

# Haemoglobinopathies on the Move: Is Europe ready?

Health and Migration  
Policy Perspectives

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This report was developed by a group of experts from the European Network for Rare and Congenital Anaemias (ENERCA) and the Thalassemia International Federation (TIF) in collaboration with the International Organization for Migration (IOM), Migration Health Division, Regional Office Brussels. Novartis Farma S.p.A. sponsored this project through in-kind support services from Burson-Marsteller Brussels.

NOTE: In this report, the term migrant is used in a very broad sense, referring not only to those who change their country of residence voluntarily but also to asylum seekers, refugees and victims of human trafficking.

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## Foreword

**Haemoglobinopathies (thalassaemia and sickle cell disease) are genetic disorders that in their severe forms are associated with chronic, life-impairing and -threatening diseases with inherent serious health sequelae that can lead to disability or even death. The World Health Organization (WHO) has recognised that haemoglobinopathies represent a growing health problem in 71% of 229 countries (which account for 89% of all births worldwide).**

Haemoglobin disorders are traditionally endemic among populations originating from Southern Europe (Italy, Greece and Cyprus), Africa, the Middle East and Asia. Through both older and more recent migration and mobility flows, these disorders are today occurring widely across the world, including the American continent, Australia, Western Europe and, in more recent years, in Northern Europe.

As a result of significant scientific and medical advances, especially in the last three decades, haemoglobin disorders can be effectively prevented, treated and managed. Increased survival, life expectancy and improved quality of life of patients in countries where programmes have been developed, implemented and nationally coordinated are well documented in the literature. Despite this, affected babies are still being born every year and patients are inadequately managed – the majority of which in the developing world, but also throughout Europe, mainly due to increased global and intra-EU mobility and migration. Accordingly, figures on the prevalence of haemoglobin disorders are largely considered underestimated due to the lack of national registries

and poor patient access to diagnosis and treatment.

Although we recognise that notable European Union initiatives do exist to reduce the psychological and economic impact of rare diseases on patients and society at large, we believe that a tailored, comprehensive, holistic approach to haemoglobinopathies is not equally applied across the EU member states. Furthermore, a clear effort to ensure equal access to healthcare by migrant populations and ethnic minority groups is lacking.

In Europe, inside and outside mobility flows and migration are broadly related to demographic and socio-economic realities, and for this reason, their effects on healthcare, welfare and social systems costs should not be underestimated. We should be investing more in responsive and evidence-based policy making, research, preventive care, healthcare professionals' education, diagnostics and treatment.

In 2006 and 2007, the WHO, through its Executive Board Meeting and World Health Assembly, adopted two Resolutions on thalassaemia<sup>1</sup> and sickle cell disease,<sup>2</sup> which represented an important step forward as political recognition of the global challenge of haemoglobinopathies. Moreover, in 2007, the European Network for Rare and Congenital Anaemias (ENERCA), with the participation of the WHO and the Thalassaemia International Federation (TIF), organised a symposium on *Immigration and Haematology: Towards the prevention of haemoglobinopathies in Europe* (Budapest, 30 August, 2007), where a joint action was proposed to call on public authorities and governments to follow up on the WHO recommendations and draw attention to the need for targeted policies to tackle severe haemoglobinopathies, and immigration and mobility flows in Europe.

Haemoglobin disorders are included in the field of rare diseases and recommendations for action in this field

have been proposed to EU countries since 2009. However, little progress has been made with regards to the development and establishment of national programmes for the prevention of this specific group of rare diseases, highly influenced by global mobility flows and immigration, reflecting the increased diversity of our societies. Piel et al. (2013) state "HbS [sickle haemoglobin] will have an increasing effect on public health systems. Our estimates can help countries and the international communities gauge the need for appropriate diagnoses and genetic counselling to reduce the number of neonates affected".<sup>3</sup> Accordingly, the need to act at the national level with the support of the EU is increasingly urgent, and is supported by recent research.

As experts, we deemed it important to develop this policy report, *'Haemoglobinopathies on the Move: Is Europe ready?'*, which, for the first time, places a spotlight on the different policies and practices of ten EU countries. The report builds on previously described initiatives by providing updated evidence for decision makers on the need to act. It also highlights the value of cost-effective policy measures, and maps areas and countries where progress is needed. We are confident that it will be of value to European policy makers in shaping the future health, migrant and social policies of the European Union and its member states.

We sincerely hope that our findings and recommendations will encourage policy makers across Europe to address the current gaps in the management of haemoglobin disorders. We hope to engage in a deeper debate on health and migration policy measures that are able to adequately respond to the rise of haemoglobinopathies in Europe and ensure equitable access to healthcare and social support, thereby contributing to improved quality and long-term cost-effectiveness of healthcare delivery.

<sup>1</sup> WHO Executive Board, 118th Session EB118.R1, Thalassaemia and other haemoglobinopathies. [http://apps.who.int/gb/ebwha/pdf\\_files/EBSS-EB118-2006-REC1/english/Res/listing/b118\\_r1-en.pdf](http://apps.who.int/gb/ebwha/pdf_files/EBSS-EB118-2006-REC1/english/Res/listing/b118_r1-en.pdf) | <sup>2</sup> WHO Fifty-Nine World Health Assembly, WHA59.20 on Sickle-cell anaemia, 2007. [http://apps.who.int/iris/bitstream/10665/21447/1/A59\\_R20-en.pdf](http://apps.who.int/iris/bitstream/10665/21447/1/A59_R20-en.pdf) | <sup>3</sup> Piel FB et al. Global epidemiology of sickle haemoglobin in neonates: a contemporary geostatistical model-based map and population estimates. *Lancet* 2013; 381:142–51

## Collaborating Experts and Organisations

## Collaborating Experts



Michael Angastiniotis

Androulla Eleftheriou

Béatrice Gulbis

### Michael Angastiniotis, MD

THALASSAEMIA INTERNATIONAL FEDERATION

Born in Famagusta, Dr Michael Angastiniotis graduated with a degree in Medicine from the University of Aberdeen (Scotland) in 1966, and obtained his DCH (Diploma in Child Health) in Paediatrics in 1976 from the Royal College of Physicians and Surgeons of Glasgow. He received his graduate training in Paediatrics in Scottish hospitals and Oxford and he returned to the UK for various courses, such as Thalassaemia (Biochemistry-Prenatal diagnosis), Genetics and Haematology/Oncology.

Dr Angastiniotis has been a member of the Thalassaemia Control Programme from the outset (1971–1972) and is Vice President of the Cyprus Society for Spastic Children. He was the Chairman of the Medical Records Committee and of the Committee for the Computerisation of the Health Service. Dr Angastiniotis has been a Special Advisor and Consultant for the control of haemoglobinopathies in the Eastern Mediterranean region. He is member of the Board of Directors of the Cyprus Institute of Neurology and Genetics, and of the WHO Expert Advisory Panel on Human Genetics. He has published a great number of scientific articles, several of which are on the topic of haemoglobinopathies and thalassaemia in particular.

Until retirement, Dr Angastiniotis was Director of the Paediatric Department of Archbishop Makarios III Hospital and the Thalassaemia Centre. Since 2004, he has been employed by TIF as a Medical Advisor.

### Androulla Eleftheriou, BSC, MSC, PhD

THALASSAEMIA INTERNATIONAL FEDERATION

Dr Androulla Eleftheriou obtained her graduate and postgraduate degrees in Biochemistry, Microbiology and Virology. She has been awarded a number of scholarships by the Cypriot government, the World Health Organization and the Fulbright Commission. Her postdoctoral fellowship was completed at the Centre for Disease Control in Atlanta, GA, USA. Dr Eleftheriou has also recently obtained a Diploma in Business Management from the University of Leicester, UK.

From 1990 until August 2006, she was Head of the Virus Reference Centre of the Cyprus Ministry of Health – a centre she was closely involved in establishing. Since 2005 she has held the post of Director at the Cyprus WHO Collaborating Centre. She has worked with TIF since 1993, initially on a voluntary basis as a Scientific Coordinator of TIF's educational programme, and since 2006 as the Executive Director of the Federation. She regularly acts as a WHO consultant on issues related to her field of expertise.

Through her work with TIF, Dr Eleftheriou has carried out numerous projects of local, national, regional and international scope, working closely with international experts, local healthcare professionals and thalassaemia associations worldwide. She is involved in many joint activities and projects, for example, the ENERCA, DOMAINE and Ithanel EU co-funded projects, and closely collaborates with other disease-specific organisations and health-orientated bodies at the European and international level as well as with official health and pharmaceutical authorities. Dr Eleftheriou is a member of numerous committees and the Pan-European Blood Safety Alliance, where she has been Secretary since 2005. She is also an appointed member of the European Union Committee of Experts in Rare Diseases (EUCERD), and a past member of the Executive Committee of the European Public Health Alliance (EPHA).

Dr Eleftheriou is the author of over 11 TIF publications, as well as several publications written in collaboration with WHO and other international bodies on a wide range of scientific topics. Dr Eleftheriou is the Chief Editor of *TIF Magazine*, which is issued quarterly and distributed to 2,500 subscribers in more than 40 countries worldwide.

### Béatrice Gulbis, MD, PhD

ERASME HOSPITAL – UNIVERSITÉ LIBRE DE BRUXELLES (BELGIUM)

Dr Béatrice Gulbis graduated in Medicine in 1984, and obtained her specialisation in Clinical Biology in 1989 from the Université Libre de Bruxelles (Belgium). Dr Gulbis is head of the Department of Clinical Chemistry and since 2010, Director of the Departments of Clinical Biology and Molecular Genetics. She is also Professor of Haematology at the Faculty of Medicine and is a genetic counsellor in the field of hereditary red blood cell disorders.

More than 15 years ago, Dr Béatrice Gulbis led the development of a red blood cell reference laboratory centre at the Erasme Hospital in Belgium, to build expertise in the diagnosis of hereditary red blood cell disorders, i.e. haemoglobinopathies, erythrocyte membrane and enzyme defects, and also neonatal and prenatal screening for haemoglobinopathies. In collaboration with the haematology department and the genetic centre of the Erasme Hospital, Dr Gulbis has led the implementation of specific phenotypic and genotypic tools with a view to offering a large panel of tests for the diagnosis of hereditary red blood cell disorders, as well as genetic counselling consultations on these diseases.

The centre's expertise has been broadly recognised at the national level (Presidency of Belgian Haematological Society subcommittee dedicated to disorders of red blood cells - <http://www.bhs.be>) and international level (leadership of a European working group devoted to sickle cell syndromes - European Network for Rare Congenital Anaemias; <http://www.enerca.org>).



Maria del Mar Mañú Pereira Patricia Aguilar Martinez Roumyana Petrova-Benedict Joan-Lluis Vives-Corrns

## Maria del Mar Mañú Pereira, BSc, PhD

HOSPITAL CLÍNIC DE BARCELONA (SPAIN)

Dr Maria del Mar Mañú Pereira graduated with a degree in Biology from the University of Barcelona (Spain) in 2002 and specialised in Human Genetics. She obtained her PhD in 2009, with a thesis on “Haemoglobinopathies and Glucose-6-phosphate dehydrogenase in Catalonia: Epidemiological and molecular genetics studies in the newborn population.”

Since 2002, she has been working as a molecular biologist at the red cell laboratory at the Hospital Clínic de Barcelona. Dr del Mar Mañú Pereira performs diagnostic tests for red cell disorders, and is leading the development of methodologies for genetic characterisation at the laboratory. Further, she also corroborates with the University of Barcelona as a professor.

Her research activity is devoted to haemoglobin and enzyme disorders; specifically, molecular characterisation and genotype-phenotype correlation. She has been involved in several projects at a national and international level. She is member of the coordination team of ENERCA (European Network for Rare and Congenital Anaemias) since 2007.

## Patricia Aguilar Martinez, MD, PhD, CHU

MONTPELLIER - SAINT ELOI HOSPITAL (FRANCE)

Dr Patricia Aguilar Martinez graduated with a degree in medicine from the Montpellier Medical School (France) in 1988 and specialised in Biology and Genetics in different French Universities, including Lyon and Paris. She obtained her PhD in 1994 (thesis on the “molecular biology of haemophilia and haemoglobinopathies”). Since 1989, she has been a consultant in genetic counselling for hereditary haematological disorders, especially haemoglobinopathies and hereditary iron overload. Since 1993, she has also been responsible for the departments of red cell disorders and molecular biology at the laboratory of haematology in the Montpellier teaching hospital. She is currently a Professor of Haematology at the Faculty of Medicine in Montpellier.

She is the coordinator of the “Centre de compétence” (clinical expert centre) on rare iron disorders and a founding member of the department of red cell disorders at the Montpellier teaching hospital. She has been the treasurer of the French Red Cell Scientific Society for 10 years.

Since 2001, she has been collaborating with the WHO Human Genetic programme, and in 2006 she was invited to join the WHO Expert Advisory Panel on Human Genetics. In 2010–2011 she was appointed for a temporary 6-month mission at WHO headquarters in Geneva as the officer in charge of the Human Genetics programme. Her mission focused on the follow up of the implementation of the 2006 WHO resolutions on thalassaemia and sickle cell disease.

Her research activity is devoted to genetic iron disorders, particularly genetic modifiers in hereditary iron overload. She has been a member of the ENERCA project since its creation, and a work package leader during the second and third phases of this project.

## Roumyana Petrova-Benedict

INTERNATIONAL ORGANIZATION FOR MIGRATION (BELGIUM)

Dr Roumyana Petrova-Benedict, IOM Senior Regional Migration Health Manager for Europe and Central Asia, is responsible for the development, implementation and coordination of IOM Migration Health programmes, liaison and partnerships.

She has over 19 years’ experience in global public health, development and research from within academic, civil society and intergovernmental institutions across several continents (Main topics: Migration Health; Primary Health Care; Human Resources for Health; Community Development and Care; Health Information Systems; TB; STIs, HIV and AIDS).

## Joan-Lluis Vives-Corrns, MD, PhD

HOSPITAL CLINIC - UNIVERSITAT DE BARCELONA (SPAIN)

Dr Vives had been Professor of Medicine and Head of the Haematology Laboratory Department of the Hospital Clínic i Provincial (University of Barcelona) since 1976, with a high input into haematology research, education and diagnosis. His main activities have been focused on the development of new procedures for the diagnosis of anaemias and haematological malignancies (acute and chronic leukaemias and lymphomas), as well as the standardisation and quality assessment of haematology laboratory practice.

His research has been mainly related to the physiopathology, molecular mechanisms and epidemiology of red blood cell disorders due to hereditary enzymopathies (G6PD, PK and others), haemoglobinopathies (sickle cell anaemia and thalassaemias) and membrane defects (hereditary spherocytosis and stomatocytosis). Dr Vives’ research has led to the creation of the Red Cell Pathology Unit, a recognised Expert Centre for the diagnosis of red blood cell defects and in the identification of low prevalence anaemias, also known as ‘rare anaemias’.

Dr Vives’ scientific career includes over 400 publications in scientific journals, participation (as an editor or collaborator) in about 20 books on haematology and the accomplishment of 28 research projects, which have been partially funded by different public health organisations, including the Spanish (Ministry of Health and Education and Science) and Catalan governments. Since 2002, Dr Vives is the leader of the public health project of the European Commission for the development of a European Reference Network (ERN) of Expert Centres in Rare Anaemias.

## Collaborating Experts and Organisations

### ENERCA | website: [www.enerca.org](http://www.enerca.org) (European Network for Rare and Congenital Anaemias)

ENERCA is co-financed by the European Commission (DG-SANCO) with the goal of preventing rare anaemias and promoting policies that lead to a healthier way of life for EU citizens. Two pivotal aspects, a specific framework for cross-border healthcare and a European cooperation on health services, are promoted.

As for many other rare diseases, a large-scale network of experts and specialists was previously non-existent in the field of rare anaemias, leading to a serious lack of information for both patients and healthcare professionals. The ENERCA Project started in 2002, with the purpose of offering an improved public health service in every aspect of rare anaemias. Common goals with the 2006 WHO Resolution include the public health role of ENERCA, its education and training programmes, the dissemination of appropriate technologies, guidelines, expert groups and the promotion of research. The yearly *European Symposium on Rare Anaemias*, organised by ENERCA in collaboration with patient associations, is greatly appreciated.

In 2013, an ENERCA Task Force prepared the recommendations for the identification of Centres of Expertise (CEs) as the nodes of the future European Reference Network (ERN) for Rare Anaemias. These recommendations have been published in a 'White Book' (in printing stage) that aims to provide stakeholders involved in national plans for rare diseases and the European Commission with the practical material and specific methodology needed for moving towards the establishment of consensus recommendations for their national CEs in rare anaemias.

Finally, in 2013–2016, ENERCA will initiate a new project to establish a telemedicine platform with three main branches: e-registry, e-learning and telediagnosis. The platform will enhance the web-based interactivity of ENERCA with patients, professionals and other stakeholders, and allow for easy access to information, healthcare, teaching and epidemiological data. Moreover, an immediate reply will be possible for the diagnosis of anaemias of unknown origin and the care of patients with chronic anaemia due to sickle cell disease and thalassaemia.

## Collaborating Organisations

### TIF | website: [www.thalassaemia.org.cy](http://www.thalassaemia.org.cy) (Thalassaemia International Federation)

TIF was established in 1986 by thalassaemia patients and parents, mainly from Mediterranean countries, the UK and USA. TIF's mission is to obtain equal access to quality healthcare for every patient with thalassaemia, through the establishment of effective control programmes across the world.

To achieve this goal, TIF focuses on:

- (i) Supporting the establishment and promotion of activities of national patients' and parents' organisations;
- (ii) Establishing networks, partnerships and collaborations with national and international health-related organisations and the international medical community;
- (iii) Supporting national health authorities in their efforts to develop effective national control strategies that include the components of prevention and clinical management.

One of the main challenges for TIF is to 'bridge the gap' that today exists between industrialised and developing countries – to extend the knowledge, experience and expertise gained in those countries that first recognised thalassaemia and developed effective control programmes, to other affected countries across the world. Since 1996, TIF has been an official interlocutor of the WHO and is considered the international reference organisation for thalassaemia. TIF's longstanding collaboration with the WHO has in recent years (2006) resulted in the adoption of resolutions on sickle cell disease and thalassaemia. A joint 5-year plan of action has been developed to put these resolutions into practice across the world.

A major focus of TIF's activities is its educational programme, aimed at spreading information and knowledge on these diseases, including prevention, management and cure, to the patient and medical community, as well as the general population. To this end, the Federation organises regular conferences, workshops and seminars, and publishes an extensive range of educational materials ([www.thalassaemia.org.cy](http://www.thalassaemia.org.cy)). Whilst TIF will continue its work on a global basis, it recognises the specific needs and challenges in the European region, which remain under-recognised and underestimated.

In response to the need to revisit and strengthen existing



policies in Europe, TIF has more recently increased its activities within the region.

TIF has developed a strong network of collaboration with European patients' associations (disease and non-disease specific), the medical community, healthcare providers and official bodies at the level of the European Union (European Commission, European Parliament, Council of Europe, etc). TIF actively participates in the work of organisations such as the European Haematology Association (EHA), European School of Transfusion Medicine (ESTM), European Public Health Alliance (EPHA), the European Organisation for Rare Diseases (Eurordis), the European Platform for Patients' Organisations, Science and Industry (EPPOSI) and the European Medicines Agency (EMA). TIF has also participated in a number of EU-funded projects, including ENERCA, Ithamet and DOMAINE, aiming to improve the services for patients with haemoglobin disorders in Europe.

### **IOM | website: [www.iom.int](http://www.iom.int)**

#### **(International Organization for Migration)**

IOM is a dynamic and growing inter-governmental organisation committed to the principle that humane and orderly migration benefits both migrants and society. As the leading global organization for migration, IOM works with migrants, governments and its partners to: assist in meeting the operational challenges of migration; advance understanding of migration issues; encourage social and economic development through migration; and uphold the human dignity and well-being of migrants ([www.iom.int](http://www.iom.int)).

The IOM Constitution recognises the link between migration and economic, social and cultural development, as well as to the right of freedom of movement.

IOM works in the four broad areas of migration management:

- Migration and development
- Facilitating migration
- Regulating migration
- Forced migration

IOM activities that cut across these areas include the

promotion of international migration law, policy debate and guidance, protection of migrants' rights, migration health and the gender dimension of migration.

The Migration Health Division (MHD) vision is that: migrants and mobile populations benefit from an improved standard of physical, mental and social wellbeing, which enables them to substantially contribute towards the social and economic development of their home communities and host societies. In Europe, IOM Regional Office (RO) Brussels works in partnership with EU institutions (DG Health and Consumers, DG Research and EU Agencies), regional networks (NDPHS, SEEHN), other international organisations, academic networks and civil society organisations in support of policy development, providing technical advice and assistance in the implementation of projects. MHD RO Brussels also collaborates closely with member states and, in particular, has supported the health agendas of the Portuguese (2007) and Spanish (2010) EU Presidencies in their priorities on migrant health and health inequalities, respectively. IOM has supported the drafting of the Bratislava Declaration on Human Rights, Health and Migration (November 2007) of the Council of Europe (CoE). Furthermore, IOM participated in the work of the CoE's Expert Committee on Mobility, Migration and Access to Health Care, whose recommendations, CM/Rec (2011), were adopted in November 2011 by the Committee of Experts of the Council of Europe.

In December 2012, IOM was awarded a direct grant co-financed by the EC/DG Health and Consumers for the EQUI-HEALTH project within the framework of the 2012 EC Public Health Programme. The action started in February 2013 and aims to improve access to quality healthcare services for migrants, Roma and other vulnerable ethnic minority groups, including irregular migrants, in the EU, the European Economic Area (EEA), Croatia and potentially Turkey.

#### **Disclaimer**

The opinions expressed in this report are those of the author(s) and do not necessarily reflect the views of the International Organization for Migration. The designations employed and the presentation of material throughout the paper do not imply the expression of any opinion whatsoever on the part of the IOM concerning the legal status of any country, territory, city or area, or of its authorities, or concerning its frontiers or boundaries.

## Key Facts and Figures about Haemoglobinopathies

**Haemoglobinopathies (thalassaemias and sickle cell disorders) are rare blood disorders affecting 330,000 infants born annually around the world.<sup>4</sup> 83% of these infants are affected by sickle cell disease and 17% by thalassaemia. According to the WHO, haemoglobinopathies present a growing health problem in 71% of 229 countries (countries that account for 89% of all births worldwide).**

Thalassaemia is defined as a partial or total decrease in the production of haemoglobin; a protein found in red blood cells, whose main function is the transport of oxygen to the tissues. This decrease is caused by mutations affecting the genes responsible for the production of the globin chains that form the haemoglobin protein. Decreased production of haemoglobin leads to anaemia.

Thalassaemia can be classified depending on the gene involved - Alpha ( $\alpha$ ) or Beta ( $\beta$ ). The level of severity (ranging from mild to major), is linked to the number of copies of the gene affected by a thalassaemia mutation

and sometimes to other co-inherited anomalies. Whereas thalassaemia affects the quantity of haemoglobin produced, sickle cell disease affects the quality. The abnormal structure of the haemoglobin molecule changes the red blood cells' shape. Their 'sickle' shape does not allow them to travel through thin blood vessels, causing blockages that deprive organs and tissues of oxygen. Because red blood cells have a very short life-span and cannot be replaced quickly, sickle cell disease patients are anaemic.

Both thalassaemia and sickle cell diseases can lead to chronic complications that severely undermine the quality of life of patients and can, in some cases, lead to death (see table 4).

There is substantial scientific evidence that haemoglobinopathies have become a global public health problem. Thalassaemia is most common among those of Italian, Greek, Middle Eastern, Asian and African descent.<sup>5</sup> However, migration has seen an increase in these diseases outside of the Mediterranean, Africa and Asia, and they are now endemic throughout Europe, the Americas and Australia.

**Table 1 | HAEMOGLOBIN DISORDERS IN NUMBERS (2007)<sup>6</sup>**

Around 7% of the global population CARRIES AN ABNORMAL HAEMOGLOBIN GENE;

300,000–500,000 children ARE BORN WITH CLINICALLY SIGNIFICANT HAEMOGLOBIN DISORDERS ANNUALLY;

About 80% of affected children ARE BORN IN DEVELOPING COUNTRIES;

About 70% ARE BORN WITH SICKLE CELL DISEASE (SCD) AND THE REST WITH THALASSAEMIA SYNDROMES;

50–80% of children with SCD DIE EACH YEAR IN LOW AND MIDDLE INCOME COUNTRIES;

50,000–100,000 children with THALASSAEMIA MAJOR DIE EACH YEAR IN LOW AND MIDDLE INCOME COUNTRIES.

<sup>4</sup> Darlison M, Modell B. Global epidemiology of haemoglobin disorders and derived service indicators. Bull World Health Organ 2008;86(6):480–487. | <sup>5</sup> US National Heart Lung and Blood Institute, What are thalassaemias? <http://www.nhlbi.nih.gov/health/health-topics/topics/thalassaemia/> | <sup>6</sup> Management of haemoglobin disorders: report of joint WHO-TIF meeting, Cyprus, November 2007. [http://www.who.int/genomics/WHO-TIF\\_genetics\\_final.pdf](http://www.who.int/genomics/WHO-TIF_genetics_final.pdf)



While general understanding of the necessary components for preventing and managing haemoglobin disorders has improved, these disorders are fairly well-recognised and managed predominantly in countries where they are traditionally endemic (e.g. Cyprus, Greece, Italy) or that have a longer tradition of receiving immigrants (e.g. France and UK). Current and future migration and mobility flows to and within the EU pose new challenges, however. In a number of countries with low incidence, evidence shows that migrant population groups (e.g. North Africa, Southern Asia and Southeast Asia but also from Southern EU countries) are under-diagnosed, and healthcare delivery systems are not sufficiently responsive to their care needs. High birth rates among these communities<sup>7</sup> (often with a consanguineous partner<sup>8</sup>) have contributed to the increased prevalence of haemoglobinopathies in traditionally non-endemic countries.<sup>9</sup>

Clinical history and prevalence of hereditary haemoglobin disorders varies considerably between different regions; countries; areas and even between different medical centres within the same area. Despite laboratory and clinical advancements, increased numbers of annual affected births and high rates of mortality and morbidity are still observed in the majority of affected countries in the developing world<sup>10</sup> and increasingly throughout the EU.

Advances in treatment have contributed to a significant improvement in patients' quality of life.<sup>11</sup> Thalassaemia major patients are treated with blood transfusions.

Three types of intensification of treatment – hydroxyurea, chronic transfusion and bone marrow/stem cell transplant – can be proposed for sickle cell disease patients.<sup>12</sup>

Iron overload is a common and potentially severe complication for patients in chronic transfusion programmes<sup>13</sup> and, therefore, patients must also receive iron chelation therapy (chelators bind to iron molecules, preventing an excess of iron in the body, which can cause a variety of conditions and death). Iron overload may also occur in thalassaemia intermedia and some severely affected sickle cell disease patients.

Treatments of sickle cell patients aim to prevent acute events, treat severe events, and prevent the onset of chronic organ damage.

Although genetically inherited, haemoglobin disorders can be diagnosed, treated and managed, thereby increasing patients' life expectancy.

In developed countries, life expectancy of patients with severe form of thalassaemia is estimated at 40–55 years, if patients comply with and have free of charge access to appropriate medical and other care.

In sickle cell disease, 50% of patients live to 40–60 years old, depending on the type of disorder, whereas only 10% of patients live longer than 60 years, depending on the sickle cell disease genotype variation.

<sup>7</sup> Modell B et al. Epidemiology of haemoglobin disorders in Europe: an overview. *Scand J Clin Lab Invest* 2007;67(1):39–69. | <sup>8</sup> Amato A, Giordano PC. Screening and genetic diagnosis of hemoglobinopathies in southern and northern europe: two examples. *Mediterr J Hematol Infect Dis* 2009;1(1):e2009007. | <sup>9</sup> Global epidemiology of haemoglobin disorders and derived service indicators, op cit. | <sup>10</sup> World Health Assembly Resolution on Sickle Cell Anaemia. WHA59.20. WHO, Geneva, Switzerland, 2006. Executive Board Resolution on Thalassaemia and Other Haemoglobinopathies. EB118.R1. | <sup>11</sup> US National Heart Lung and Blood Institute, What is sickle cell anemia? <http://www.nhlbi.nih.gov/health/health-topics/topics/sca/> | <sup>12</sup> Montalembert M et al. ENERCA clinical recommendations for disease management and prevention of complications of sickle cell disease in children. *Am J Hematol* 2011;86(1):72–75. | <sup>13</sup> Galactéros F, de Montalembert M. Sickle cell disease: a short guide to management, Chapter 13. In: European School of Haematology (ESH) Handbook on Disorders of Erythropoiesis, Erythrocytes and Iron Metabolism. European School of Haematology; 2009. [http://www.ironcurriculum.esh.org/Activity/1819\\_03/1819\\_03\\_chapter13.pdf](http://www.ironcurriculum.esh.org/Activity/1819_03/1819_03_chapter13.pdf)

## Key Facts and Figures about Haemoglobinopathies

**Table 2 | ESTIMATIONS OF THE NUMBER OF CARRIERS IN THE COUNTRIES STUDIED<sup>14</sup>**

COUNTRY	TOTAL POPULATION	TOTAL NUMBER OF IMMIGRANT CARRIERS OF $\beta$ THAL	TOTAL NUMBER OF CARRIERS OF $\beta$ -THALASSAEMIA IN THE INDIGENOUS POPULATION	TOTAL NUMBER OF IMMIGRANT CARRIERS OF HbE	TOTAL NUMBER OF IMMIGRANT CARRIERS OF SICKLE CELL	TOTAL NUMBER OF IMMIGRANT CARRIERS OF HbC	CARRIER IMMIGRANTS AS A PROPORTION OF THE TOTAL POPULATION	CARRIERS OF Hb DISORDERS AS A PROPORTION OF THE TOTAL POPULATION
AUSTRIA	8,210,281	11,842	8,210	2,453	4,675	708	0.24%	0.34%
BELGIUM	10,438,353	19,403	10,438	4,073	39,250	5,169	0.65%	0.75%
CYPRUS	840,407	3,991	121,019	354	583	20	0.58%	15.00%
DENMARK	5,543,453	6,772	5,543	4,083	2,277	330	0.24%	0.34%
FRANCE	64,057,792	98,219	64,058	32,607	172,600	47,884	0.54%	0.65%
GERMANY	82,329,758	128,419	82,330	22,955	53,883	7,135	0.25%	0.36%
GREECE	10,737,429	29,289	837,519	536	7,626	183	0.35%	8.70%
ITALY	61,261,254	75,748	2,572,972	9,463	72,870	21,416	0.29%	6.50%
THE NETHERLANDS	16,715,999	27,656	16,716	13,751	30,329	7,703	0.47%	0.57%
SPAIN	47,042,984	57,257	715,053	2,434	92,601	27,796	0.38%	1.90%
SWEDEN	9,482,855	21,092	9,483	12,593	8,720	912	0.46%	0.56%
UK	63,047,162	107,694	63,047	27,124	145,038	25,290	0.48%	0.58%

These results are the nearest figures that are calculated on the available data on immigrant populations. It was assumed that Northern European populations have a thalassaemia carrier rate of 0.1% in their indigenous populations and no carriers of the sickle cell gene. The importance is that in the countries where the prevalence is high in the indigenous population (Italy, Greece and Cyprus), there are national policies to meet the needs of these disorders. In the rest of Europe the proportion of immigrants is approximately similar, yet only the UK and France have disease specific policies. The carrier frequency is rising most rapidly in Belgium and Spain where national planning is most urgently needed.

<sup>14</sup> Angastiniotis M, Vives Corrons J-L, Soteriades ES, Eleftheriou A. The Impact of Migrations on the Health Services for Rare Diseases in Europe: The example of Haemoglobin Disorders, in: The Scientific World Journal Volume 2013

Table 3 | DISEASE BURDEN IN ASSOCIATION WITH THE EXPECTED BIRTH RATE AND THE NUMBER OF KNOWN PATIENTS<sup>15</sup>

COUNTRY	EXPECTED THALASSAEMIA BIRTHS/ 1,000 LIVE BIRTHS	EXPECTED SICKLE CELL DISEASE BIRTHS/1,000 LIVE BIRTHS	NUMBER OF KNOWN OR ESTIMATED PATIENTS WITH THALASSAEMIA SYNDROMES	NUMBER OF KNOWN OR ESTIMATED PATIENTS WITH SICKLE CELL SYNDROMES
AUSTRIA	0.0015	0.00008	NA	NA
BELGIUM	0.002	0.0035	100	400
CYPRUS	5.5	0.0018	639	40
DENMARK	0.0018	0.00004	NA	NA
FRANCE	0.0016	0.0018	571	10,000
GERMANY	0.0016	0.0001	1,500	3,000
GREECE	1.6	0.009	3,241	1,080
ITALY	0.46	0.11	7,000	6,000
THE NETHERLANDS	0.0018	0.0008	250	750
SPAIN	0.067	0.0011	113	>200
SWEDEN	0.0025	0.00021	50	100
UK	0.0018	0.0013	920	15,000

NA = not available

Table 4 | CO-MORBIDITIES AND COMPLICATIONS CAUSED BY HAEMOGLOBIN DISORDERS<sup>16 17 18</sup>

Thalassaemia	Sickle Cell Disease
Chronic anaemia.	Chronic anaemia.
Iron overload (from the disease or blood transfusion), and resultant complications in the cardiac, hepatic and endocrine systems.	Acute Anaemia.
Increased risk of infections (mainly transfusion related, e.g. hepatitis B and C).	Adverse events related to vaso-occlusion and severe damage of the brain, heart, lungs, kidney, liver, bones and spleen. Vasculopathy leading to pulmonary hypertension, priapism, leg ulcers and cerebrovascular disease.
Bone abnormalities and osteoporosis.	Increased risk of infections related to hyposplenism.

<sup>15</sup> Ibidem. <sup>16</sup> Wood B, Higgs B. Molecular basis of thalassaemia syndromes, Chapter 10. In: European School of Haematology (ESH) Handbook on Disorders of Erythropoiesis, Erythrocytes and Iron Metabolism. European School of Haematology: 2009. [http://www.esh.org/files/doc/IRON2009\\_CAP.10\(250-263\).pdf](http://www.esh.org/files/doc/IRON2009_CAP.10(250-263).pdf) <sup>17</sup> Galanello R, Origa R. Management of thalassaemia, Chapter 11. In: European School of Haematology (ESH) Handbook on Disorders of Erythropoiesis, Erythrocytes and Iron Metabolism. European School of Haematology: 2009. [http://www.esh.org/files/doc/IRON2009\\_CAP.11\(264-285\).pdf](http://www.esh.org/files/doc/IRON2009_CAP.11(264-285).pdf) <sup>18</sup> Sickle cell disease: a short guide to management (Chapter 13), F. Galactéros, M. de Montalembert, European School of Haematology (ESH) Handbook on Disorders of Erythropoiesis, Erythrocytes and Iron Metabolism, December 2009, [http://www.ironcurriculum.esh.org/Activity/1819\\_03/1819\\_03\\_chapter13.pdf](http://www.ironcurriculum.esh.org/Activity/1819_03/1819_03_chapter13.pdf)

## Methodology

This report was developed on behalf of a group of experts in the field of haemoglobinopathies working for ENERCA and the Thalassemia International Federation (TIF) (hereafter, Collaborating Experts; see previous section) between March 2012 and May 2013. Contents are based on targeted desk research and one-to-one interviews with national policy makers and relevant stakeholders, including healthcare professionals, experts, patient organisations and government officials. The scope, methodology, research and interview questionnaires were agreed with the Collaborating Experts and TIF. Burson-Marsteller, a public affairs and communication agency, was responsible for carrying out the research and interviews. Novartis Farma S.p.A sponsored this project through in-kind support services from Burson-Marsteller Brussels.

The report provides a description, to the best of our knowledge, of the current policy environment on the basis of the best available factual information and relevant stakeholder opinions. It is not intended as a scientific or statistical report.

### DESK RESEARCH

Relevant information was gathered from each of the 10 EU member states with regard to the prevalence of haemoglobinopathies (particularly thalassaemia and sickle cell disease), the cost of these disorders to healthcare budgets, as well as the relevant policies, guidelines and practices in place at the national and, in some cases, regional level. The information comes from a range of sources, including government and patient association websites, published scientific literature and media reports. Regarding the prevalence and cost data, it should be noted that, at the time of publication, there were few comprehensive, comparative European data sources available.

### INTERVIEWS

Building on the desk research, one-to-one interviews were carried out in each of the 10 countries with three or four national stakeholders representing health ministries, haematologists and other healthcare professionals, patients and families. The interviews were conducted according to a pre-defined questionnaire. The interviews aimed to further assess: the burden and cost of haemoglobinopathies; the existence of specialised measures for prevention, diagnosis and care, including support to families at the national level; and the extent of their implementation. The information provided during interviews has not been attributed to individuals. In some cases, interviewees agreed to provide approved statements, which have been included in the report. A list of consulted organisations is included on page 72 of this report. While attempts were made to consult a range of stakeholder groups in the preparation of each country report, in a minority of cases, responses from all groups, including government officials, were not available within the given time period. In such situations, reports were written on the basis of the best information available.

### EXPERT REVIEW

The Collaborating Experts, TIF and IOM carried out a comprehensive review of the key findings, conclusions and policy recommendations. Preliminary results were also discussed with relevant stakeholders during a dedicated panel session in the context of the 3rd Pan-European Conference on Haemoglobinopathies and Rare Anaemias, organised by the TIF in Cyprus, on 26 October 2012, and the ENERCA Symposium.

### FEEDBACK

The partners involved welcome feedback on this report. They recognise that there may have been new developments in some countries since this research was completed, and that further explanation or clarification may be needed for some country factsheets.

Please send any comments to the following email address: [tif@thalassaemia.org.cy](mailto:tif@thalassaemia.org.cy) and [enerca@enerca.org](mailto:enerca@enerca.org)

## EU and Country Factsheets

- EU
  - Belgium
  - Cyprus
  - France
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  - The Netherlands
  - Spain
  - Sweden
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-

## EU Factsheet

### KEY FACTS AND FIGURES



Haemoglobinopathies are classified as 'rare diseases' in Europe.<sup>19</sup>

#### PREVALENCE DATA FROM EU RARE DISEASE WEB PORTAL ORPHANET<sup>20</sup>:

Sickle cell disease: 15/100,000 PEOPLE

$\beta$ -thalassaemia: 0.5/100,000 PEOPLE

### RARE DISEASES

The European Union has made remarkable progress in the area of rare diseases, having adopted a number of policy initiatives that aim to:

- Support knowledge and access to information on rare diseases<sup>21</sup>
- Incentivise research and development of medicinal products intended for the treatment of rare diseases<sup>22</sup>
- Encourage prevention, exchange of information and management of rare diseases<sup>23</sup>
- Provide a comprehensive strategy framework for EU action to improve prevention and care of rare diseases, through increased cooperation across the EU and support for targeted actions at a national level<sup>24 25</sup>

The 2009 Council Recommendation on an action in the field of rare diseases,<sup>25</sup> although not legally binding, denotes political commitment from the 27 member states to address rare diseases. As a consequence, national action plans on rare diseases are being drafted across the EU.

The 2009 Council Recommendation also called on the EU member states to develop comprehensive EU-wide guidelines for rare disease screening and diagnosis. The European Commission has promoted efforts to compare and evaluate practices of neonatal screening for rare disorders at a national level.<sup>26</sup>

As part of this work, the European Network of Experts on Newborn Screening (EUNENBS)<sup>27</sup> was established and has recently published a report on the practices of newborn screening for rare disorders in Member States of the European Union, as well as Candidate, Potential Candidate and EFTA Countries.<sup>28</sup> The opinion provided by a group of leading experts<sup>29 30</sup> includes a set of recommendations on the provision of information to healthcare professionals and parents on neonatal screening, and the need to ensure that free screening for rare diseases is offered to all infants across the EU.

<sup>19</sup> International Statistical Classification of Diseases and Related Health Problems 10th Revision (ICD-10), World Health Organization, 2010. <http://apps.who.int/classifications/icd10/browse/2010/en#/D55> | <sup>20</sup> Prevalence of rare diseases: bibliographic data, Orphanet Report Series, number 1, May 2012. [http://www.orpha.net/orphacom/cahiers/docs/GB/Prevalence\\_of\\_rare\\_diseases\\_by\\_alphabetical\\_list.pdf](http://www.orpha.net/orphacom/cahiers/docs/GB/Prevalence_of_rare_diseases_by_alphabetical_list.pdf) | <sup>21</sup> Programme of Community Action on Rare diseases (1999-2003), European Commission, 1999. [http://ec.europa.eu/health/archive/ph\\_overview/previous\\_programme/rare\\_diseases/raredis\\_wpgm99\\_en.pdf](http://ec.europa.eu/health/archive/ph_overview/previous_programme/rare_diseases/raredis_wpgm99_en.pdf) | <sup>22</sup> European Parliament and Council Regulation No 141/2000 of 16 December 1999 on Orphan Medicinal Products. [http://europa.eu/legislation\\_summaries/internal\\_market/single\\_market\\_for\\_goods/pharmaceutical\\_and\\_cosmetic\\_products/121167\\_en.htm](http://europa.eu/legislation_summaries/internal_market/single_market_for_goods/pharmaceutical_and_cosmetic_products/121167_en.htm)



## EU Factsheet

### HEALTH INEQUALITIES

### MIGRANT HEALTH

EU policy makers have increasingly recognised the need for targeted policy action to address health inequalities across the EU.

In its ‘Solidarity in health: reducing inequalities in the EU’ communication, dated October 2010, the Commission stressed the need to address the difficulty for certain groups including “people from some migrant or ethnic minority backgrounds” in accessing and benefiting from high-quality healthcare.<sup>31</sup> In its resolution of 8 March 2011 on reducing health inequalities in the EU,<sup>32</sup> the European Parliament called on the EU member states to ensure that “information on health, healthy lifestyles, healthcare, prevention opportunities, early diagnosis of diseases and suitable treatments is available in a form and in languages that everyone can understand, using new information and communication technologies”.

Reducing health inequalities remains one of the main policy priorities under the proposed third multi-annual programme of EU action in the field of health for the period 2014–2020.<sup>33 34</sup> The ‘Health for Growth’ programme is expected to support actions aimed at enhancing patient access to cross-border healthcare through the creation of reference networks and registries for rare diseases.

Migrant and health are two interconnected policy areas, with significant implications for the wellbeing of the EU population.

The Lisbon Treaty, which came into force on 1 December 2009, gives legally binding jurisdiction to the Charter of Fundamental Rights of the European Union.<sup>35</sup>

The Lisbon Treaty has reiterated that respect for human rights is one of the values upon which the EU is founded (*Article 6 of the Treaty on the functioning of the European Union*) and included ‘wellbeing’ as a new objective of the EU (*Articles 2 and 3 of the Treaty on the functioning of the European Union*). Articles of the Treaty are available at <http://eur-lex.europa.eu/en/treaties/new-2-47.htm>.

Article 35 of the Charter states that “everyone has the right of access to preventive health care and the right to benefit from medical treatment, thus giving the EU a more predominant role in preventive care.”

This recognition of a universal right to health forms another basis for future EU debate and work on migrants’ access to health services. Member states are also bound by other international instruments that recognise health as a human right, including the Constitution of the World Health Organization of 1946 and the Universal Declaration of Human Rights in 1948.<sup>36</sup>

<sup>323</sup> Decision No. 1786/2002/EC of the European Parliament and of the Council of 23 September 2002 adopting a programme of Community action in the field of public health (2003–2008), 2002. <http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=CELEX:32002D1786:EN:NOT> <sup>24</sup> Communication from the Commission on Rare Diseases, European Commission, 11 November 2008. [http://ec.europa.eu/health/ph\\_threats/non\\_com/docs/rare\\_com\\_en.pdf](http://ec.europa.eu/health/ph_threats/non_com/docs/rare_com_en.pdf) <sup>25</sup> Council Recommendation of 8 June 2009 on an action in the field of rare diseases. Official Journal of the European Union, 2009. <http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:C:2009:151:0007:0010:EN:PDF> <sup>26</sup> Evaluation of population newborn screening practices for rare disorders in Member States of the European Union, Executive Agency for Health and Consumers, European Commission, January 2012. [http://ec.europa.eu/eahc/health/tenders\\_H09C2.html](http://ec.europa.eu/eahc/health/tenders_H09C2.html) <sup>27</sup> Newborn Screening in Europe Expert Opinion document, EU Tender “Evaluation of population newborn screening practices for rare disorders in Member States of the European Union”, January 2012. [http://ec.europa.eu/eahc/documents/news/Expert\\_opinion\\_document\\_on\\_NBS\\_20120108\\_FINAL.pdf](http://ec.europa.eu/eahc/documents/news/Expert_opinion_document_on_NBS_20120108_FINAL.pdf) <sup>28</sup> Newborn Screening in Europe Expert Opinion document. <sup>29</sup> Newborn Screening in Europe Expert Opinion document, op.cit. <sup>30</sup> Martina Cornel, Tessel Rigter, and Stephanie Weinreich from the VU University Medical Centre; Peter Burgard, Georg F. Hoffmann, Kathrin Rupp and Martin Lindner from the Department of Paediatrics of the University of Heidelberg; J. Gerard Loeber from RIVM; Domenica Taruscio and Luciano Vittozzi from the National Centre for Rare Diseases of the Italian Institute for Health <sup>31</sup> Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions – Solidarity in health: reducing health inequalities in the EU, European Commission, 20 October 2009. [http://ec.europa.eu/health/ph\\_determinants/socio\\_economics/documents/com2009\\_en.pdf](http://ec.europa.eu/health/ph_determinants/socio_economics/documents/com2009_en.pdf) <sup>32</sup> Reducing Health Inequalities: European Parliament Resolution of 8 March 2011, on reducing health inequalities in the EU, 2011. <http://www.europarl.europa.eu/sides/getDoc.do?pubRef=-//EP//NONSGML+TA+P7-TA-2011-0081+0+DOC+PDF+VO//EN> <sup>33</sup> Proposal for a Regulation of the European Parliament and of the Council on establishing a Health for Growth Programme, the third multi-annual programme of EU action in the field of health for the period 2014–2020, European Commission, 9 November 2011. [http://ec.europa.eu/health/programme/docs/prop\\_prog2014\\_en.pdf](http://ec.europa.eu/health/programme/docs/prop_prog2014_en.pdf) <sup>34</sup> Multiannual Financial Framework, 2013. <http://ec.europa.eu/budget/reform> <sup>35</sup> Charter of Fundamental Rights of the European Union, 2000. [http://www.europarl.europa.eu/charter/pdf/text\\_en.pdf](http://www.europarl.europa.eu/charter/pdf/text_en.pdf) <sup>36</sup> Pace P. What can be done in EU Member States to better protect the health of migrants? *Eurohealth* 2010;16(1):5–10

## MIGRANT HEALTH

In 2006, the WHO issued dedicated recommendations to governments to establish comprehensive plans addressing thalassaemia and sickle cell disease.<sup>37</sup>

Certain EU member states have issued comprehensive guidance on migrant health to practitioners in a number of areas, including chronic diseases, anaemias and haemoglobin disorders. However, research conducted by the International Organization for Migration and published by the London School of Economics shows disparities in the approach to migrant health across the EU and highlights the value of increased EU coordination in this respect.<sup>38 39</sup>

Research also shows that disaggregated data on migrants' status and health are also severely lacking and need to be collected.<sup>40 41</sup>

EU policy makers have recently debated the need for policy action in order to improve migrant access to healthcare services, and thus ensure the respect of the universal right to health, in accordance with their international obligations.

The EU Agency for Fundamental Rights has drawn political attention to the fact that national practices diverge widely with regard to access to healthcare of irregular migrants.<sup>42</sup> The European Parliament has called on the European Commission to further assess this issue and emphasised the need for policy action in order to ensure and promote access to appropriate healthcare for people at risk of exclusion, including migrants and intra-European Union citizens.<sup>43</sup>

## HAEMOGLOBINOPATHIES

With the exception of the ENERCA project and website (which addresses rare anaemias), initiatives at an EU level remain general for rare diseases, with no policy strategies in place that specifically address haemoglobinopathies or increasing population diversity.

The EUNENBS report on the practices of newborn screening for rare disorders in Member States of the European Union, and Candidate, Potential Candidate and EFTA Countries found significant variations in haemoglobinopathies screening panels and quantitative cut-off levels across different countries.<sup>44</sup>

The EUNENBS report recommended harmonising the approach to the screening of haemoglobin disorders by setting up regular evaluation and correction of cut-off values.

Stakeholders expect that the report will now be followed by an EU Council recommendation on newborn screening for rare conditions.<sup>45</sup>

<sup>37</sup> WHO, 2006. World Health Assembly Resolution on Sickle Cell Anaemia. WHA59.20. Executive Board Resolution on Thalassaemia and Other Haemoglobinopathies. EB118.R1. WHO, Geneva, Switzerland, 2006.

<sup>38</sup> Eurohealth Volume 16 Number 1, London School of Economics - Health and Social Care, 2010, Pace, P., London School of Economics, What can be done in EU Member States to better protect the health of migrants?, 2010, <http://www2.lse.ac.uk/LSEHealthAndSocialCare/pdf/eurohealth/VOL16No1/Pace.pdf> | <sup>39</sup> Ursula K-T, Sonja N-Z, Birgit M. Access to healthcare for undocumented migrants in the EU: A first landscape of NowHereland. Eurohealth. 2010;16(1):13-16, Available at: [http://www.lse.ac.uk/LSEHealthAndSocialCare/pdf/eurohealth/VOL16No1/Karl\\_Trummer.pdf](http://www.lse.ac.uk/LSEHealthAndSocialCare/pdf/eurohealth/VOL16No1/Karl_Trummer.pdf) | <sup>40</sup> WHO, 2008, World Health Assembly Resolution on Health of Migrants, WHA 61.17. WHO, Geneva, Switzerland, 2008. | <sup>41</sup> European Centre for Disease Prevention and Control (ECDC). Improving HIV data comparability in migrant populations and ethnic minorities in EU/EEA/EFTA countries: findings from a literature review and expert panel. Stockholm, Sweden, August 2011, Available at: [http://ecdc.europa.eu/en/publications/publications/1108\\_ter\\_improving\\_hiv\\_data\\_comparability\\_in\\_migrants.pdf](http://ecdc.europa.eu/en/publications/publications/1108_ter_improving_hiv_data_comparability_in_migrants.pdf) | <sup>42</sup> Migrants in an irregular situation: access to healthcare in 10 European Union Member States, European Union Agency for Fundamental Rights, 2011. [http://fra.europa.eu/fraWebsite/attachments/FRA-2011-fundamental-rights-for-irregular-migrants-healthcare\\_EN.pdf](http://fra.europa.eu/fraWebsite/attachments/FRA-2011-fundamental-rights-for-irregular-migrants-healthcare_EN.pdf) | <sup>43</sup> Question for oral answer to the Commission by Nadja Hirsch, et al. on behalf of the ALDE Group on Access to health care for irregular migrants on 15 November 2011, European Parliament, <http://www.europarl.europa.eu/sides/getDoc.do?pubRef=-//EP//TEXT+OQ+O-2011-000301+0+DOC+XML+VO//EN> and European Parliament Resolution on the situation of fundamental rights in the European Union, 12 December 2012, Available at: <http://www.europarl.europa.eu/sides/getDoc.do?pubRef=-//EP//TEXT+TA+P7-TA-2012-0500+0+DOC+XML+VO//EN> | <sup>44</sup> Newborn Screening in Europe Expert Opinion document. | <sup>45</sup> First meeting of the EUCERD: summary report, European Union Committee of Experts on Rare Diseases, 9-10 December 2011. <http://nestor.orpha.net/EUCERD/upload/file/Meeting/EUCERD/EUCERDPublicSummaryReport101210.pdf>

## EU Factsheet

### DATA COLLECTION AND COORDINATION

### EU REFERENCE NETWORKS

Despite efforts since 1979 to improve the prevention and care of rare diseases through effective disaggregated data sharing mechanisms, the Council of the EU recently recognised that there are still difficulties linked to coordination between countries and the shortcomings of national systems.<sup>46</sup>

Past and recent initiatives to support data collection and analysis on rare diseases include the ORPHANET web portal, which contains information on more than 6,000 rare diseases including haemoglobinopathies; the European Surveillance on Congenital Anomalies (EUROCAT) network and the recently launched Joint Action on patient registries;<sup>47</sup> and the ENERCA project and its website ([www.enerca.org](http://www.enerca.org)), which contains information on 62 rare anaemias including disease definition and data on reference centres and healthcare professionals.

The Directive on the Application of patients' rights in cross-border healthcare, adopted in March 2011,<sup>48</sup> provides for the establishment of a European Reference Network on Rare Diseases, to disseminate knowledge and sharing of best practices as a means of increasing access to quality care and treatment facilities.

One of the main objectives of ENERCA,<sup>49</sup> supported by the European Commission, is the establishment of a European Reference Network (ERN) of Centres of Expertise (CEs) on Rare Anaemias. The first goal is the preparation of recommendations for the identification of Centres of Expertise on Rare Anaemias in Europe and the establishment of the ERN.

These recommendations will be published in the ENERCA White Book, which will be released in 2013, and will include recommendations on services to be provided by CEs in rare anaemias and for the establishment of national networks and the ERN.

<sup>46</sup> Council Recommendation of 8 June 2009 on an action in the field of rare diseases, 8 June 2009, <http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:C:2009:151:0007:0010:EN:PDF>

<sup>47</sup> Joint Action on patient registries, <http://ec.europa.eu/eahc/news/news139.html>, 2012. <sup>48</sup> Directive 2011/24/EU of the European Parliament and of the Council of 9 March 2011 on the application of patients' rights in cross-border healthcare, 2011. [http://eur-lex.europa.eu/smartapi/cgi/sga\\_doc?smartapi!celexplus!prod!CELEXnumdoc&lg=EN&numdoc=32011L0024](http://eur-lex.europa.eu/smartapi/cgi/sga_doc?smartapi!celexplus!prod!CELEXnumdoc&lg=EN&numdoc=32011L0024)

<sup>49</sup> European Network for Rare and Congenital Anaemias (ENERCA), 2013. <http://www.enerca.org/about-enerca/objectives.html>

## EU Factsheet

### RESEARCH AND INNOVATION

### POLICY OUTLOOK

The 2009 Council Recommendation on a European action in the field of rare diseases<sup>50</sup> called for the improvement of the funding and coordination of research programmes, with a particular emphasis on funding for rare diseases under the Seventh European Research Framework (FP7).

There are currently 66 ongoing research projects on rare diseases under FP7, with total funding of around EUR 325 million.<sup>51</sup>

A number of past and current EU-funded projects focus on data collection, prevention and treatment of haemoglobinopathies, including thalassaemia and sickle cell disease.<sup>52 53 54</sup>

The future EU Framework Programme for Research and Innovation (2014–2020), Horizon 2020,<sup>55</sup> is also expected to support international cooperation for the development of new diagnoses and care therapies in the area of rare diseases.

By the end of 2013, the European Commission is expected to publish an implementation report on both the Council Recommendation and Commission Communication on EU action on rare diseases, in order to assess the progress and impact of the measures undertaken at a national and EU level, and to evaluate the need for further action to improve healthcare for patients affected by rare diseases.

The European Union Committee of Experts on Rare Diseases (EUCERD) will support the Commission in this task.<sup>56</sup> Member states are also working on the implementation of commitments to address health inequalities, as a follow-up to the European Commission's Communication on 'Solidarity in Health: reducing health inequalities in the EU'. In particular, within the framework of Europe 2020<sup>57</sup> (the 10-year strategy for Europe's smart and inclusive growth), the EU plans to support member states' and stakeholders' actions against inequalities, in particular by targeting the use of EU funds to reduce health inequalities between regions. It also regularly produces statistics and reports on inequalities in the EU, with specific recommendations and strategies to reduce them.

The EU Directive on Cross-border Healthcare is expected to be fully implemented across EU member states by October 2013. The Directive, however, only applies to people legally residing in an EU member state and with health insurance and, therefore, does not cover all types of migrant or even non-insured EU citizens.

<sup>50</sup> Council Recommendation of 8 June 2009 on an action in the field of rare diseases, Official Journal of the European Union, 8 June 2009, <http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:C:2009:151:0007:0010:EN:PDF> | <sup>51</sup> Rare Disease Research in FP7, Outlook in Horizon 2020, IRDiRC European Commission, 24 May 2012. [http://www.google.be/url?sa=t&rct=j&q=&esrc=s&source=web&cd=1&ved=OCFEQFjAA&url=http%3A%2F%2Fdownload.eurordis.org%2Fecrd2012%2FT4S0402\\_IEROLA.pdf&ei=hRUnULDeJ-ms0QWMzaDvBA&usg=AFQjCNH3BDhZeX0AA4J8dF02yredod-7L0w&sig2=VRlbWH4vsg-vh7vd6VV2A](http://www.google.be/url?sa=t&rct=j&q=&esrc=s&source=web&cd=1&ved=OCFEQFjAA&url=http%3A%2F%2Fdownload.eurordis.org%2Fecrd2012%2FT4S0402_IEROLA.pdf&ei=hRUnULDeJ-ms0QWMzaDvBA&usg=AFQjCNH3BDhZeX0AA4J8dF02yredod-7L0w&sig2=VRlbWH4vsg-vh7vd6VV2A) | <sup>52</sup> Deferiprone Evaluation in Paediatrics, European Commission, European R&D Projects, 2013. [http://cordis.europa.eu/projects/rcn/97667\\_en.html](http://cordis.europa.eu/projects/rcn/97667_en.html) | <sup>53</sup> Electronic Infrastructure for Thalassaemia Research Network, 2013. <http://www.ithanet.eu> | <sup>54</sup> Infrastructure for Thalassaemia Research Network, European R&D Projects, European Commission, 2013. <http://cordis.europa.eu/projects/index.cfm?fuseaction=app.details&TXT=haemoglobinopathies&FRM=1&STP=10&SIC=&PGA=&CCY=&PCY=&SRC=&LNG=en&REF=80459> | <sup>55</sup> Proposal for a Council Decision establishing the Specific Programme Implementing Horizon 2020 - The Framework Programme for Research and Innovation (2014–2020), European Commission, 30 November 2011. <http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=CELEX:52011PC0811:EN:NOT> | <sup>56</sup> 2012 Report of the state of the art of rare disease activities in Europe of the EUCERD, European Union Committee of Experts on Rare Diseases, 2012. <http://www.eucerd.eu/upload/file/Reports/2012ReportStateofArtRDActivities.pdf> | <sup>57</sup> Europe 2020. [http://ec.europa.eu/health/europe\\_2020\\_en.htm](http://ec.europa.eu/health/europe_2020_en.htm)

## Country Factsheet: Belgium

### KEY FACTS AND FIGURES



Belgium is not considered an endemic country for haemoglobinopathies. While estimated prevalence data exist, this information does not specify migrants' status, country of origin or gaps at the time of diagnosis.

A Belgian survey from 2007 estimated the number of patients with a major haemoglobinopathy at approximately 500.<sup>58</sup> This includes around 400 (or 0.0036% of the total population) with sickle cell disease (SCD) and around 100 with thalassaemia major (0.0009% of the total population).<sup>59</sup>

Every year, 26 conceptions are estimated to be affected by SCD, and two by  $\beta$ -thalassaemia major.<sup>60</sup> The annual number of pregnancies at risk for SCD is estimated to be 105 and for  $\beta$ -thalassaemia, 10.<sup>49</sup>

Healthcare professionals interviewed believe that the number of new patients diagnosed every year is greatly dependent on current and previous migration flows into Belgium. Larger cities such as Brussels and Liège have the highest number of SCD cases, with an incidence of 1:1,600 and 1:1,950 respectively.<sup>58</sup> The prevalence of SCD is expected to substantially increase in the near future due to a decreased mortality rate. The prevalence of thalassaemia is expected to remain relatively low (precise data and estimates are not available, but in Brussels the incidence is around 1:40,000).

The patient's country of origin is considered an important prevalence factor for haemoglobinopathies. Data from 2005 shows that 3.9% of immigrants to Belgium originated from countries with a high prevalence of haemoglobinopathies.<sup>61</sup> Data from 2009 found that 97.5% of parents in Belgium with neonates suffering from SCD come from sub-Saharan Africa and those with thalassaemia are mostly from North Africa, Italy or the Middle East.<sup>62</sup>

There are no official Belgian data on main sequelae and mortality linked to haemoglobinopathies. The average life expectancy of haemoglobinopathy patients in Belgium is estimated to be more than 40 years, which according to healthcare professionals is mainly due to increased access to new medication and care, as well as absence of malarial infection. Experts generally consider the most common causes of mortality as heart iron overload linked to thalassaemia and infections and long-term organ damage linked to SCD.

In Belgium there is no official registry of relevant data on haemoglobinopathies. Since 2007, The Red Blood Cell Disorders Subcommittee of the Belgian Haematological Society<sup>63</sup> has maintained a centralised Belgian registry of patients with SCD.<sup>64</sup> The registry can only be accessed by healthcare professionals, for data privacy reasons.

The federal government does plan, however, to establish a registry to gather and analyse data on a number of rare diseases, as a follow-up to the EU Council Recommendation on Rare Diseases of 8 June 2009.<sup>66</sup> The registry is expected to integrate existing rare disease records that have already been developed on a voluntary and/or local level (including haemoglobinopathies registries), and to gather new data. Unfortunately, progress with the registry has been slow due to budgetary cuts.

In collaboration with the *Institut national d'assurance maladie-invalidité*<sup>67</sup> (INAMI, National Institute of Health and Disability Insurance), a universal neonatal screening programme for haemoglobinopathies has been conducted in the Brussels region since December 1994.<sup>68</sup> The aim of this programme is mainly to improve healthcare for patients with SCD. The programme has significantly reduced morbidity and mortality associated with SCD in infancy and early childhood.<sup>69</sup>

There are no data on the estimated costs linked to haemoglobinopathies.

<sup>58</sup> Gulbis B, et al. on behalf of the Red Blood Cell Disorders Subcommittee of the Belgian Haematological Society. An estimation of the incidence and demographic picture of the major hemoglobinopathies in Belgium (from a confidential inquiry). *Hemoglobin* 2008;32(3):279-285 | <sup>59</sup> Gulbis B, et al. Haemoglobinopathies in Belgium. *Belgian Journal of Haematology* 2010;1:50-560 | <sup>60</sup> Modell B, Darlison M, Birgens H, Cario H, Faustino P, Giordano PC, Gulbis B, Hopmeier P, Lena, Russo D, Romao L, Theodorsson E. Epidemiology of haemoglobin disorders in Europe: an Overview. *Scand J Clin Lab Invest*. 2007;67(1):39-69. Review. | <sup>61</sup> Haemoglobinopathies in Belgium, op.cit. | <sup>62</sup> Haemoglobinopathies in Belgium, op.cit. | <sup>63</sup> The Red Blood Cell Disorders Subcommittee of the Belgian Haematological Society, 2013. <http://www.bhs.be/committees/red-cells> | <sup>64</sup> Belgian Registry of Patients with SCD, <https://www.drepano.be/> | <sup>65</sup> Haemoglobinopathies in Belgium, op. cit. | <sup>66</sup> Council Recommendation of 8 June 2009 on an action in the field of rare diseases, Official Journal of the European Union, 8 June 2009, <http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:C:2009:151:0007:0010:EN:PDF> | <sup>67</sup> Institut national d'assurance maladie-invalidité (INAMI), 2013. <http://www.inami.fgov.be/homefr.htm> | <sup>68</sup> Gulbis B, et al. Neonatal haemoglobinopathy screening: review of a 10-year programme in Brussels. *J Med Screen* 2006;13(2):76-78 | <sup>69</sup> Lê PQ, et al. Sickle cell disease from Africa to Belgium, from neonatal screening to clinical management. *Med Trop (Mars)* 2010;70(5-6):467-470

## Country Factsheet: Belgium

### POLICY FOCUS

### THE NATIONAL RARE DISEASE PLAN

In Belgium, haemoglobinopathies are not a focus of specific health policy. Although the government recognises that improvements in disease awareness, screening and treatment could be made, it considers that patients generally access high quality care.

In 2011 the Belgian Ministry of Social Affairs and Public Health tasked a healthcare experts group (ETHEALTH)<sup>70</sup> to develop recommendations for adapting current Belgian healthcare policies to the presence of migrants and ethnic minorities in Belgium. The ETHEALTH report<sup>71</sup> was published in December 2011.

The report highlighted the need to establish targeted strategies to evaluate the inequalities linked to ethnicity, and to take an 'intercultural' approach across public healthcare professional bodies.

Experts also recommended implementing targeted measures to strengthen prevention, health promotion and access to quality care at hospital and primary care levels, in particular by raising awareness and competence levels in the delivery of care to cultural and ethnic minority groups.<sup>72</sup>

In June 2012, following the ETHEALTH recommendations, the Belgian Council of Ministers<sup>73</sup> approved measures aimed at enhancing access to healthcare among the migrant population. The proposed measures include a series of financial mechanisms to ensure affordability of care and treatment, and are expected to benefit patients with a rare or orphan disease from 2013.<sup>74</sup>

Haemoglobinopathies fall in the category of rare diseases in Belgium (they affect less than five in 10,000 people). Over 65,000 patients and their families are estimated to be affected by a rare disease in Belgium.<sup>75</sup>

The Belgian government plans to establish a rare disease plan and registry.<sup>76</sup> The Fund for Rare Diseases and Orphan Drugs<sup>77</sup> was appointed responsible for coordinating this work by the Minister of Social Affairs and Public Health and INAMI.

The Fund is administered by the Fondation Roi Baudouin (FRB, an independent foundation for the improvement of living conditions<sup>78</sup>), which has received state funding for this general interest mission. The main objective of the Fund is to create a comprehensive framework to improve the diagnosis and care of patients, including access to validated information, and patients' and families' quality of life.

In its recommendations from September 2011, the Fund outlined an initial set of suggestions and steps to shape the 5-year rare diseases plan, under four main pillars of action:

- Organisation of specific clinical skills to improve health outcomes for patients
- Codification and identification of rare diseases
- Increase awareness on the issue, informing stakeholders and empowering patients
- Access to diagnosis, medication, treatment and care, with its direct and indirect costs<sup>79</sup>

<sup>70</sup> Rapport: Vers des soins de santé interculturels, Federal Public Service, Food Chain Safety and Environment, 21 December 2011. [http://www.health.belgium.be/eportal/Myhealth/19074155\\_FR?fodnlang=en](http://www.health.belgium.be/eportal/Myhealth/19074155_FR?fodnlang=en) <sup>71</sup> Vers des soins de santé interculturels, V. Lorant, I. Derluyn, M. Dauvrin, I. Coune, H. Verrept, ETHEALTH, December 2011, [http://www.health.belgium.be/filestore/19074152\\_FR/Ethealth\\_FR.pdf](http://www.health.belgium.be/filestore/19074152_FR/Ethealth_FR.pdf) <sup>72</sup> Rapport: vers des soins de santé interculturels, FPS Health, Food chain safety and Environment, op.cit <sup>73</sup> Conseil des ministres du 15 juin 2012, SPF Chancellerie du Premier Ministre, 15 June 2012. <http://www.presscenter.org/fr/pressrelease/20120615/conseil-des-ministres-du-15-juin-2012> <sup>74</sup> Mormont M. De meilleurs soins pour les plus vulnérables. Alter Echo, 28 June 2012. [http://www.alterechos.be/?p=breves&d=i&c=a&art\\_id=22573&listby=day&art\\_date=2012\\_06\\_28](http://www.alterechos.be/?p=breves&d=i&c=a&art_id=22573&listby=day&art_date=2012_06_28) <sup>75</sup> Mieux soutenir les personnes atteintes de maladies rares!, V. Lievin, rtl.be, 15 February 2012, <http://blogs.rtl.be/santevousbien/2012/02/15/mieux-soutenir-les-personnes-atteintes-de-maladies-rares/> <sup>76</sup> Plan belge pour les maladies rares, J. Ventura, EURODIS, 30 January 2012, <http://www.eurordis.org/fr/content/plan-belge-pour-les-maladies-rares> <sup>77</sup> Fonds Maladies rares et médicaments orphelin, Fondation Roi Baudouin, 2011. <http://www.kbs-frb.be/fund.aspx?id=293667&langtype=2060> <sup>78</sup> Fondation Roi Baudouin, Mission: Working together for a better society, 2013. <http://www.kbs-frb.be/content.aspx?id=290655&langtype=1033> <sup>79</sup> Recommandations et propositions Plan belge pour les Maladies Rares, Fonds Maladies Rares et Médicaments Orphelins, 2011. <http://www.debra-belgium.org/fr/debra-belgium/maladies-rares/>



## THE NATIONAL RARE DISEASE PLAN

The experts in charge of drafting the recommendations consulted healthcare professionals and patient associations. There are currently no plans to consult them on the final draft plan, however. At the time of writing this report, the Health Minister's office (along with experts from INAMI) were incorporating the Fund recommendations into a draft rare diseases plan, although the timing for its adoption remains imprecise. Stakeholders, including the FRB, fear that current cost containment measures could delay these key developments.

However, a few of the recommendations have started to be implemented. The *Institut Scientifique de la Santé Publique*<sup>80</sup> (ISP, Public Health Scientific Institute) has drafted a concept paper on the future registry of rare diseases in Belgium<sup>81</sup> and has set up ORPHANET Belgium, a reference portal for rare diseases and orphan drugs.

Measures to improve collaboration with patient organisations and healthcare professionals have also been taken. The Health Ministry's programme entitled 'Priority to chronic illness populations', led by INAMI, created the *Observatoire des maladies chroniques* (observatory of chronic diseases).<sup>82</sup> The Observatory is a consultation forum for healthcare professionals and patient associations, brought together through the *Alliance Belge des Maladies Rares - Radiorg* (Belgian Rare Diseases Alliance).<sup>83</sup>

## PREVENTION AND SCREENING

**There is no official prenatal screening programme for haemoglobinopathies in place. However, a test before conception to detect SCD carriers is recommended to individuals at risk, based on family records and ethnic origin.**<sup>84</sup>

In 2004, the Belgian authorities noted a lack of relevant evidence in favour of establishing systematic prenatal screening for haemoglobinopathies. In 2009, the Belgian Haematological Society issued guidelines for prenatal and neonatal screening for haemoglobinopathies.<sup>85</sup> These recommendations were adopted by a network of Belgian obstetricians (GGOLFB).<sup>86</sup> They recommend screening of all women between the 8th and 10th weeks of pregnancy, through a blood test and evaluation of family origins. If the woman is identified as carrier of a haemoglobinopathy, the partner will also be tested in order to assess the risk of a major haemoglobinopathy for the foetus.

Systematic screening programmes on haemoglobinopathies for neonates are practised for all maternity wards in Brussels Region and in Liège. Members of a red blood cell committee within the Belgian Haematological Society have implemented specific protocols on the diagnosis of haemoglobinopathies, how to inform and educate patients and their families, and how to ensure follow-up care.<sup>87</sup>

According to consulted healthcare professionals, screening and diagnosis of health sequelae are conducted according to the needs of the patient, and vary depending on the financial and technological resources of the hospital. Patients believe that access to transcranial Doppler screening for SCD children and magnetic resonance screening for thalassaemia patients is not consistent across the country, as it depends largely on the expertise of healthcare professionals in the hospital. Professionals consider that there is still a lack of information and training for health professionals in these domains.

<sup>80</sup> Belgian Scientific Institute for Public Health, 2013. <https://www.wiv-isp.be/Pages/EN-Home.aspx?pf1g=1033> | <sup>81</sup> Orphanet Belgique, 2013. <http://www.orpha.net/national/BE-FR/index/page-d-accueil/> | <sup>82</sup> Communiqué de presse: Priorité aux malades chroniques! Inauguration de l'Observatoire des maladies chroniques, INAMI, 2 May 2012. <http://www.inami.fgov.be/news/fr/press/pdf/press2012050201.pdf> | <sup>83</sup> Radiorg (Belgian Rare Diseases Alliance), 2013. <http://www.radiorg.be/fr/> | <sup>84</sup> Review Hematology, Hemoglobinopathies in Belgium, B. Gulbis, A. Ferster, F. Vertongen; on behalf of the hereditary red blood cell disorders subcommittee - Belgian Hematological Society, *Belgian Journal of Hematology* Volume 1, Issue 2, December 2010 | <sup>85</sup> Haemoglobinopathies in Belgium, op.cit | <sup>86</sup> Guide de consultation prénatale, Alexander S, et al. Guide de consultation prénatale. de Boeck, 2009. | <sup>87</sup> Gulbis B. Neonatal screening for haemoglobinopathies: what is the situation in Belgium? February 2010. [http://www.kuleuven.be/vakb/pages/VAKB03feb10/VAKB\\_Gulbis\\_screening\\_hemoglobinopathies.pdf](http://www.kuleuven.be/vakb/pages/VAKB03feb10/VAKB_Gulbis_screening_hemoglobinopathies.pdf)

## Country Factsheet: Belgium

### TREATMENT AND CARE

### EDUCATION

The Belgian social security system ensures partial reimbursement of costs related to treatment, medication, visits to physicians, and tests. The level of financial support received by a patient depends on the established annual reimbursement ceiling, and the patient's income and health insurance plan.<sup>88 89</sup>

Care for patients with haemoglobinopathies in Belgium is mainly provided by specialised doctors. Where neonatal screening is implemented, there is a reference person for haemoglobinopathies in each hospital involved. A preliminary contact is made with the paediatric centre when there is no specialist doctor.

Some hospitals have reference centres and receive funds from INAMI to finance specific treatment and care in the disease in which they are specialised. Currently, there is no reference centre for haemoglobinopathies subsidised by INAMI. Stakeholders recognise this as a consequence of budgetary constraints.

Healthcare professionals consider that specialised centres in the country provide a high level quality of care to haemoglobinopathies patients. They also note that access to specialised care is ensured where it is needed. Furthermore, public authorities believe that given the country's small size, travelling and access to healthcare in Belgium is not considered to be a major problem. Regular oral chelation and blood transfusion therapy is widely accessible across the country; organ transplants are carried out only in key health care facilities in bigger cities. Healthcare professionals stress, however, that increased funding is crucial in order to ensure a holistic approach to patient care. Experts note that the specialised services in hospitals do not currently have the financial resources necessary for the training of healthcare professionals on a more local level, which they believe is essential for quality of care.

Patients and healthcare professionals agree that only a few doctors are fully aware of the health risks, sequelae and social impact derived from haemoglobinopathies. Awareness among the general public is considered to be almost non-existent.

Healthcare professionals also stress that patient education is the largest challenge in the management of haemoglobinopathies. Physicians remain patients' main source of information on these diseases. In Brussels, dedicated sessions are regularly organised by healthcare professionals to inform patients about the diseases, linked health risks, importance of good care and follow up, what to do in case of crisis, etc. Currently, health services often lack the cultural mediation and interpretation services to effectively handle patients of migrant origin, posing difficulties in communication and the dissemination of information. (<http://www.uclouvain.be/cps/ucl/doc/ir-irss/images/ETHEALTH-FINAL1.pdf>)

Government sources acknowledge that diagnosis could be improved with awareness raising campaigns as well as patient and physician education and training. These elements are managed at a regional and community level, however. The Fondation Roi Baudouin recommendations for the future plan on rare diseases include specific measures aimed to raise awareness and increase knowledge of rare diseases among patients, people at risk, healthcare professionals and other relevant target groups such as teachers and social workers. However, haemoglobinopathies are not specifically mentioned, and patients and experts agree that haemoglobinopathies do not appear to be a priority. Patient associations and groups of medical experts have developed educational materials and communication tools focused on haemoglobinopathies that are available to the public, such as a comic strip style cartoon for SCD children and their families.<sup>90</sup> These patient organisations, mostly run by volunteers, provide advice, information and support to both migrant and non-migrant patients and other important stakeholders, including patient families. The patient group Action Drepanocytose is expected to release a report outlining its activities in 2013.

<sup>88</sup> La sécurité sociale belge, INAMI, 2010. [http://www.inami.be/presentation/fr/social\\_security/index.htm](http://www.inami.be/presentation/fr/social_security/index.htm) | <sup>89</sup> Maximum à facturer, INAMI, 2010. [http://www.riziv.fgov.be/citizen/fr/medical-cost/SANTH\\_4\\_3.htm](http://www.riziv.fgov.be/citizen/fr/medical-cost/SANTH_4_3.htm) | <sup>90</sup> Neonatal haemoglobinopathy screening: review of a 10-year programme in Brussels, op.cit | <sup>91</sup> Recommandations et propositions de mesures en vue du Plan belge pour les Maladies Rares, Fonds Maladies Rares et Médicaments Orphelins, Fondation Roi Baudouin, October 2010, [http://www.kbs-frb.be/uploadedFiles/KBS-FRB/05\)\\_Pictures\\_documents\\_and\\_external\\_sites/09\)\\_Publications/PUB\\_2024\\_PlanBelgePourLesMaladiesRares\\_DEF.pdf](http://www.kbs-frb.be/uploadedFiles/KBS-FRB/05)_Pictures_documents_and_external_sites/09)_Publications/PUB_2024_PlanBelgePourLesMaladiesRares_DEF.pdf)

## Country Factsheet: Belgium

### POLICY OUTLOOK

#### EXPECTED POLICY DEVELOPMENTS AND TIMELINE

Ongoing work to develop a national plan on rare diseases (timing for adoption remains imprecise).<sup>91</sup>

Ongoing dialogue with health authorities (INAMI) to establish systematic neonatal screening across the country.

Implementation of targeted measures to enhance migrants' access to healthcare (as of 2013).

#### INTERVIEWEES' SUGGESTED POLICY ACTIONS

Government officials regard the screening, care and treatment of haemoglobinopathies as already quite positive from a qualitative point of view.

Haemoglobin disorders are not a health priority for action for the current Belgian government. Government sources also note that improved patient and healthcare professional education can help address a number of existing gaps, but this falls under the competences of the regions.

Healthcare professionals however stress that there is a need for urgent measures to ensure holistic, long-term and quality care for haemoglobinopathies patients.

Healthcare professionals call on the government to support the establishment of a dedicated, long-term plan at national level, as this is seen as an essential step to improve the delivery of care in Belgium.

**“WE GOT AND GET SUPPORT FROM NATIONAL AUTHORITIES. THIS HAS ALLOWED US TO IMPROVE DIAGNOSIS, PREVENTION AND TREATMENT OF PATIENTS WITH HAEMOGLOBINOPATHIES.**

**NEVERTHELESS, NOW IS NOT THE TIME TO LOSE THE ACTIONS THAT HAVE BEEN IMPLEMENTED AND NOT TO CONTINUE THE WORK. WE WOULD SAVE TIME AND MONEY IF THERE IS A LONG-TERM VISION AND A CENTRALLY ORGANISED STRUCTURE TO ENSURE DELIVERY OF QUALITY CARE TO PATIENTS WITH MAJOR HAEMOGLOBINOPATHIES.”**

Béatrice Gulbis, Erasme Hospital, Université Libre de Bruxelles

## Country Factsheet: Cyprus

### KEY FACTS AND FIGURES



Thalassaemia is endemic in Cyprus. It is estimated that around one in 1,000 Cypriots suffer from thalassaemia.<sup>92</sup> There is a national registry on thalassaemia for healthcare professionals of the Thalassaemia Centres that are part of the Cyprus Ministry of Health. In order to facilitate management and patients' access, an electronic version is being implemented.<sup>93</sup>

The latest data from the national Cyprus Register indicate that there are 962 patients with thalassaemia in Cyprus (592  $\beta$ -thalassaemia major, 93  $\beta$ -intermedia and 277 HbH disease). Epidemiological data demonstrate a 17%  $\beta$ -thalassaemia carrier rate amongst the Cypriot population, of which 20–25% are carriers of  $\alpha$ -thalassaemia.

Sickle cell disease (SCD) is considerably less common, with only an estimated 44 cases in the country, according to the Cyprus Registry.<sup>94</sup> As such, SCD is considered a rare anaemia, although there is concern that the number of patients is rising.<sup>95</sup>

In the last three decades, the number of newborns with haemoglobin disorders has decreased dramatically. In 1979, for example, there were only 18 cases, as opposed to the projected 77 expected cases for that year.<sup>96</sup> Between 1991 and 2001, only five babies were born with thalassaemia, one every 2–3 years. Furthermore, no babies with thalassaemia have been born since 2006 in the North of Cyprus.<sup>97</sup>

Notably, however, there has been a recent rise in the number of affected births: in 2011, there were six new thalassaemia births, with a further six in 2012. At the time of writing, there have been four new cases reported in 2013. This is a new trend, which deserves further analysis.

Survival of patients with haemoglobinopathies is high. Over 90% of patients live beyond 45 years old, making Cyprus the country with perhaps the oldest population of thalassaemia patients. Cardiac failure, liver disease and infection are considered the main complications leading to death amongst thalassaemia patients.<sup>98</sup>

There are no official data available on the costs related to haemoglobinopathies in Cyprus. In the 1970s, the government, with the support of the WHO, estimated that if Cyprus did not implement an effective prevention programme, by 2010 more than 50 percent of the population would need to donate blood in order to support affected haemoglobinopathy patients. This would have resulted in estimated costs equivalent to 70% of the entire national health budget.<sup>99</sup>

Currently, it is estimated that approximately 12% of the healthcare budget is allocated to support prevention, care and management of thalassaemia (screening and care of haemoglobinopathy patients is provided free of charge).

<sup>92</sup> Kolnagou A, Kontoghiorghe GJ. Advances in the prevention and treatment are changing thalassaemia from a fatal to a chronic disease. experience from a Cyprus model and its use as a paradigm for future applications. *Hemoglobin* 2009;33(5):287–295 | <sup>93</sup> Activity Report 2011 and Plan of Activities 2012, Thalassaemia International Organisation (TIF), 2011. <http://www.thalassaemia.org.cy/docs/2012%20TIF%20Plan%20F%20Action.pdf> | <sup>94</sup> Figures derived from the National Register in Cyprus. | <sup>95</sup> Ευριπίδου Ν. Μια νέα απειλητική ασθένεια (A New Threatening Disease), 13 January 2012. <http://www.sigmalive.com/simerini/news/health/455080> | <sup>96</sup> Angastiniotis MA, Hadjiminias MG. Prevention of thalassaemia in Cyprus. *Lancet* 1981;1(8216):369–371 | <sup>97</sup> Bozkurt G. Results from the North Cyprus thalassaemia prevention programme. *Haemoglobin* 2007;31(2):257–264 | <sup>98</sup> Telfer P. Survival of medically treated thalassaemia patients in Cyprus. Trends and risks factors over the period 1980–2004. *Haematologica* 2006;91:1187–1192 | <sup>99</sup> Η θαλασσαιμία βρίσκεται παντού στην Μεσόγειο (Thalassaemia is found everywhere in the Mediterranean), speech by Mr Christophias, President of the Republic of Cyprus, 28 January 2010. <http://www.sigmalive.com/simerini/news/local/233372>

**“ENSURING ACCESS TO QUALITY AND INNOVATIVE CARE FOR THALASSAEMIA PATIENTS IS A HIGH PRIORITY FOR THE CYPRUS GOVERNMENT.”**

Sotiroulla Christou, Medical Head  
of the Cyprus Thalassaemia Centre

## Country Factsheet: Cyprus

### POLICY FOCUS

The main health policy priorities of Cyprus include cancer and rheumatic diseases, as well as maternal health.<sup>100</sup> The national control programme and clinical management of haemoglobinopathies is one of the highest public health priorities.

Haemoglobinopathies are not considered rare diseases in Cyprus,<sup>101</sup> although SCD is identified as a rare anaemia.<sup>102</sup> A dedicated thalassaemia national plan has been in place since 1969, under the Ministry of Health with the close support and collaboration of the Ministry of Education.<sup>103</sup> Cyprus' policies are considered effective, largely due to the accompanying education programmes that aim to raise awareness and acceptance of screening amongst the general population.<sup>104</sup> However, experts stress that there is an increasing impact of migration on health and, therefore, health services should be alert to epidemiological change and the need to develop policies for comprehensive services to meet the needs of these disorders (e.g. informed choice and access to expert management). Experts further emphasise that the active involvement of the medical community, including physicians, paediatricians and obstetricians, as well as religious, family planning associations and social workers, has been instrumental to the success of awareness and prevention measures in Cyprus.<sup>105 106</sup>

The Greek Orthodox Church<sup>107</sup> has actively encouraged the prevention strategy with the introduction of the premarital certificate in 1983. Following prenatal diagnosis, pregnancy termination is the choice of a great majority of 'at risk' couples in Cyprus, an option that is provided by the public healthcare service and is free of charge. Stakeholders paved the way to improving prevention and care by developing initiatives that were later endorsed and established at policy level (e.g. national blood donation programme<sup>108 109</sup>). It is believed that Cyprus is the first country to establish voluntary, non-remunerated blood donation policies that have led to the self-sufficiency of Cyprus in terms of adequate blood supply for the care of patients on a national level, including those with thalassaemia. In recent years, however, blood shortages have become more frequent. This has an impact on thalassaemia patients, who may have to postpone treatments. Cypriot patients and parents have also driven international mobilisation and recognition of thalassaemia, leading to the creation of the Thalassaemia International Federation<sup>110</sup> (TIF) in 1986, headquartered in Nicosia.

Patients and experts agree that general awareness of the risks, sequelae and social impact linked to haemoglobin disorders is high across the country. Indeed, awareness-raising was addressed under the first Thalassaemia Programme adopted by the government in 1969.<sup>111</sup> However, according to patients, although understanding in society is considered to be good overall, difficulties still remain in the private sector, with companies sometimes still reluctant to hire individuals with thalassaemia. In response, the government is promoting the fact that advances in care and treatment have resulted in thalassaemia patients being able to lead normal careers, without significant absenteeism.

<sup>100</sup> Health Strategies, Cyprus Ministry of Health, 2013. [http://www.moh.gov.cy/moh/moh.nsf/page75\\_gr/page75\\_gr?OpenDocument](http://www.moh.gov.cy/moh/moh.nsf/page75_gr/page75_gr?OpenDocument) | <sup>101</sup> Annual Report 2010, Cyprus Ministry of Health, 2010. [http://www.moh.gov.cy/moh/moh.nsf/EEBCAF0CDB3C0C4FC22577BB0026941E/\\$file/Ετήσια%20Εκθεση%202010.pdf](http://www.moh.gov.cy/moh/moh.nsf/EEBCAF0CDB3C0C4FC22577BB0026941E/$file/Ετήσια%20Εκθεση%202010.pdf) | <sup>102</sup> FMια νέα απειλητική ασθένεια (A New Threatening Disease), Νατάσα Ευριπίδου (Natasa Eyripidou), η Σημερινή, 13 January 2012, <http://www.sigmalive.com/simerini/news/health/455080> | <sup>103</sup> Angastiniotis M, et al. How thalassaemia was controlled in Cyprus. *World Health* 1986;(7):291-297 | <sup>104</sup> How thalassaemia was controlled in Cyprus, op.cit | <sup>105</sup> Prevention of Thalassemias and other haemoglobin disorders, op. cit. | <sup>106</sup> How thalassaemia was controlled in Cyprus, op. cit. | <sup>107</sup> In part this is due to the active lobbying of the Cypriot Antianaemic Society, which began to advocate for mandated government screening. They were regularly refused, as no legislator would vote for it, fearing condemnation by the Orthodox Church. As a consequence, physicians and parents addressed Archbishop Chrysostomos (then the head of the Church) and the bishops who consult with him on Church policy. | <sup>108</sup> A National blood donation programme, Παγκύπριας Συντονιστικής Επιτροπής Αιμοδοσίας και Διαφώτισης (ΣΕΑΔ) | <sup>109</sup> Grigoriou P. H... αιμοδιψούσα Κύπρος: δηλώνουν αιμοδότες, αλλά αίμα σπανίως δίνουν (Bloodthirsty Cyprus: many register as blood donors but rarely give blood), 3 November 2010. <http://www.sigmalive.com/simerini/news/local/322015> | <sup>110</sup> Thalassaemia International Federation, 2013. <http://www.thalassaemia.org/> | <sup>111</sup> How thalassaemia was controlled in Cyprus, op. cit.

**“THANKS TO THE OUTSTANDING EFFORTS LED BY THE NATIONAL BLOOD DONATION PROGRAMME, THE CYPRUS GOVERNMENT AND THE PANCYPRIAN THALASSAEMIA ASSOCIATION AS EARLY AS THE 1970s, THE SCREENING AND PREMARITAL PROGRAMME HAS BEEN EXTREMELY EFFECTIVE. WE ARE, IN ESSENCE, THE LAST GENERATION OF PATIENTS WITH THALASSAEMIA IN CYPRUS.”**

President of the Pancyprian Thalassaemia Association

## Country Factsheet: Cyprus

### PREVENTION AND SCREENING

### TREATMENT AND CARE

Screening programmes have been widely implemented in Cyprus since the 1970s. Experts believe that they have been crucial in reducing the thalassaemia newborn rate in Cyprus to nearly zero.

Carrier detection is conducted by specialised units and made available to the entire population, although it is not mandatory. As part of the country's prevention methods, premarital counselling is also offered. The premarital certificate required by the Greek Orthodox Church does not disclose in any way screening results, which remain confidential to the patients and do not in any way affect the patients' choice or right to marry. Experts have highlighted that the support of the Church has been crucial in improving the prevention of haemoglobin disorders.<sup>112</sup> Among Turkish Cypriots, screening is organised by the civil authorities. Screening consists of a blood test and specialised molecular analysis, when required.

Healthcare professionals believe that government support guarantees access to quality prevention and care for all affected patients. Experts welcome awareness and commitment from policy-makers in improving quality of life and healthcare delivery for haemoglobinopathy patients.

Discussions are ongoing with regard to re-establishing educational programmes within schools, in order to encourage screening and prevent conception of affected fetuses. Formal education on inherited anaemias has been introduced into the school curriculum for children over 15 years of age. Targeted campaigns are also organised by patients, the Ministry of Health and the Thalassaemia Centres. Schools often organise screening on their own initiative as part of haemoglobinopathies education campaigns.

Four dedicated thalassaemia centres exist across the country (Nicosia, Paphos, Limassol and Larnaka). The Nicosia Thalassaemia Centre has been designated an official collaborating centre by the WHO since 1986.

The Centre is responsible for ensuring the consistency and quality of care, in line with the recommendations on the clinical management of treatment for thalassaemia, developed by the Thalassaemia International Federation.<sup>113</sup> Relevant experts within the medical community and the Ministry of Health are currently working together to develop a national strategy including guidelines and clinical protocols on SCD prevention and care management.<sup>114</sup> Currently, the public healthcare system provides free screening and diagnosis for all individuals, as well as free care and regular treatment for patients. Experts, therefore, consider that the policy response, funding and access to care in Cyprus are among the best in the world. Clinical applied research is a high priority for both the government and specialised medical units. Research focuses on improving existing and developing new therapies to address haemoglobinopathies and derived diseases, such as chronic viral hepatitis, osteoporosis, and heart and liver complications linked to iron overload.<sup>115</sup> Experts and patient stress, however, that a number of challenges remain, including the need to improve healthcare professional training and awareness of haemoglobinopathies, and increase the number of specialised haematologists practising on the island. Patient groups, supported by the Ministry of Health, have run public education campaigns to prevent stigmatisation and promote labour integration.<sup>116</sup> There are also measures to promote the employment of affected patients within the public and private sectors. Current care provisions do not include psychological support for haemoglobinopathies patients; only the Thalassaemia Centre of Nicosia offers this service. Patients and healthcare professionals have highlighted that the emotional toll of dealing with the disease is not negligible and, therefore, all the dedicated centres should offer psychological support to patients. Experts also consider that access to innovative treatments should be made available to all patients across the country.

<sup>112</sup> Prevention of thalassaemia in Cyprus, M.A. Angastiniotis, M.G. Hadjiminias, *Lancet*, 14 February 1981, 1(8216):369–71, <http://www.ncbi.nlm.nih.gov/pubmed/6109998> | <sup>113</sup> Θεραπευτικός Οδηγός για την Κλινική Αντιμετώπιση της Θαλασσαιμίας, Therapeutic Guide for the Clinical Care of Thalassaemia, Thalassaemia International Federation, 2010. [http://www.thalassaemia.org.cy/pdf/Guidelines\\_in\\_Greek\\_2010.pdf](http://www.thalassaemia.org.cy/pdf/Guidelines_in_Greek_2010.pdf) | <sup>114</sup> Μια νέα απειλητική ασθένεια (A New Threatening disease), Νατάσα Ευριπίδου (N. Eyrpidou), η Σημερινή, 13 January 2012, <http://www.sigmalive.com/simerini/news/health/455080> Πρώτο Παγκύπριο Εργαστήριο Δρεπανοκυταρικής Αναιμίας (First Pancyriot Workshop on Sickle Cell Disease), H. Kathimerini, 12 January 2012, <http://www.kathimerini.com.cy/index.php?pageaction=k&modid=1&artid=73136> | <sup>115</sup> Annual Report 2010, Cyprus Ministry of Health, [http://www.moh.gov.cy/moh/moh.nsf/EEBCAF0CDB3C0C4FC22577BB0026941E/\\$file/Ετήσια%20Εκθεση%202010.pdf](http://www.moh.gov.cy/moh/moh.nsf/EEBCAF0CDB3C0C4FC22577BB0026941E/$file/Ετήσια%20Εκθεση%202010.pdf) | <sup>116</sup> How thalassaemia was controlled in Cyprus, op. cit.



## Country Factsheet: Cyprus

### POLICY OUTLOOK

#### EXPECTED POLICY DEVELOPMENTS AND TIMELINE

Stakeholders agree that prevention, care and management of haemoglobin disorders are comprehensive and effective in Cyprus. Further improvement will be linked to the future development of new therapies.

There is ongoing consideration of the establishment of screening programmes at schools.

#### INTERVIEWEES' SUGGESTED POLICY ACTIONS

- Access to psychological support
- Access to innovative treatment across the country
- Introduction of haemoglobinopathies education in the national curriculums of paediatricians, haematologists, endocrinologists, hepatologists, cardiologists and other specialisations
- Ensuring preconception and prenatal screening, as well as awareness amongst interracial couples

**“WIDELY IMPLEMENTED SCREENING PROGRAMMES ACCOMPANIED BY STRONG EDUCATION CAMPAIGNS, AND THE ACTIVE INVOLVEMENT OF THE BROADER COMMUNITY, HAVE BEEN INSTRUMENTAL IN REDUCING THE NUMBER OF NEW THALASSAEMIA PATIENTS TO ALMOST ZERO IN CYPRUS.**

**INCREASED COORDINATION AND A COMPREHENSIVE POLICY FRAMEWORK AT EU LEVEL IS NEEDED IN ORDER TO ADDRESS HAEMOGLOBIN DISORDERS EFFECTIVELY ACROSS THE EU.”**

Androulla Eleftheriou, Thalassaemia International Federation

## Country Factsheet: France

### KEY FACTS AND FIGURES

According to a recent issue of the epidemiological bulletin of the Institut de Veille Sanitaire<sup>117</sup> (InVS, French Institute of Health Surveillance) Sickle Cell Disease (SCD) is the most common haemoglobinopathy and the most frequent disease detected by neonatal screening in France.<sup>118</sup>

Healthcare professionals estimate the number of SCD patients in France to be over 10,000 cases.<sup>119</sup> A recent update from the *Registre Français des Thalassémies* (French Register for Thalassaemia) recorded approximately 500 thalassaemia patients in the French territory.<sup>120 121</sup> The percentage of the total population with SCD is, therefore, approximately 0.023% and with thalassaemia is 0.0005%.<sup>122</sup>

National newborn screening results<sup>123</sup> indicate that every year, 409 babies are born with SCD in France<sup>124</sup> and the French Overseas Departments and Territories (including the West Indies) in 2010.<sup>125</sup> On average, nine conceptions are affected by thalassaemia every year.<sup>126</sup>

Experts believe that there is a direct link between the number of new patients diagnosed every year and migration flows in France.<sup>127</sup> The highest concentration of haemoglobinopathy patients are in big cities (about 50% of SCD patients in France live in the Paris area)<sup>126 128</sup> and French Overseas Territories, where 10 to 12% of the population are SCD carriers<sup>126 129</sup> (equivalent to one in 280 births). In France, neonatal screening has revealed that the majority of the affected population originate from Sub-Saharan Africa, North Africa, Overseas Territories and the Mediterranean basin.<sup>130</sup> While the number of thalassaemia patients has been fairly stable for the past 18 years, the number of SCD patients is expected to rise substantially, up to 20,000 patients in 2020.<sup>131</sup>

Estimates indicate that average life expectancy amongst patients with thalassaemia major, appropriately transfused and chelated, is now higher than 40–50 years of age.<sup>132 133</sup> Median age at death of patients with SCD is 36 years, which is twice as high as 20 years ago, as a result of better care.<sup>134</sup> According to the current literature, the number of annual deaths of a result of SCD has increased from an average of 13 per year in 1981 to 1985, to 40 per year in 2001 to 2005, with 70% of these deaths occurring in the Paris area and French Overseas Territories. An increase in the number of hospitalizations due to SCD has also been observed; however, it is not clear whether this is due to a growing number of SCD sufferers or simply an increased level of care of existing SCD patients.<sup>135</sup>

Over the period 2004–2009, an average of 7,355 patients with SCD were cared for every year. During the same period, the number of hospitalisations amongst SCD patients increased by 3.5% every year.<sup>136</sup> Severe anaemias, serious infections and cardiovascular accidents are considered the most common complications of SCD.<sup>137</sup>

Since 2006, the national Reference Centre for Thalassaemia has gathered data on thalassaemia patients and care across the country for the French Thalassaemia Registry, which is supported by the French national plan on rare diseases and the Ministry of Health.<sup>138</sup> There is no comprehensive registry for SCD.<sup>139</sup>

Data on the costs of haemoglobinopathies in France are not available. Furthermore, disaggregated data on migrants' cultural and religious background and/or country of origin are not available.

<sup>117</sup> <http://www.invs.sante.fr/> | <sup>118</sup> Plan national maladies rares 2011–2014, French Ministries of Economy Finance and Industry, Research and Education, Employment and Health, 2011. [http://www.orpha.net/actor/Orphanews/2011/doc/Plan\\_national\\_maladies\\_rares.pdf](http://www.orpha.net/actor/Orphanews/2011/doc/Plan_national_maladies_rares.pdf) | <sup>119</sup> Montalembert | <sup>120</sup> Thuret I, Badens C. Bêta thalassémie majeure quelle est la situation en France. *La Revue du Praticien*, 20 October 2009 | <sup>121</sup> Thuret I. Personal communication, French Society of Hematology, 2013 | <sup>122</sup> Léta thalassémie majeure quelle est la situation en France, op. cit. | <sup>123</sup> La drépanocytose en France : des données épidémiologiques pour améliorer la prise en charge, Institut de veille sanitaire, *Bulletin Épidémiologique*, 3 July 2012. <http://www.invs.sante.fr/Publications-et-outils/BEH-Bulletin-epidemiologique-hebdomadaire/Derniers-numeros-et-archives/Archives/2012/BEH-n-27-28-2012/> | <sup>124</sup> La drépanocytose, Orphanet, March 2011. <http://www.orpha.net/data/patho/Pub/fr/Drepanocytose-FRfrPub125v01.pdf> | <sup>125</sup> La drépanocytose en France : des données épidémiologiques pour améliorer la prise en charge, Institut de veille sanitaire, *Bulletin épidémiologique*, 3 July 2012, <http://www.invs.sante.fr/Publications-et-outils/BEH-Bulletin-epidemiologique-hebdomadaire/Derniers-numeros-et-archives/Archives/2012/BEH-n-27-28-2012/> | <sup>126</sup> Bêta thalassémie majeure quelle est la situation en France, I. Thuret, C. Badens, *La revue du praticien*, 20 October 2009d | <sup>127</sup> Bardakdjian J, Wajcman H. Épidémiologie de la drépanocytose. *La Revue du Praticien* 2004;54:1531–1533 | <sup>128</sup> La drépanocytose, une chance pour la France? 2007. [http://www.youtube.com/watch?v=aBJAEd7f6Ns&feature=player\\_embedded](http://www.youtube.com/watch?v=aBJAEd7f6Ns&feature=player_embedded) | <sup>129</sup> Education thérapeutique pour les enfants drépanocytaires, op. cit. | <sup>130</sup> Thuret I, et al. Neonatal screening for sickle cell disease in France: evaluation of the selective process. *J Clin Pathol* 2010;63(6):548–551. | <sup>131</sup> Elion J. Sickle Cell Disease in France and international collaboration, 2007. [http://www.cehmob.org.br/simposio/imagens/download/palestras/ELION\\_Jacques.pdf](http://www.cehmob.org.br/simposio/imagens/download/palestras/ELION_Jacques.pdf) | <sup>132</sup> Bêta thalassémie majeure quelle est la situation en France, op. cit. | <sup>133</sup> Thuret I, et al. Complications and treatment of patients with  $\beta$ -thalassaemia in France: results of the National Registry. *Haematologica* 2010;95(5):724–729

## Country Factsheet: France

### POLICY FOCUS

### PREVENTION AND SCREENING

Thalassaemia and SCD are considered rare diseases in France, even though the frequency of SCD is higher than five out of 10,000 in certain areas.

The first national plan on rare diseases was adopted in 2005 with the aim of “ensuring equity in the access to diagnosis, treatment and provision of care”. The plan also raised awareness of rare diseases amongst patients, health professionals and the general public, and has established the basis for comprehensive screening, diagnosis and improved care across the country.<sup>140</sup> The second plan was developed for the period 2011–2014, following consultation with relevant patient groups and healthcare professionals.<sup>141</sup> The plan focuses particularly on SCD patients, due to the significant challenges of the disorder, and builds on the previous plan’s measures to improve knowledge on rare diseases, cooperation amongst the relevant stakeholders, and follow-up care and support for people with disabilities. The plan emphasises the need to implement nationwide protocols for healthcare professionals on diagnosis and care, and to improve patient education and access to healthcare for patients in the Overseas Territories by enhancing eHealth solutions and adequate patient disease self-management.

The French health plan for Overseas Territories<sup>142</sup> includes a set of measures to address haemoglobin disorders, namely the establishment of data collection and analysis protocols for thalassaemia and SCD, awareness activities and screening of all newborns. Contacted patients, however, are deeply disappointed that most of the described measures have not yet been implemented.

The Haute Autorité de Santé (HAS, French Health Authority) has adopted nationwide protocols for healthcare professionals on the diagnosis and care of thalassaemia major and intermedia,<sup>143</sup> and SCD in children and teenagers<sup>144</sup> as well as adults.<sup>145</sup>

The protocols include guidelines on screening, diagnosis and information for patients, long-term care and treatment, prevention and genetic counselling, as well as the prevention and care of complications. A particular focus on the prevention and care of complications in children with SCD, and the need to educate children, parents and teachers.<sup>146</sup>

As described previously, neonatal screening in France targets the at-risk population (i.e. babies whose parents originate from Africa, French Indies, South Europe, Asia, Middle West, North America and Brazil).<sup>147</sup> In the West Indies-Guyana region, SCD screening is carried out on the entire population. Patients point out that screening should be extended to every child in all regions, in consideration of the high representation of ethnic groups across the country. The diagnosis of thalassaemia major is usually made during the first month of life, when the child shows signs and symptoms of anaemia,<sup>148</sup> or in the neonatal period when the family is already known to be at risk.

Prenatal diagnosis of thalassaemia or SCD is proposed to couples considered ‘at risk’ (i.e. if both parents are carriers and, occasionally, if there is uncertainty of the father’s status).<sup>149 150</sup> Experts note that prenatal screening is generally accepted by couples at risk for a

<sup>134</sup> La drépanocytose en France : des données épidémiologiques pour améliorer la prise en charge, Institut de veille sanitaire, Bulletin épidémiologique, 3 July 2012, <http://www.invs.sante.fr/Publications-et-outils/BEH-Bulletin-epidemiologique-hebdomadaire/Derniers-numeros-et-archives/Archives/2012/BEH-n-27-28-2012/> | <sup>135</sup> Suzan F, Paty A-C. Etudes descriptive de la mortalité et des hospitalisations liées à la drépanocytose en France. Bulletin épidémiologique hebdomadaire, 3 July 2012. [http://www.invs.sante.fr/content/download/40267/186491/version/2/file/beh\\_27\\_28\\_2012.pdf](http://www.invs.sante.fr/content/download/40267/186491/version/2/file/beh_27_28_2012.pdf) | <sup>136</sup> Maladies rares: Mortalité liée à la drépanocytose, Institut de veille sanitaire, 2 December 2010. [http://www.invs.sante.fr/surveillance/maladies\\_rares/mortalite\\_drepanocytose.htm#5](http://www.invs.sante.fr/surveillance/maladies_rares/mortalite_drepanocytose.htm#5) | <sup>137</sup> Maladies rares : Mortalité liée à la drépanocytose, Institut de veille sanitaire, op. cit. | <sup>138</sup> Les Thalassémies, French Ministry of Health, 14 April 2009. <http://www.sante.gouv.fr/les-thalassemies.html> | <sup>139</sup> Tchernia G, et al. [A center in Paris for screening and counselling sickle cell patients and carriers]. Bull Acad Natl Med 2008;192(7):1349–1359 | <sup>140</sup> Plan national maladies rares 2011–2014, op. cit. | <sup>141</sup> Plan national maladies rares 2011–2014, op. cit. | <sup>142</sup> Plan Santé Outre-Mer 2009, French Ministry of Health, July 2009. [http://www.sante.gouv.fr/IMG/pdf/Plan\\_sante\\_outre-mer-2.pdf](http://www.sante.gouv.fr/IMG/pdf/Plan_sante_outre-mer-2.pdf) | <sup>143</sup> Syndromes thalassémiques majeurs et intermédiaires: Protocole national de diagnostic et de soins pour une maladie rare, Haute Autorité de Santé, July 2006. [http://www.has-sante.fr/portail/upload/docs/application/pdf/2008-07/pnds\\_thalassemies\\_final\\_web.pdf](http://www.has-sante.fr/portail/upload/docs/application/pdf/2008-07/pnds_thalassemies_final_web.pdf) | <sup>144</sup> Syndromes drépanocytaires majeurs de l'enfant et de l'adolescent: Protocole national de diagnostic et de soins pour une maladie rare", Haute Autorité de Santé, January 2010. [http://www.has-sante.fr/portail/upload/docs/application/pdf/2010-04/ald\\_10\\_pnds\\_drepano\\_enfant\\_web.pdf](http://www.has-sante.fr/portail/upload/docs/application/pdf/2010-04/ald_10_pnds_drepano_enfant_web.pdf) | <sup>145</sup> Syndromes drépanocytaires majeurs de l'adulte: Protocole national de diagnostic et de soins pour une maladie rare, by the Haute Autorité de Santé, January 2010. [http://www.has-sante.fr/portail/upload/docs/application/pdf/2010-04/ald\\_10\\_guide\\_drepano\\_adulte\\_web.pdf](http://www.has-sante.fr/portail/upload/docs/application/pdf/2010-04/ald_10_guide_drepano_adulte_web.pdf) | <sup>146</sup> La bêta-thalassémie, Orphanet, June 2008. <http://www.orpha.net/data/patho/Pub/fr/BetaThalassemie-FRfrPub51.pdf> PAI: Projet d'Accueil Individualisé, Ecole supérieure de l'éducation nationale, de l'enseignement supérieur et de la recherche (ESEN), March 2012, <http://www.esen.education.fr/?id=79&a=84&cHash=e800bc08c1> | <sup>147</sup> Informations et conseils: Drépanocytose, Direction Générale de la Santé, Ministère de la Santé et des Solidarités, December 2006. [http://www.sante.gouv.fr/IMG/pdf/drepanocytose\\_info.pdf](http://www.sante.gouv.fr/IMG/pdf/drepanocytose_info.pdf) | <sup>148</sup> La bêta-thalassémie, Orphanet, June 2008, <http://www.orpha.net/data/patho/Pub/fr/BetaThalassemie-FRfrPub51.pdf> | <sup>149</sup> La bêta-thalassémie, Orphanet, June 2008. <http://www.orpha.net/data/patho/Pub/fr/BetaThalassemie-FRfrPub51.pdf> | <sup>150</sup> Informations et conseils: Drépanocytose, Direction Générale de la Santé, Ministère de la Santé et des Solidarités, December 2006. [http://www.sante.gouv.fr/IMG/pdf/drepanocytose\\_info.pdf](http://www.sante.gouv.fr/IMG/pdf/drepanocytose_info.pdf)

## PREVENTION AND SCREENING

severe form of thalassaemia.<sup>151</sup> For SCD, in many cases, parents who know they are SCD carriers will choose not to undertake prenatal diagnosis or, once the prenatal diagnosis is performed, will decide to continue the pregnancy of an affected child.<sup>152</sup>

Information and awareness campaigns in France are generally driven by patient associations. There are no specific government campaigns on haemoglobinopathies. In 2006, the Haute Autorité de Santé published a set of factsheets and cards, mostly in French but with specific sections also in English, aimed at raising awareness of thalassaemia and SCD.<sup>153 154 155 156</sup> Healthcare professionals and patients agree on the need for increased awareness of haemoglobinopathies, health risks, sequelae and social impact among the general public and patients. They identify a lack of targeted measures to reach out to the at-risk population, and a lack of adequate funding. Individualised support in schools is in place for children with disabilities.<sup>157</sup> Patients believe, however, there is a need for intensified measures to raise awareness of haemoglobin disorders among education professionals in order to enhance education, prevent stigmatisation and ensure adequate support from school staff.

Experts also stress the need to implement education measures for the at-risk population that address specific barriers to the access of quality information and care, such as language differences. Patients have also called on the government to ensure the implementation and strengthening of awareness measures foreseen under the Health Plan for the Overseas Territories. Promoting specialised training and education for general practitioners and nurses on haemoglobinopathies is also considered an important step for improving patient care.

## TREATMENT AND CARE

**Specialised thalassaemia and SCD care is provided in three reference centres and 14 expertise centres across the country.**

These centres also provide access to the latest screening techniques both for haemoglobin disorders and complications (e.g. transcranial Doppler scanning to prevent stroke in SCD children, and MRI imaging for iron overload in thalassaemia). The centres are responsible for diagnosis, therapeutic management, psychological and social support, developing and disseminating care protocols, coordinating research and participating in epidemiological monitoring. They provide healthcare education and training activities, promoting information to patients and their families for adequate self-management of the disease, coordination of multidisciplinary teams and dialogue with patient groups.<sup>158 159 160</sup>

Patients have expressed concern over disparities in the level of quality of specialised care provided across the country. The need to travel to reference (or expertise) centres remains a significant challenge for some patients.

Care is provided free of charge and, according to healthcare experts, costs linked to travel to reference and expertise centres are also reimbursed. Healthcare professionals and patients stress the need to ensure broad patient access to all available innovative treatments, and also agree that increased financial support to research on genetic therapies is crucial in order to improve the prevention and care of haemoglobinopathies.

It is, however, noteworthy that the first thalassaemia patient to be successfully cured by gene therapy was French and was cared for in France.<sup>161</sup>

<sup>151</sup> Bêta thalassémie majeure quelle est la situation en France, op. cit. <sup>152</sup> La drépanocytose en France : des données épidémiologiques pour améliorer la prise en charge, Institut de veille sanitaire, Bulletin épidémiologique, 3 July 2012, <http://www.invs.sante.fr/Publications-et-outils/BEH-Bulletin-epidemiologique-hebdomadaire/Derniers-numeros-et-archives/Archives/2012/BEH-n-27-28-2012/> <sup>153</sup> Informations et conseils : Drépanocytose, Direction Générale de la Santé, Ministère de la Santé et des Solidarités, December 2006, [http://www.sante.gouv.fr/IMG/pdf/drepanocytose\\_info.pdfop.cit](http://www.sante.gouv.fr/IMG/pdf/drepanocytose_info.pdfop.cit). <sup>154</sup> Informations et conseils : Thalassémies, Direction Générale de la Santé, Ministère de la Santé et des Solidarités, January 2009, [http://www.sante.gouv.fr/IMG/pdf/DGS-Thalassemie\\_Conseils\\_BD.pdf](http://www.sante.gouv.fr/IMG/pdf/DGS-Thalassemie_Conseils_BD.pdf) <sup>155</sup> Emergency and Healthcare Card: Sickle cell disease, Direction Générale de la Santé, Ministère de la Santé et des Solidarités, December 2006. [http://www.sante.gouv.fr/IMG/pdf/drepanocytose\\_soins.pdf](http://www.sante.gouv.fr/IMG/pdf/drepanocytose_soins.pdf) <sup>156</sup> Emergency and Healthcare Card: Thalassémies, by the Direction Générale de la Santé, Ministère de la Santé et des Solidarités, January 2009. [http://www.sante.gouv.fr/IMG/pdf/DGS-Thalassemie\\_Soins\\_Specimen\\_BD.pdf](http://www.sante.gouv.fr/IMG/pdf/DGS-Thalassemie_Soins_Specimen_BD.pdf) <sup>157</sup> PAI: Projet d'Accueil Individualisé, Ecole supérieure de l'éducation nationale, de l'enseignement supérieur et de la recherche (ESEN), March 2012. <http://www.esen.education.fr/?id=79&a=84&cHash=e800bc08c1> <sup>158</sup> Circulaire DHOS/DGS/04/SD5D no 2006-479 du 9 novembre 2006 relative à l'appel à projets auprès des centres hospitaliers universitaires en vue de l'obtention du label de centre de référence pour une maladie ou un groupe de maladies rares, French Ministry of Health, 9 November 2006. <http://www.sante.gouv.fr/fichiers/bo/2006/06-12/a0120033.htm> <sup>159</sup> Centres de référence labellisés et centres de compétences désignés pour la prise en charge d'une maladie rare ou d'un groupe de maladies rares, Orphanet, Les Cahiers d'Orphanet, October 2010. [http://www.sante.gouv.fr/IMG/pdf/Liste\\_des\\_centres\\_de\\_compétences\\_et\\_de\\_references\\_par\\_groupe.pdf](http://www.sante.gouv.fr/IMG/pdf/Liste_des_centres_de_compétences_et_de_references_par_groupe.pdf) <sup>160</sup> Un centre de référence: c'est quoi?, French Ministry of Health, 17 February 2009. <http://www.sante.gouv.fr/un-centre-de-reference-c-est-quoi.html> <sup>161</sup> Cavazzana-Calvo M, et al. Transfusion independence and HMG2A activation after gene therapy of human  $\beta$ -thalassaemia. *Nature* 2010;467:318–322

## TREATMENT AND CARE

The Haute Autorité de Santé protocols of care<sup>162 163 164</sup> include guidance to healthcare professionals on the treatment and long-term care of thalassaemia and SCD, including the prevention and care of related complications. The guidelines for SCD include reference to psychological, cultural and ethnic aspects of the disorder, and note that special attention should be paid to the cultural background of patients, as in France this disorder affects mainly migrants or their children. Furthermore, the guidelines recommend that patients are referred to psychologists and, in some cases, interpreters/mediators or 'transcultural psychiatrists'. Thalassaemia guidelines, however, only recommend referral to a psychologist and include no reference to cultural competency, possibly because thalassaemia patients in France used to be mainly of indigenous or South European (Greek or Italian) origin.

According to patients, long distances and non-coverage of some transportation costs, a lack of preparedness by the healthcare system and shortage of specialists are the major barriers to improving patient access to quality care. They stress the need to implement targeted awareness campaigns focused on haemoglobinopathies, in order to address the barriers patients face in accessing quality information and care.

Psychological support should also be strengthened, in particular for teenagers, through improved education and training for medical staff, psychologists and social workers. Patients further highlight the need for targeted measures to prevent stigmatisation and facilitate labour integration of people with haemoglobin disorders.

<sup>162</sup> Syndromes drépanocytaires majeurs de l'enfant et de l'adolescent: Protocole national de diagnostic et de soins pour une maladie rare, Haute Autorité de Santé, January 2010, [http://www.has-sante.fr/portail/upload/docs/application/pdf/2010-04/ald\\_10\\_pnds\\_drepano\\_enfant\\_web.pdf](http://www.has-sante.fr/portail/upload/docs/application/pdf/2010-04/ald_10_pnds_drepano_enfant_web.pdf)

<sup>163</sup> Syndromes drépanocytaires majeurs de l'adulte : Protocole national de diagnostic et de soins pour une maladie rare, Haute Autorité de Santé, January 2010, [http://www.has-sante.fr/portail/upload/docs/application/pdf/2010-04/ald\\_10\\_guide\\_drepano\\_adulte\\_web.pdf](http://www.has-sante.fr/portail/upload/docs/application/pdf/2010-04/ald_10_guide_drepano_adulte_web.pdf)

<sup>164</sup> Syndromes thalassémiques majeurs et intermédiaires : Protocole national de diagnostic et de soins pour une maladie rare, Haute Autorité de Santé, July 2006, [http://www.has-sante.fr/portail/upload/docs/application/pdf/2008-07/pnds\\_thalasseemies\\_final\\_web.pdf](http://www.has-sante.fr/portail/upload/docs/application/pdf/2008-07/pnds_thalasseemies_final_web.pdf)

## POLICY OUTLOOK

## EXPECTED POLICY DEVELOPMENTS AND TIMELINE

In the context of the current rare diseases plan, discussions are ongoing on the establishment of universal SCD screening. This includes a pilot study in a selection of public and private hospitals in Ile-de-France and the creation of a dedicated expert group on screening.

## INTERVIEWEES' SUGGESTED POLICY ACTIONS

- Further development of measures and protocols to ensure consistent screening and care practices
- Specialised healthcare professional education and training across different specialisms
- Increased recruitment of specialised practitioners
- Increased access to screening and care equipment (e.g. echo Doppler), as well as treatment
- Increased financial support for research on gene therapies

**“CONTINUITY AND ADEQUATE FINANCIAL SUPPORT ARE CRUCIAL IN ORDER TO BUILD UPON THE PROGRESS ACHIEVED SO FAR.”**

Patricia Aguilar Martinez, Laboratoire d'hématologie biologique, CHU de Montpellier

## Country Factsheet: Germany

### KEY FACTS AND FIGURES



Estimates from 2012 suggest that approximately 400,000 people in Germany are carriers of haemoglobinopathy genes.<sup>165</sup> This results in an estimated prevalence rate of 0.5 to 1% of the overall population, and 5% of the immigrant population.<sup>166</sup>

Amid the changing, multiethnic population, there are 15 million people with an immigrant background.<sup>167</sup> More than 9 million of the immigrants now living in Germany come from countries considered to be of 'risk' for haemoglobinopathies.<sup>168</sup>

There is no comprehensive registry or precise information available on the number of carriers or patients with overt disease. Experts estimate that there are approximately 4,500 haemoglobinopathy patients being treated in Germany (1,500 thalassaemia patients and 3,000 sickle cell disease [SCD] patients).

Around 30 new patients are diagnosed with thalassaemia every year. A 2012 running trial on newborn screening for SCD in Berlin diagnosed seven newborns between January and June with this disease. Studies by the Ulm hospital analysed the national origin of affected patients in Germany and showed the following distribution: Turkey (40.0%), Italy and Greece (28.3%), Middle East and Africa (16.1%), Asia (6.6%), Germany (4.5%), and other southern and eastern Europe (4.5%).<sup>169</sup> In Germany there is no central registry for haemoglobinopathies, despite attempts from the Ministry of Education and Research to fund a number of data collection projects in the 1990s. The availability of data varies across the different regions since only four hospitals have a department dedicated to haemoglobinopathies (Ulm, Hamburg, Düsseldorf, Berlin).<sup>170</sup>

Amongst the main sequelae linked to SCD, experts highlight vasoocclusive crises and chronic organ insufficiency, chronic respiratory failure with pulmonary hypertension, possible iron overload and endocrinological failures for patients in a chronic transfusion programme.<sup>171</sup> The anaemia itself is considered as a 'minor' complication.<sup>172</sup> The main sequelae linked to thalassaemia include cardiovascular diseases, hypopigmentation of the skin, liver fibrosis or cirrhosis, diabetes mellitus, and growth disorders.<sup>173</sup>

There is no official data on the costs linked to haemoglobinopathies in Germany. However, healthcare professionals estimate that the cost of treatment and care of a haemoglobinopathies patient could be around €50,000 per patient, per year, or more when taking into account late diagnosis and co-morbidities associated to these disorders.

Experts have attempted to analyse changes in the epidemiology of haemoglobinopathies in Germany as a result of the immigration flows from African, Arab, and Asian countries in the last decades.<sup>174</sup> New flows are likely contributors to a drastic increase not just in the number, but also in the genetic and clinical heterogeneity of haemoglobin defects in Germany.<sup>175</sup> In the absence of precise data, estimations suggest that, in 1998, about 350 children and adults suffered from SCD (mainly in former West Germany);<sup>176</sup> the number of SCD patients increased more than eight-fold in 12 years, to 3,000 estimated patients in 2010.<sup>177</sup>

As a result, haemoglobinopathies are considered amongst the most common of rare diseases in Germany. Healthcare professionals note that SCD and thalassaemia patients account for the majority of patients in paediatric haematology units and require significant healthcare resources.

<sup>165</sup> Kohne E, Kleihauer E. Haemoglobinopathies in Germany—a longitudinal study over four decades. *Deutsches Ärzteblatt International* 2009;107(5):65–71 | <sup>166</sup> Kohne E. Haemoglobinopathies. *Clinical Manifestations, Diagnosis, and Treatment*. *Deutsches Ärzteblatt International* 2011;108(31–32):532–540 | <sup>167</sup> Bevölkerung nach detailliertem Migrationsstatus. Statistisches Bundesamt: Fachserie 1, Reihe 2,6: Ausgaben 2005, 2006, 2007 | <sup>168</sup> Kohne E, Kleihauer E. Haemoglobinopathies: A Longitudinal Study Over Four Decades. *Deutsches Ärzteblatt International*. 2010; 107(5):65–71 | <sup>169</sup> Haemoglobinopathies in Germany—a longitudinal study over four decades, op. cit. | <sup>170</sup> Haemoglobinopathies in Germany—a longitudinal study over four decades, op. cit. | <sup>171</sup> Dickerhoff R, Heimpel H. Sichelzellerkrankheiten, *Onkopedia*, June 2010. <http://www.dgho-onkopedia.de/onkopedia/leitlinien/sichelzellerkrankheiten> | <sup>172</sup> Kulosik A. Haemoglobinopathies are on the increase. *Deutsches Ärzteblatt International* 2010;107(5):63–64 | <sup>173</sup> Klose T, Putzker M. Thalassämie. *Blut als Heilmittel*. *Pharmazeutische Zeitung online*, 2003. [http://www.pharmazeutische-zeitung.de/index.php?id=medizin1\\_47\\_2003](http://www.pharmazeutische-zeitung.de/index.php?id=medizin1_47_2003) | <sup>174</sup> Haemoglobinopathies in Germany—a longitudinal study over four decades, op. cit., p. 69 | <sup>175</sup> Haemoglobinopathies in Germany—a longitudinal study over four decades, op. cit., p. 69 | <sup>176</sup> Dickerhoff R, et al. Sichelzellerkrankungen in Deutschland. *Verlaufsbeobachtungen über zehn Jahre*. *Deutsches Ärzteblatt* 1998;95:A-1675–1679 | <sup>177</sup> Haemoglobinopathies in Germany—a longitudinal study over four decades, op. cit., p. 70



## Country Factsheet: Germany

### POLICY FOCUS

### PREVENTION AND SCREENING

Migration and health issues are increasingly being addressed by policy-makers and public authorities at regional and federal level through targeted strategies.<sup>178</sup>

At the moment of drafting this report, the German government was preparing its national plan on rare diseases, expected by 2013, and had plans to establish rare disease centres of expertise across the country. However, none of these initiatives are expected to include any particular focus on haemoglobinopathies, as the German Ministry for Health wishes to take a horizontal policy approach rather than giving priority to any particular group of diseases (rare diseases are those listed in orphanet).<sup>179</sup> Nevertheless, healthcare professionals, namely haemoglobinopathy experts, hope for the establishment of centres of reference in the area of haemoglobinopathies.

The absence of policy focus and framework for haemoglobinopathies is reflected in the lack of structured dialogue with haemoglobinopathy stakeholders in the shaping of initiatives. Since March 2010, the *Nationales Aktionsbündnis für Menschen mit seltenen Erkrankungen* (NAMSE, National Coalition for People with Rare Diseases) has brought relevant patient groups together and is expected to provide input and issue recommendations on the future Rare Diseases Plan.<sup>180</sup> Migrant representatives, however, are not included in the Coalition.

Patients and healthcare professionals regret that there is low general awareness of thalassaemia and SCD; the health risks linked to haemoglobin disorders; and the risk of stigmatisation, social and labour exclusion of affected patients. Knowledge about these diseases amongst general practitioners is also considered generally poor.

There are no targeted education measures at schools aimed at preventing and raising awareness of haemoglobinopathies and their impact amongst the youngest populations.

There are no prevention and screening programmes for haemoglobin disorders in place in Germany, or prenatal or neonatal screening to detect thalassaemia and SCD carriers and patients.

A neonatal SCD screening pilot project was carried out in Berlin in 2012.

**“IN GERMANY IT IS NECESSARY TO FORM MULTIDISCIPLINARY CENTRES WHERE ALL HAEMOGLOBINOPATHIES PATIENTS, ACROSS ALL AGE GROUPS, CAN BE TREATED.”**

Holger Cario, University Hospital Ulm

<sup>178</sup> Presentation of best practices in the areas of migration and health, website of the German Government, 2007. [http://www.bundesregierung.de/Content/DE/Publikation/IB/gesundheit-und-integration.pdf?\\_\\_blob=publicationFile](http://www.bundesregierung.de/Content/DE/Publikation/IB/gesundheit-und-integration.pdf?__blob=publicationFile) <sup>179</sup> Alphabetical list of rare diseases in orphanet, 2013. [http://www.orpha.net/consor/cgi-bin/Disease\\_Search\\_List.php](http://www.orpha.net/consor/cgi-bin/Disease_Search_List.php) <sup>180</sup> National Action Alliance for people with rare diseases (Namse), 2013. <http://www.namse.de/startseite.html>

## Country Factsheet: Germany

### TREATMENT AND CARE

Patients with thalassaemia require regular transfusions for the rest of their life. They also need iron-elimination treatment and, in most cases, complex endocrinological management.

If a suitable donor can be found, haematopoietic stem-cell transplantation should be performed. Despite these disease manifestations, optimised treatment protocols have improved the prognosis of affected children, so that they can now survive into adulthood. As a result of optimised treatment and care, which has led to improved life expectancy, haemoglobin disorders are increasingly treated in adult internal medicine care.<sup>181</sup>

Care and treatment of haemoglobinopathy patients is ensured by specialised centres across the country (Ulm, Hamburg, Düsseldorf, Berlin). In the specialised centres, patients receive information about their disease and how it can be managed most effectively on a daily basis. Expert groups on haemoglobinopathies have developed guidelines for diagnosis and treatment of thalassaemia and SCD, which do not include any references to cultural competence.<sup>182</sup> Patients and healthcare professionals, have reported that the level of quality of the specialised care differs greatly among these units.

A number of patient groups from therapy areas including thalassaemia, SCD and chronic diseases play an active role in providing patient support.<sup>183 184 185</sup>

Ensuring a holistic approach to care and optimal management of the disease is one of the major concerns for both patients and healthcare professionals. Both stress the need to ensure appropriate transition between paediatric and adult treatment.

Interviewees stated that one of the main challenges for patients remains access to specialised physicians and hospitals/clinics. They consider that this is a result of not having enough fully dedicated centres for haemoglobinopathies, or a dedicated register. Appropriate follow-up care is a major concern too; patients say there is no entitlement to receive specialist advice regularly (e.g. once a year) in order to ensure proper follow-up and therapy continuation. Improvements in psychological and social support are also considered crucial steps in order to ensure quality care in Germany.

Experts recognise that healthcare systems are not well-prepared to deal with affected patients, causing barriers to optimal haemoglobinopathy prevention and management. Particular measures are needed to target population groups effectively, due to language, socio-economic, cultural, and religious factors. Experts stress that these factors should be taken into account when designing new models for the prevention and care of haemoglobinopathies patients in Germany.

<sup>181</sup> Haemoglobinopathies are on the Increase, A. Kulosik, Deutsches Ärzteblatt International 2010; 107(5): 63–64, p.63 | <sup>182</sup> Gesellschaft für Pädiatrische Onkologie und Hämatologie, Leitlinie Thalassämie, 2010. [http://www.awmf.org/uploads/tx\\_szleitlinien/025-0171\\_S1\\_Thalassaemie.pdf](http://www.awmf.org/uploads/tx_szleitlinien/025-0171_S1_Thalassaemie.pdf), Leitfaden für die Betreuung von Sichelzellerkrankungen, 2012. [http://www.haemoglobin.uni-bonn.de/sichelzelleitfaden2012\\_10.pdf](http://www.haemoglobin.uni-bonn.de/sichelzelleitfaden2012_10.pdf), and Sichelzell-Informationszentrum für Deutschland, Information über die Sichelzellerkrankung für Patienten 2010. [http://www.haemoglobin.uni-bonn.de/sichelzellerkrankung2010\\_7.html](http://www.haemoglobin.uni-bonn.de/sichelzellerkrankung2010_7.html) | <sup>183</sup> Interessensgemeinschaft Sichelzellerkrankheit und Thalassaemie e.V., 2013. <http://www.ist-ev.org/index.php> | <sup>184</sup> Help for thalassaemia without borders, 2013. <http://www.thogde.org/c5> | <sup>185</sup> Alliance for Chronic rare diseases (Achse), 2013. <http://www.achse-online.de> | <sup>186</sup> Bundesministerium für Gesundheit (BMG- Ministry for Health), Seltene Erkrankungen, 2013. <http://www.bmg.bund.de/praevention/gesundheitsgefahren/seltene-erkrankungen.html>

## Country Factsheet: Germany

### POLICY OUTLOOK

#### EXPECTED POLICY DEVELOPMENTS AND TIMELINE

The German rare disease plan is expected by 2013,<sup>186</sup> along with the creation of reference centres.

#### INTERVIEWEES' SUGGESTED POLICY ACTIONS

- Establishment of reference centres focused on haemoglobinopathies
- Creation of a dedicated registry to assess care needs and outcomes across the country
- Measures to ensure access to specialised care and comprehensive follow-up across the country
- Improving psychological and social support for affected patients

**“HAEMOGLOBINOPATHIES IN GERMANY SHOULD BE MORE FOCUSED ON BY DOCTORS AS THEY ARE NOT AS RARE AS THOUGHT. THEY NEED SPECIAL TREATMENT IN SPECIALISED CENTRES.”**

Regine Gosse, UKE Hamburg

**“HEALTH POLITICIANS AND DOCTORS SHOULD LOOK CLOSER AT THE ALARMING RISE OF HAEMOGLOBINOPATHIES IN GERMANY.”**

Jürgen Beith, Thalassämiehilfe ohne Grenzen  
(Help for thalassaemia patients without borders)

## Country Factsheet: Greece

### KEY FACTS AND FIGURES



### Haemoglobinopathies are the most common genetic disorders in Greece,<sup>187</sup> where they are considered endemic.

Haemoglobinopathy patients numbered 4,506 patients in 2012 (National Registry of Haemoglobinopathies) – approximately 0.04% of the Greek population.<sup>188</sup> New patients diagnosed every year/incidence: approximately six to seven new cases per year.<sup>189</sup> The carrier rate is estimated at 8% of the population for  $\beta$ -thalassaemia and 8% for  $\alpha$ -thalassaemia. Data from 2007 indicates between 2,500 and 3,000 registered  $\beta$ -thalassaemia patients.<sup>190</sup> The carrier rate is estimated at 1% of the population for sickle cell anaemia.<sup>191</sup> It is estimated that several hundreds of patients suffer from sickle cell disease (SCD).<sup>192</sup>

Available data indicate that between 1998 and 2003, there were 37 births of patients affected by haemoglobin disorders in Greece:<sup>193</sup> 19 were in families with immigrant origin (11 Albanians, four Africans, four Roma); in seven cases, prenatal screening by private laboratories failed to diagnose the diseases; in six cases from In Vitro Fertilization (IVF) the donor had not been examined; in three cases the parents decided to continue with the pregnancy despite the presence of a haemoglobin disorder; in two cases the doctor had not suggested the screening to identify whether the foetus or parents were carriers. Healthcare professionals believe that the existence of affected newborns is directly linked to: poor or lack of prenatal medical care due to financial reasons or low educational level; late diagnosis, due to unawareness of time limitations for prenatal diagnosis amongst obstetricians and couples at risk; and religious, social and ethical reasons.<sup>194</sup> Experts highlight that the number of newborns affected by haemoglobin disorders has significantly decreased in the past decades from approximately 120 per year in the late 1970s,

to approximately six in the early 1990s onwards.<sup>195</sup> The prevalence of thalassaemia carriers in Greece is considered high: 7–8% on average. In certain regions, such as Lesbos, Corfu and Karditsa, the rate is 15% or higher.<sup>196</sup> The number of  $\beta$ -thalassaemia carrier births is considered to be more than 100,000 yearly. 1–1.5% of Greeks are SCD carriers.<sup>197</sup> Data from 1969 estimated the overall incidence of  $\beta$ -thalassaemia trait as 7.7%. This was significantly higher in certain areas such as the Ionian islands (14%) and Rhodes (16–20%). Each year around 200 carrier children were born; in total 2,500 patients were suffering from  $\beta$ -thalassaemia, significantly fewer than today since most did not survive puberty.<sup>198</sup> SCD was restricted to a few areas, like Orchomenos (20%) and Chalikidiki (23%), where there was a high frequency of malaria in the past.<sup>199</sup>

The number of couples at risk of having a baby affected by a haemoglobin disorder is estimated to be between 800 and 1,000 every year,<sup>200</sup> which would result in 120–130 newborns suffering from a haemoglobinopathy per year if no preventative measures were undertaken.<sup>201</sup> The first National Registry for Haemoglobinopathies in Greece (NRHG) was established in 2009 by the Greek Society of Haematology. It gathers data from all patients affected by thalassaemia major (TM), thalassaemia intermedia (TI), “H” Haemoglobinopathy (HH) and SCD.<sup>202</sup>

There are no data on the estimated cost linked to haemoglobinopathies in Greece. Experts stress that life expectancy amongst haemoglobinopathy patients significantly improved after 2005, due to technological and medical developments. For example, due to MRI scanning, the number of deaths from heart failure is significantly lower. Linked cardiovascular and liver problems are considered the main cause of death amongst affected patients.<sup>203</sup>

<sup>187</sup> Voskaridou E, et al., on behalf of the Greek Haemoglobinopathies Study Group. A national registry of haemoglobinopathies in Greece: Deducted demographics, trends in mortality and affected births. *Ann Hematol* 2012;91(9):1451–1458 | <sup>188</sup> A national registry of haemoglobinopathies in Greece: Deducted demographics, trends in mortality and affected births, E. Voskaridou et al. on behalf of the Greek Haemoglobinopathies Study Group, *Ann Hematol*, 19 April 2012, <http://www.ncbi.nlm.nih.gov/pubmed/22526366> | <sup>189</sup> Management of Haemoglobin Disorders Report of Joint WHO-TIF Meeting, op. cit. | <sup>190</sup> Management of Haemoglobin Disorders Report of Joint WHO-TIF Meeting. | <sup>191</sup> Management of Haemoglobin Disorders Report of Joint WHO-TIF Meeting, Nicosia, Cyprus, 16–18 November 2007, [http://www.who.int/genomics/WHO-TIF\\_genetics\\_final.pdf](http://www.who.int/genomics/WHO-TIF_genetics_final.pdf) | <sup>192</sup> Loukopoulos D. Haemoglobinopathies in Greece: prevention programme over the past 35 years. *Indian J Med Res* 2011;134(4):572–576 | <sup>193</sup> Management of Haemoglobin Disorders Report of Joint WHO-TIF Meeting, op. cit. | <sup>194</sup> A national registry of haemoglobinopathies in Greece: Deducted demographics, trends in mortality and affected births, E. Voskaridou et al., on behalf of the Greek Haemoglobinopathies Study Group, *Ann Hematol*, 19 April 2012, <http://www.ncbi.nlm.nih.gov/pubmed/22526366> | <sup>195</sup> Components for setting up large thalassaemia prevention programs. The paradigm of Greece, D. Loukopoulos, Biomedical Research Foundation, Academy of Athens, Presentation in the 12th International Conference on Thalassaemia and the Hemoglobinopathies, Antalya 2011. | <sup>196</sup> 20 Χρόνια Ελληνική Ομοσπονδία Θαλασσαιμίας (20 years Greek Thalassaemia Federation) Album, Greek Thalassaemia Federation, 2010. [http://eotha.gr/files/LEFKOMA\\_EOTHA.pdf](http://eotha.gr/files/LEFKOMA_EOTHA.pdf) | <sup>197</sup> Components for setting up large thalassaemia prevention programs- The paradigm of Greece, op. cit. | <sup>198</sup> Matsaniotis N, Kattamis C. Thalassemsias, a social problem in Greece. *Ann Soc Belge Med Trop* 1969;49(3):223–230 | <sup>199</sup> Thalassemsias, a social problem in Greece, op. cit. | <sup>200</sup> 20 Χρόνια Ελληνική Ομοσπονδία Θαλασσαιμίας (20 years Greek Thalassaemia Federation) Album, Greek Thalassaemia Federation, 2010, [http://eotha.gr/files/LEFKOMA\\_EOTHA.pdf](http://eotha.gr/files/LEFKOMA_EOTHA.pdf) | <sup>201</sup> Haemoglobinopathies in Greece: prevention programme over the past 35 years, Dimitris Loukopoulos, *Indian J Med Res*; 134(4): 572–576, October 2011, <http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3237258/> | <sup>202</sup> A national registry of haemoglobinopathies in Greece: Deducted demographics, trends in mortality and affected births. | <sup>203</sup> A national registry of haemoglobinopathies in Greece: Deducted demographics, trends in mortality and affected births.

## Country Factsheet: Greece

### POLICY FOCUS

The main current focus of the Greek government in the field of health is to contain costs and ensure the overall sustainability of the healthcare system.

The 'Institutional Framework for Thalassaemia and Sickle Cell Disease Units',<sup>204</sup> adopted in 2007 by the Ministry of Health, recognised official 'reference centres' and set minimum standards for specialised staff according to the number of patients treated in each centre. The Thalassaemia Committee of the Central Board of Health has undertaken the development of obligatory unified guidelines for the operation of both public and private units. However, most thalassaemia units cooperate closely and the guidelines established by the Laiko Reference Centre are widely used. These include the analysis of quantitative and qualitative data as well as assessment of the overall care delivery.

Particular emphasis is placed on the continuous education of the medical, nursing and technology staff. Also emphasised are compliance with medical ethics and the application of new therapeutic protocols, which should be certified by the Thalassaemia Committee of the Central Health Board (KESY), an advisory body to the Ministry of Health. However, implementation of the other aspects of the overall framework has been poor, largely due to lack of sufficient funds, unwillingness on the part of certain stakeholders involved and ineffective controls.

In line with international classifications, haemoglobin disorders are considered rare diseases.<sup>205</sup> The Greek 'National Action Plan for Rare Diseases 2008–2012' (Εθνικό Σχέδιο Δράσης για τις Σπάνιες Παθήσεις 2008–2012), adopted by the Health Ministry, aims to provide a comprehensive, horizontal strategy for the holistic care of rare diseases. Key priorities include the Ministry's involvement in the collection of valid and reliable information through the dedicated registry of rare diseases, which was created independently by the Hellenic Haematology Association, in order to inform future policy development. This is considered crucial in order to continue improving the quality of healthcare delivery and the quality of life of patients.<sup>206</sup> The Rare Disease Plan does not include any specific focus on haemoglobin disorders.

The Special Committee for Thalassaemia of the Central Board of Health (Κεντρικό Συμβούλιο Υγείας-ΚΕΣΥ) is a dedicated advisory body that brings together healthcare professionals specialised in thalassaemia, patients and civil society representatives. This Committee is responsible for providing input and advice to the Ministry of Health on any new policy developments related to haemoglobinopathies. Healthcare professionals, however, regret that the actual consideration and implementation of its proposals largely depend on mobilisation and pressure from stakeholders such as patient associations.

<sup>204</sup> Θεσμικό Πλαίσιο για τις Μονάδες Μεσογειακής Αναμίας και Δρεπανοκυτταρικής Νόσου (Institutional Framework for Thalassaemia and Sickle Cell Disease Units), Φ.Ε.Κ. 2267, Ημ.Δημοσίευσης 28/11/2007, Νόμος Αριθμ. Υ4α/οικ149906 | <sup>205</sup> Εθνικό Σχέδιο Δράσης για τις Σπάνιες Παθήσεις 2008–2013 (National Plan for Rare Diseases 2008–2013), Υπουργείο Υγείας (Ministry of Health), 2008. <http://www.yyka.gov.gr/articles/health/domes-kai-drasesis-gia-thn-ygeia/ethnika-sxedia-drashs/95-ethnika-sxedia-drashs> | <sup>206</sup> Εθνικό Σχέδιο Δράσης για τις Σπάνιες Παθήσεις 2008–2013 (National Plan for Rare Diseases 2008–2013), Υπουργείο Υγείας (Ministry of Health), <http://www.yyka.gov.gr/articles/health/domes-kai-drasesis-gia-thn-ygeia/ethnika-sxedia-drashs/95-ethnika-sxedia-drashs>

**“GREECE HAS BEEN A LEADER COUNTRY IN RESEARCH AND PROVIDING HOLISTIC, QUALITY CARE AND PREVENTION OF HAEMOGLOBINOPATHIES.”**

Ersi Voskaridou, Director of the Centre for Thalassaemia at the Laiko General Hospital

## Country Factsheet: Greece

### PREVENTION AND SCREENING

A 'National Prevention Programme' for haemoglobinopathies has been in place since 1974 and sets out guidelines for the screening and diagnosis of haemoglobinopathies. The programme was first established as a pilot project by the academic and medical community. Since then, it has been integrated into the National Healthcare System,<sup>207</sup> and widely implemented across the country.

Key measures aim to: raise awareness of haemoglobinopathies amongst the general public; ensure adequate screening in dedicated units in central and peripheral public hospitals; and provide genetic counselling and patient information about available prenatal diagnosis to high-risk couples.<sup>208</sup> As part of this initiative, education campaigns on haemoglobinopathies in primary and secondary schools are also in place. The programme includes measures to: facilitate access to personalised genetic counselling; apply European Molecular Genetics Quality Network (EMQN) best practices of external quality control and quality management; and take into account increased needs due to the influx of high numbers of immigrants from high-prevalence countries.<sup>209</sup>

Screening mainly targets couples engaged to be married, as they are considered easier to reach out to and more receptive to relevant information about how to prevent haemoglobin disorders, compared with other groups (younger population) or settings (schools).<sup>210</sup> Prenatal and neonatal screening to detect the risk of thalassaemia and sickle cell anaemia is not mandatory, but is widely recommended by most doctors and free of charge for patients. In total, there are 15 carrier identification units in peripheral hospitals in areas with high frequency of the respective genes.<sup>211</sup> When necessary, screening is referred to the Central Unit in Athens for further

investigation and molecular studies.<sup>212</sup> As a consequence, the prevention unit at Laiko Hospital screens approximately 10,000 people every year. Healthcare professionals agree on the importance of ensuring quality controls and validation of screening results.<sup>213</sup>

Along with Cyprus, Italy (Sardinia) and the UK (London), Greece hosts one of the main European Prevention Centres for haemoglobinopathies. This centre, in Athens, runs the largest number of prenatal diagnoses in Europe.<sup>214</sup>

Patients are informed individually if they are identified as disease carriers. In situations when both parents are carriers, they are called to receive further information and genetic counselling, usually delivered by a social worker or other specialised professional.<sup>215</sup> In addition to medical factors, ethical, legal, social and family aspects are taken into account when providing advice to carriers. Prenatal diagnosis and pregnancy termination is generally accepted by couples at risk.<sup>216</sup>

Experts consider access to innovative screening techniques to be optimal. As an example, the Laikon Hospital provides DNA testing to diagnose thalassaemia in the very early stages of pregnancy to 500–600 pregnant mothers. Thalassaemia and SCD units in Greece participate in research protocols and regularly assess and introduce the application of new methods.<sup>217</sup> Healthcare professionals agree on the importance of improving prenatal diagnosis, however. The medical community is particularly encouraged by recent and ongoing research into prenatal diagnosis, through blood testing of the mother, rather than by amniocentesis.<sup>218</sup> In Vitro Fertilisation (IVF) pre-implantation diagnosis for thalassaemia is carried out by the Laboratory of Medical Genetics in the Children's Hospital 'Agia Sophia', and a number of private IVF centres.<sup>219</sup>

<sup>207</sup> Management of Haemoglobin Disorders Report of Joint WHO-TIF Meeting, op. cit. | <sup>208</sup> Management of Haemoglobin Disorders Report of Joint WHO-TIF Meeting, op. cit. | <sup>209</sup> Management of Haemoglobin Disorders Report of Joint WHO-TIF Meeting, op. cit. | <sup>210</sup> Haemoglobinopathies in Greece: prevention programme over the past 35 years, D. Loukopoulos, *Indian J Med Res*; 134(4): 572–576, October 2011, <http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3237258/> | <sup>211</sup> Haemoglobinopathies in Greece: prevention programme over the past 35 years, op. cit. | <sup>212</sup> Haemoglobinopathies in Greece: prevention programme over the past 35 years, op. cit. | <sup>213</sup> Haemoglobinopathies in Greece: prevention programme over the past 35 years, op. cit. | <sup>214</sup> 20 Χρόνια Ελληνική Ομοσπονδία Θαλασσαιμίας (20 years Greek Thalassaemia Federation) Album, Greek Thalassaemia Federation, 2010, [http://eotha.gr/files/LEFKOMA\\_EOETHA.pdf](http://eotha.gr/files/LEFKOMA_EOETHA.pdf) | <sup>215</sup> Laiko Hospital, [http://www.laiko.gr/index.php?option=com\\_content&view=article&id=81&Itemid=125](http://www.laiko.gr/index.php?option=com_content&view=article&id=81&Itemid=125) | <sup>216</sup> Haemoglobinopathies in Greece: prevention programme over the past 35 years, op. cit. | <sup>217</sup> Interview with E Voskaridou, Laiko Hospital, Centre for Thalassaemia and Haemoglobinopathies | <sup>218</sup> Μεσογειακή αναμία: Πλησιάζει η θεραπεία, *Vita magazine*, September 2008. <http://www.vita.gr/html/ent/248/ent.7248.asp> | <sup>219</sup> Μεσογειακή αναμία: Πλησιάζει η θεραπεία, op. cit.



## PREVENTION AND SCREENING

However, healthcare professionals in specialised public hospital units are significantly more experienced in detecting even rare haemoglobin variants, compared to private laboratories. Screening and diagnosis of health sequelae associated with haemoglobinopathies are also available. However, healthcare professionals and patients regret that differences in level of access exist across the country. For example, only a few units in Greece provide transcranial Doppler scanning for stroke prevention to children with SCD. For thalassaemia and non-transfusion-dependent thalassaemia patients (NTDT), MRI scanning is available to all patients, but is only carried out by limited units in Athens and Thessaloniki.

Information campaigns to raise awareness of haemoglobinopathies prevention and screening amongst the general population are regularly organised by patient and medical associations.<sup>220</sup> The media also frequently reports on haemoglobinopathies issues, although experts' involvement with the media has become limited due to their increased workload.

Thalassaemia is particularly addressed within educational programmes at school, in coordination with medical and patient associations. However, patients highlight that the outcomes and success of these initiatives varies and largely depends on the involvement and knowledge of the teachers.

Experts consider that lack of awareness and cultural and religious beliefs remain the main reasons for the high prevalence of new cases of babies born with haemoglobin disorders every year. Awareness of healthcare professionals also plays a key role in ensuring timely screening and diagnosis. The major challenge remains reaching out and conveying adequate information to migrant populations (e.g. Albanians, Pakistanis, Afghans and Africans). Experts believe that further efforts should be made in order to raise awareness amongst these target groups.

## TREATMENT AND CARE

Due to the traditionally high prevalence of haemoglobinopathies in Greece, experts consider that the country has achieved significantly major progress in ensuring quality care and treatment of affected patients, when compared to other European countries.

Due to modern technology and medical developments, reductions in mortality and improved life expectancy and quality of life of patients have been achieved.

In 1974, leading experts in the field of haemoglobin disorders from the Reference Centre of Thalassaemia in Laiko Hospital developed guidelines on the screening, diagnosis, care and treatment of haemoglobinopathies. These guidelines are intended for use by healthcare professionals, and although not compulsory or formally endorsed by the public authorities, the guidelines are widely accepted and implemented across the country. Experts consider that an effective holistic approach to haemoglobinopathies prevention and care is effectively implemented across Greece. Specialised medical units for thalassaemia and SCD exist in most major cities of the country - 38 units are operating in public hospitals, the largest being at the Agia Sofia Children's Hospital, treating approximately 700 patients.

Experts consider that research and the introduction of innovative therapies is a high priority for the specialised medical units, once approval has been given by the National Organisation for Medicines (Εθνικός Οργανισμός Φαρμάκων- ΕΟΦ). Research and successful results to manage and prevent iron overload among thalassaemia patients carried out by the Thalassaemia Transfusion Unit in Corinth<sup>221</sup> (Μονάδα Μεταγγίσεων Μεσογειακής Αναιμίας του Γενικού Νοσοκομείου Κορίνθου)<sup>222</sup> Bone marrow transplants are successfully carried out in specialised transplant centres. Experts expect that future developments in gene therapy will provide an efficient treatment for these diseases. The "G. Papanikolaou" hospital in Thessaloniki is currently carrying out promising research in this field,

<sup>220</sup> Hellenic Society of Haematology, 2013. <http://www.eae.gr/new2/index.asp> | <sup>221</sup> Thalassaemia Transfusion Unit of the General Hospital of Corinth, 2007. <http://www.hospkorinthos.gr/erga-aima.html> | <sup>222</sup> Μεσογειακή αναιμία: Πλησιάζει η θεραπεία, Vita magazine, September 2008, <http://www.vita.gr/html/ent/248/ent.7248.asp>



## TREATMENT AND CARE

in cooperation with the Department of Medical Genetics of the University of Washington, US.

Patients and professionals recognise the key role that patient groups have traditionally played in ensuring patient support and advocacy for improved policies and care for haemoglobinopathy patients. Patients strongly recognise that social perceptions and attitudes towards haemoglobinopathy patients have improved significantly. Whereas in the 1970s, patients faced major social exclusion, people have now become more aware of and knowledgeable about these disorders. Both patients and professionals agree that no particular problems of social exclusion and stigmatisation persist, except for possible sporadic cases in very small and isolated communities.

Specific policies are in place to facilitate the social, educational and professional inclusion of patients, such as access to tertiary education without sitting exams,<sup>223</sup> and measures to support recruitment of people with disabilities in the public and private sectors.<sup>224</sup> Along with free access to care, patients are entitled to additional financial support, tax benefits and full pension upon retirement after 15 years of work.<sup>225</sup> There is also commitment to integrate professionals with thalassaemia and SCD into the National Health System (ESY) hospitals. Care is free of charge for insured patients. Unemployed people and migrant patients are often uninsured, and therefore do not benefit from coverage and access to healthcare services, with the exception of asylum seekers. Experts note this makes it even more difficult to provide care and outreach to affected patients, and regret that the current economic situation has led to increasingly restricted resources, which are primarily allocated to insured patients.

There is general consensus that the present lack of resources has led to decreased capacity for delivering quality care in Greece. Staff shortages due to lack of recruitment to replace retired personnel; scarce or poor equipment; and waiting lists are amongst the major challenges and concerns identified by experts. Despite efforts from healthcare professionals to make the most cost-effective use of resources without jeopardising the level of quality care, they are deeply concerned that it is increasingly difficult to treat patients in a timely manner. Patients stress that while complications and secondary conditions were adequately dealt with in the past,

the quality of care has dramatically deteriorated in the past years due to the lack of experienced and specialised medical staff.

Patient associations also believe that the current economic situation has had a tremendous negative psychological impact on patients, making care, treatment compliance and their general quality of life even more difficult. The lack of adequate funding for measures to support education, social support and labour integration of affected patients is of great concern to patient associations. Patients call on the government to ensure greater cooperation and consensus between the Ministry of Health, healthcare professionals and patient associations, in order to address the socioeconomic situation and impact of healthcare delivery.

Centralised guidelines and protocols on testing and treatment that are aimed at all public and private hospital and laboratories involved in the screening, treatment and care of haemoglobinopathies are also needed. Other required measures include: improved healthcare quality management (currently under development by the Thalassaemia Committee of the Central Health Board); extending financial support to cover all equipment needed; oral iron chelation therapy; and travel costs incurred to receive blood transfusions. Another major challenge faced by healthcare professionals and patients is the shortage of blood reserves, partly due to a lack of a blood donation culture. Blood needs are met mainly through mandatory blood donations by patients' relatives, while volunteer donors provide only 40% of all donations. More than 30 blood units per patient are needed every year for blood transfusions.

Insufficient financial support for research programmes is also highlighted by experts as a major challenge in Greece. While it is possible to access EU funding, research efforts remain outside the formal scope of work of professionals and largely depend on individual commitment and dedication after normal working hours. There are no specific educational/training programmes for healthcare professionals. The 'Institutional Framework for Thalassaemia and Sickle cell disease Units',<sup>226</sup> published in 2007, encourages the continuous education of professionals through seminars and conferences, which are regularly organised by healthcare and medical associations, and the broader scientific community.

<sup>223</sup> Law.3794/2009 (ΦΕΚ 156 Α/4-9-2009) | <sup>224</sup> Law 2643/98 (ΦΕΚ 220 τ. Α/28-09-1998) | <sup>225</sup> 20 Χρόνια Ελληνική Ομοσπονδία Θαλασσαιμίας (20 years Greek Thalassaemia Federation) Album, op. cit. | <sup>226</sup> Θεσμικό Πλαίσιο για τις Μονάδες Μεσογειακής Αναμίας και Δρεπανοκυτταρικής Νόσου (Institutional Framework for Thalassaemia and Sickle-Cell Disease Units), Φ.Ε.Κ. 2267, Ημ.Δημοσίευσης 28/11/2007, Νόμος Αριθμ. Υ4α/οικ149906

## Country Factsheet: Greece

### POLICY OUTLOOK

The Ministry of Health and the Central Board of Health (KESY) have stated their intention to become involved in the full implementation and continuation of the National Registry for Haemoglobinopathies, in cooperation with the Hellenic Society of Haematology (HSH) that established and carried out the project in 2012.

The Thalassaemia Committee of the Central Board of Health has undertaken the development of unified obligatory guidelines for the operation of units, both public and private. However, most thalassaemia units are in close cooperation and the guidelines established by the Laiko Reference Centre are widely used.

### INTERVIEWEES' SUGGESTED POLICY ACTIONS

Regarding prevention and screening, experts involved in policy development stress the need to ensure the proper operation of the private laboratories and IVF centres.

It is important that a formal licensing procedure and adequate controls are in place. Furthermore, medical and paramedical staff should benefit from the experience and accumulated knowledge of healthcare professionals in public hospital units, such as the Laiko reference centre, through a structured training system.

Regarding the quality of treatment and care, the full implementation of the Institutional Framework for Thalassaemia and Sickle cell disease Units, established in 2007, is an absolute priority for healthcare professionals, patients and policy-makers.

**“THE CURRENT ECONOMIC AND COST CUTS ARE RESULTING IN A CONTINUED DETERIORATION OF THE QUALITY AND ACCESS TO CARE FOR PATIENTS.”**

Vasilis Dimos, President of the Greek Thalassaemia Federation (EOTHA Patient organisation)

## Country Factsheet: Italy

### KEY FACTS AND FIGURES

### POLICY FOCUS

Haemoglobinopathy disorders are endemic in Italy. Estimates say there are over 7,000 people affected by haemoglobinopathies in Italy.<sup>227</sup>

In addition, about 3 million people are estimated to be symptom-free carriers at risk of transmitting haemoglobinopathies to their children.<sup>228</sup> However, experts believe that the number of patients may be higher, since these estimations do not take account of immigration flows from North Africa.

Reliable, comparable data across the country is not available. Since 2001, Italian regions are responsible for gathering data on rare diseases, including thalassaemia and SCD. The Centro Nazionale Malattie Rare (CNMR – National Centre for Rare Diseases) is responsible for compiling and analysing this data.<sup>229</sup>

Sicily is the only region that currently has a comprehensive registry on haemoglobinopathies. Healthcare professionals regret that the steps taken so far to enhance data gathering and evaluation have not led to tangible progress.

**“WE DESPERATELY NEED CONCERTED ACTION TO GUARANTEE PATIENTS AFFECTED BY HAEMOGLOBINOPATHIES GOOD QUALITY CARE AND MUCH LONGER LIFE EXPECTANCY, ALLOWING PATIENTS TO REACH OLD AGE.”**

Tommasina Iorno, President ATDL

Italy's healthcare system is jointly managed by national, regional and local authorities.

While the Ministry of Health<sup>230</sup> is responsible for setting the overall healthcare policy priorities and objectives, regional governments are responsible for their implementation, including issuing treatment guidelines and guaranteeing the delivery of healthcare services in local health units.

The current policy priorities in the area of health include: ensuring sustainable and effective use of the existing resources; improving quality of care through enhanced prevention; monitoring systems; coordination between primary and hospital care units as well as clinical networks; and ensuring a holistic approach to patient-centred care.<sup>231</sup> However, there is no reference to health equity in the current policy priorities.

Haemoglobinopathies are considered rare diseases in Italy. The *Istituto Superiore di Sanità* (ISS – Italian National Institute of Health, the main technical and scientific body within Italian National Health Service) is currently developing the first National Plan on Rare Diseases, with the involvement of medical associations, patient groups and other relevant stakeholders. The plan is expected to set up a comprehensive framework for the prevention, diagnosis and long-term care of rare diseases and to improve coordination amongst the different involved regional bodies. A public consultation involving patients' associations, National Network of Rare Diseases' units and scientific societies was concluded on 8 February 2013, to gather suggestions aiming at improving the plan's first draft.<sup>232</sup>

<sup>227</sup> Talassemia, l'8 maggio la Giornata Internazionale. In Italia la migrazione fa aumentare i casi (media article), I. Ciancaleoni Bartoli, Osservatorio Malattie Rare, April 2011. <http://www.osservatoriomalattierare.it/talassemia/655-talassemia-l8-maggio-la-giornata-internazionale-in-italia-la-migrazione-fa-aumentare-i-casi>

<sup>228</sup> Marchi Foundation, July 2013. <http://www.fondazioneemarchi.it/la-thalassemia/>

<sup>229</sup> National Centre for Rare Diseases (ISS), <http://www.iss.it/cnmr/>

<sup>230</sup> Italian Ministry of Health, 2013. <http://www.salute.gov.it/>

<sup>231</sup> Ministry of Health, Priorities 2012. <http://www.salute.gov.it/ministero/sezTrasparenzaDettaglio.jsp?label=trasp&notSez=trasp1&idP=934&id=939>

<sup>232</sup> Italian Ministry of Health. National Plan on Rare Diseases, 2012. <https://www.salute.gov.it/malattieRare/paginaInternaMalattieRare.jsp?id=3296&menu=piano&lingua=italiano>

## POLICY FOCUS

The plan is not expected to establish specific measures on particular diseases but to refer to rare diseases in general.

There are no national targeted policies addressing haemoglobinopathies and migrant health. Experts and patients regret the lack of policy developments required in response to the continuous rise in the number of haemoglobinopathy patients, partially influenced by increasing migration flows from North Africa. Experts also note that poor policy focus on these disorders is linked to the economic and social status of patients, who are often neglected in the development of health policies.

Different patient groups across the country have recently gathered together to draw political attention to haemoglobinopathies and raise general awareness.<sup>233</sup>

Experts stress the need for improved concerted action and educational programmes on haemoglobinopathies aimed at patients as well as doctors.

## PREVENTION AND SCREENING

Since the 1970s, a few Italian regions have implemented neonatal and prenatal screening and diagnosis programmes on  $\beta$ -thalassaemia (e.g. Sicily, Sardinia and the Po Delta)<sup>234</sup>

However, there is no screening programme in most of the regions or at national level.<sup>235</sup> Healthcare professionals believe that access to prenatal and neonatal screening generally varies significantly as a result of differing policies and resources available across the different regions. Healthcare professionals consider that in regions where access to innovative prenatal screening (e.g. celocentesis, embryonal selection)<sup>236</sup> is widely available, the number of births of patients suffering haemoglobin disorders has decreased, and the number of patients suffering thalassaemia major has been reduced to almost zero.<sup>237</sup> Information on the specific situation of migrants is not available.

Screening programmes are available in some regions where haemoglobinopathies are endemic. In Sicily, for example, screening is free for all women aged between 14 and 50, pregnant women, and family members of patients.<sup>238</sup>

Prenatal genetic counselling is widely available to haemoglobinopathy patients.<sup>239</sup> Yet healthcare professionals believe that this practice remains inconsistent amongst migrant patients, especially those from North Africa. Healthcare professionals consider the inconsistency to be mainly as a consequence of cultural attitudes and perceptions against prenatal screening and pregnancy termination.

<sup>233</sup> Talassemia, le associazioni di pazienti insieme per la nascita di United Onlus (media article), Osservatorio Malattie Rare, May 2012. <http://www.osservatoriomalattierare.it/talassemia/2145-talassemia-le-associazioni-di-pazienti-insieme-per-la-nascita-di-united-onlus>; Costituita united federazione delle associazioni per la talassemia e la drepanocitosi, Hemoex, May 2012. [http://www.hemoex.it/main/index.php?id\\_pag=47&id\\_inf\\_articolo=113](http://www.hemoex.it/main/index.php?id_pag=47&id_inf_articolo=113) <sup>234</sup> The first antenatal diagnosis of Beta-Thalassaemia in Europe was conducted in Cagliari (Sardinia) in 1977. <http://www.aslcagliari.it/index.php?xsl=7&s=692&v=2&c=2492>; [http://veprints.unica.it/683/1/PhD\\_Roggero\\_Simona.pdf](http://veprints.unica.it/683/1/PhD_Roggero_Simona.pdf). <sup>235</sup> Allargamento degli screening e delle esenzioni, le Regioni fanno da sole (media article), Osservatorio Malattie Rare, March 2011. <http://www.osservatoriomalattierare.it/screening-neonatale/574-allargamento-degli-screening-e-delle-esenzioni-le-regioni-fanno-da-sole->; Talassemia, l'8 maggio la Giornata Internazionale. In Italia la migrazione fa aumentare i casi, Osservatorio Malattie Rare, April 2011. <http://www.osservatoriomalattierare.it/talassemia/655-talassemia-8-maggio-la-giornata-internazionale-in-italia-la-migrazione-fa-aumentare-i-casi>; Immigrazione ed emoglobinopatie: o ci attiviamo subito per prevenirle o... è già emergenza, A. Amato, Previassme Foundation, <http://www.fondazionepreviasme.it/allegati/Amato.pdf> <sup>236</sup> Contro la talassemia, un nuovo test prenatale, Umberto Veronesi Foundation, April 2011. <http://www.fondazioneveronesi.it/la-tua-salute/ginecologia/contro-la-talassemia-un-nuovo-test-prenatale/589>; La Celocentesi, eccellenza della ricerca siciliana ed esempio della collaborazione pubblico privato (media article), I. Ciancaleoni Bartoli, Osservatorio Malattie Rare, December 2011. <http://www.osservatoriomalattierare.it/talassemia/311-la-celocentesi-eccellenza-della-ricerca-siciliana-ed-esempio-della-collaborazione-pubblico-privato> <sup>237</sup> Immigrazione ed emoglobinopatie: o ci attiviamo subito per prevenirle o... è già emergenza, op. cit. <sup>238</sup> Italian Society for Thalassaemias and Haemoglobinopathies, [http://www.site-italia.org/legislazione\\_vigente.php?134122434941.45](http://www.site-italia.org/legislazione_vigente.php?134122434941.45) <sup>239</sup> Diagnosi prenatale, il prof Novelli su Repubblica: 'Prioritaria la consulenza genetica sui genitori' (media article), Osservatorio Malattie Rare, January 2011. <http://www.osservatoriomalattierare.it/attualita/408-diagnosi-prenatale-il-prof-novelli-su-repubblica-prioritaria-la-consulenza-genetica-sui-genitori>

## PREVENTION AND SCREENING

Experts believe, however, that it could also be due to healthcare professionals' poor competences when treating patients with different cultural backgrounds.

Screening and prevention of complications is carried out as part of follow-up care to patients affected by haemoglobinopathies. Individual experts in regions (such as Lombardia) have developed voluntary guidelines on the diagnosis of health sequelae targeted to healthcare professionals.<sup>240</sup> Cardiac complications are considered the most common cause of death among people affected by thalassaemia major.<sup>241</sup> Innovative tools such as T2\* magnetic resonance are currently used in eight university hospitals across the country in order to improve diagnosis of these linked problems. Hepatitis C amongst young thalassaemia major transfusion-dependent patients, hepatic complications and severe renal and lung deficiencies are also sequelae of major concerns for patients.<sup>242</sup>

General awareness of haemoglobinopathies and patient education are considered to be low, and vary across regions. Healthcare professionals stress the importance of patient education on the health sequelae linked to haemoglobinopathies and the role that doctors play in this. Experts also note there is a need to increase awareness of migrant health related issues, in order to improve diagnosis and care in medical emergency services and peripheral hospitals. They express regret that only a few education and information campaigns focused on haemoglobinopathies have been conducted in middle and high schools in the last decades. Such campaigns would help improve prevention of these diseases and fight patient stigmatisation.

Patient groups and professionals have called on the government to implement specific educational programmes on haemoglobin disorders at school, and also as part of medical professional training. Scientific societies (such as Società Italiana Talassemie ed Emoglobinopatie, SITE – Italian Society for Thalassaemias and Haemoglobinopathies)<sup>243</sup>, foundations, patient groups, and healthcare professionals regularly run awareness activities targeted at the medical community and the general public.<sup>244</sup>

Recent studies suggested existing preventive programmes for severe haemoglobinopathies should adapt to changes in population ethnicities, and that screening for haemoglobinopathies at school age is an efficient strategy.<sup>245</sup> Healthcare professionals underline the importance of implementing primary prevention, based upon better information, diagnostics and counselling.<sup>246</sup>

The Italian Lazio region organises universal screening at school level. The *Centro Studi Microcitemia Roma* (CSMR), which provides care to people with haemoglobinopathies and promotes studies and research, has developed prevention, diagnostic and educational programmes targeting immigrants as well as secondary school students and young adults. CSMR curricular and extra-curricular programmes aim to guarantee equal access to prevention and medical care, especially considering the epidemiologically changing context of the Lazio region, due to high migratory fluxes.

<sup>240</sup> Thalassaemia Guidelines, G. Fiorelli, C. Vullo, Associazione Talassemici e Drepanocitici Lombardi (ATDL), 1999. [http://www.google.be/url?sa=t&rct=j&q=&esrc=s&source=web&cd=1&ved=0CFwQFJA&url=http%3A%2F%2Fwww.osservatoriomalattierare.it%2Fdocumenti%2Fdoc\\_download%2F3-linee-guida-talassemia&ei=AyYAUkm3MKrX0QXf\\_aizBw&usq=AFQjCNE71pM4-QT59Nh-J-D9xLMpxXRwA&sig2=zs7wEtQZ2x32V0wjrdRFvw](http://www.google.be/url?sa=t&rct=j&q=&esrc=s&source=web&cd=1&ved=0CFwQFJA&url=http%3A%2F%2Fwww.osservatoriomalattierare.it%2Fdocumenti%2Fdoc_download%2F3-linee-guida-talassemia&ei=AyYAUkm3MKrX0QXf_aizBw&usq=AFQjCNE71pM4-QT59Nh-J-D9xLMpxXRwA&sig2=zs7wEtQZ2x32V0wjrdRFvw) <sup>241</sup> Talassemia: arriva il T2 star multislice, il sistema non invasivo per quantificare l'accumulo di ferro nel cuore (media article), I. Giancaleoni Bartoli, Osservatorio Malattie Rare, October 2010. <http://www.osservatoriomalattierare.it/talassemia/204-talassemia-arriva-il-t2-star-multislice-il-sistema-non-invasivo-per-quantificare-l-accumulo-di-ferro-nel-cuore>; <sup>242</sup> For a summary of Thalassaemia complications see: [http://www.fondazionegiambone.it/?page\\_id=342](http://www.fondazionegiambone.it/?page_id=342) <sup>243</sup> Italian Society for Thalassaemias and Haemoglobinopathies, <http://www.site-italia.org/> <sup>244</sup> Confronto medici e pazienti sulla talassemia nella regione veneto (media article), Hemoex, November 2011. [http://www.hemoex.it/main/index.php?id\\_pag=47&id\\_inf\\_articolo=78](http://www.hemoex.it/main/index.php?id_pag=47&id_inf_articolo=78); La cura della talassemia e drepanocitosi al s. orsola malpighi di bologna, Hemoex, January 2012. [http://www.hemoex.it/main/index.php?id\\_pag=47&id\\_inf\\_articolo=89](http://www.hemoex.it/main/index.php?id_pag=47&id_inf_articolo=89); Piera Cutino Foundation <http://www.pieracutino.it/>; Giambone Foundation, <http://www.fondazionegiambone.it/>; ATDL, <http://www.atdl.it/old/index.html> <sup>245</sup> Prevention strategies for severe hemoglobinopathies in endemic and nonendemic immigration countries: the Latium example, A. Amato, P. Grisanti, M. Lerone, D. Ponzini, P. Di Biagio, M.P.Cappabianca and P.C. Giordano, Prenatal Diagnosis 2009; 29:1171-1174 <sup>246</sup> Screening and genetic diagnosis of hemoglobinopathies in Southern and Northern Europe: two examples, A. Amato and P.C.Giordano, Medit J of Hemat Infect Dis 2009, 1(1): E2009007

## Country Factsheet: Italy

### TREATMENT AND CARE

Access to healthcare services is free of charge for patients.<sup>247</sup> In each region there is at least one specific referral centre for haemoglobin disorders.<sup>248</sup> Once referral centres diagnose patients, they are usually referred to hospital units for follow-up care and treatment.

Healthcare professionals regret that teenagers and young adults are not systematically taken care of by transition centres in order to address particular needs and conditions that arise at these life stages. Often care is provided by paediatricians.<sup>249</sup>

There are no existing national guidelines or protocols for care and treatment of haemoglobinopathies for doctors and nursing staff. Professionals consider guidelines a necessary step to improve care and mortality rates amongst young patients. Also, poor knowledge about complications often leads to mis- or under-diagnosis. Italian patient groups have published guidelines on prevention and treatment of different haemoglobinopathies.<sup>250</sup> Healthcare professionals welcome the work done by the European Network for Rare and Congenital Anaemias (ENERCA) that aims to develop guidelines for care and treatment of patients and raise general awareness on rare and congenital anaemias.<sup>251</sup> In some regions, such as Sicily<sup>252</sup>, Lombardia, Emilia-Romagna and Veneto, there are special training programmes for healthcare professionals.

There are relevant ongoing research projects in the area of gene therapy.<sup>253</sup> Experts highlight

the need for increased financial support for research, in particular taking into consideration that these are the most common rare diseases.

Patients and professionals believe that the current organisation of care is leading to wide disparities in access to quality care for patients across the different regions. The lack of specialised doctors and nurses as well as limited financial resources are also seen as major challenges. Some regions, such as Sicily, have developed specific policies to guarantee consistent quality care for haemoglobinopathies.<sup>254</sup>

Support for haemoglobinopathy patients is organised at regional level and therefore varies widely across the country. At national level, transfusion dependent haemoglobinopathy patients are entitled to a monthly allowance.<sup>255</sup> Eligible patients, however, only include those older than 35 and who have contributed to the social security system for at least 10 years. Patients who are recognised as having the status of 'registered disabled' are entitled to the financial and social support measures. There are no disaggregated data available on migrants. If migrants are permanent residents they are entitled to financial and social support.

Exceptionally, some Italian regions have implemented targeted social support measures for haemoglobinopathy patients.<sup>256</sup> Healthcare professionals recognise that these patients need increased targeted support, and they generally face difficulties in accessing other general support measures for people with disabilities.

<sup>247</sup> DECRETO 18 maggio 2001, n. 279: Regolamento di istituzione della rete nazionale delle malattie rare e di esenzione dalla partecipazione al costo delle relative prestazioni sanitarie, ai sensi dell'articolo 5, comma 1, lettera b), del decreto legislativo 29 aprile 1998, n. 124. (GU n. 160 del 12-7-2001- Suppl. Ordinario n.180/L), <http://www.iss.it/binary/geme/cont/DM279-2001.1234897572.pdf>; <http://www.salute.gov.it/malattieRare/paginaInternaMalattieRare.jsp?id=707&menu=rete&lingua=italiano> <sup>248</sup> Reference centres tend to conglomerate under their responsibility as well as conduct screening and diagnosis for a number of rare diseases. <sup>249</sup> Treatment of patients with Thalassaemia at Policlinico di Modena, [http://www.hemoex.it/main/index.php?id\\_pag=47&id\\_inf\\_articolo=110](http://www.hemoex.it/main/index.php?id_pag=47&id_inf_articolo=110) <sup>250</sup> The Geneva Foundation for Medical Education and Research have gathered a series of links to these guidelines, [http://www.gfmer.ch/Guidelines/Anemia\\_emoglobinopatie\\_it/Anemia\\_emoglobinopatie\\_mt.htm](http://www.gfmer.ch/Guidelines/Anemia_emoglobinopatie_it/Anemia_emoglobinopatie_mt.htm); ATDL guidelines on prevention and treatment of thalassaemia, <http://www.google.com/url?sa=t&rct=j&q=&esrc=s&source=web&cd=1&ved=OCG0QFjAA&url=http%3A%2F%2Fwww.osservatoriomalattierare.it%2Fdocumenti%2F3-linee-guida-talassemia&ei=7wQhUOKjPMb80QXqjoDQCg&usq=AFQjCNE71pM4-QT59NhJ-D9xlLMpxXRwA&sig2=hV9PivqlyYfV6LMYjXePg> <sup>251</sup> ENERCA, <http://www.enerca.org/> <sup>252</sup> Fondazione cutino e cemi insieme per la lotta alla talassemia nel mediterraneo (media article), I. Ciancaleoni Bartoli, Osservatorio Malattie Rare, May 2011. <http://www.osservatoriomalattierare.it/talassemia/661--fondazione-cutino-e-cemi-insieme-per-la-lotta-alla-talassemia-nel-mediterraneo> <sup>253</sup> Talassemia, avanti verso la terapia genica, Telethon Foundation, July 2010. <http://www.telethon.it/news-video/news/talassemia-avanti-verso-terapia-genica-0>; <http://www.telethon.it/ricerca-progetti/malattie-trattate/talassemie>; Ricerca: accordo tra molmed e telethon per la terapia genica di sei malattie rare (media article), Osservatorio Malattie Rare, March 2011. <http://www.osservatoriomalattierare.it/telethon/575-ricerca-accordo-tra-molmed-e-telethon-per-la-terapia-genica-di-sei-malattie-rare> <sup>254</sup> Talassemia, a palermo parte il progetto ospedale a domicilio, I. Vacca, Osservatorio Malattie Rare, March 2011. <http://www.osservatoriomalattierare.it/talassemia/1883-talassemia-a-palermo-parte-il-progetto-ospedale-a-domicilio> <sup>255</sup> Disposizioni per la formazione del bilancio annuale e pluriennale dello Stato (legge finanziaria 2002), Italian Parliament, 2001. <http://www.camera.it/parlam/leggi/01448l.html> <sup>256</sup> Italian Society for Thalassaemias and Haemoglobinopathies, [http://www.site-italia.org/legislazione\\_vigente.php?134122434941.45](http://www.site-italia.org/legislazione_vigente.php?134122434941.45)



## Country Factsheet: Italy

### POLICY OUTLOOK

#### EXPECTED POLICY DEVELOPMENTS AND TIMELINE

The *Istituto Superiore di Sanità* (ISS - Italian National Institute of Health) is currently developing the first National Plan on Rare Diseases. A first draft of the plan was published in October 2012, however as it currently stands (from the consultation) no measures specifically targeting migrants are envisaged.

The plan is expected to be adopted by the end of 2013.<sup>257</sup>

#### INTERVIEWEES' SUGGESTED POLICY ACTIONS

- Improved coordination of regional referral centres on haemoglobinopathies
- Nationwide policies and guidelines specifically addressing screening, diagnosis, care and support for haemoglobinopathies patients
- Increased funding for research on rare anaemias
- Specific educational programmes for medical professionals including doctors and nurses, on diagnosis and care of haemoglobin disorders and secondary complications

**“HAEMOGLOBINOPATHIES ARE NOT ONLY RARE DISEASES. THEY ARE EMERGING DISEASES. POLICY MAKERS NEED TO DEDICATE MUCH MORE ATTENTION AND INTERVENTIONS TO THESE ANAEMIAS, IN ORDER TO IMPROVE PATIENTS’ QUALITY OF LIFE AND ENSURE EARLY DIAGNOSIS. RESEARCH IS KEY IN THIS PROCESS.”**

Lucia De Franceschi, University of Verona

<sup>257</sup> Italian Ministry of Health. National Plan on Rare Diseases (2012): <https://www.salute.gov.it/malattieRare/paginaInternaMalattieRare.jsp?id=3296&menu=piano&lingua=italiano>

## Country Factsheet: The Netherlands

### KEY FACTS AND FIGURES

According to the Erasmus Medical Centre, the Netherlands has 1,000 patients with haemoglobinopathies. Every year 60 children are born with a haemoglobin disorder.

Data from the specialised government agency *Rijksinstituut voor Volksgezondheid en Milieu* (RIVM) states that between five and ten children are born every year in the Netherlands with  $\beta$ -thalassaemia major,<sup>258</sup> and between 20 and 40 children are diagnosed with  $\alpha$ -thalassaemia.<sup>259</sup> Sickle cell disease (SCD) affects 40 to 60 newborns every year.<sup>260</sup>

Patient groups estimate that the total number of patients affected by SCD is approximately 750 (50% of them are estimated to be younger than 15 years). The number of thalassaemia patients is estimated to amount to 250, of whom, 60% are children.<sup>267</sup> Experts, however, consider that migrant patients are not included in the current estimates, and suggest that there are around 50 new affected patients amongst this population every year.

Experts attribute the national increase in the number of haemoglobinopathy patients in the last years to increasing immigration flows from Western Africa, Surinam and the Dutch Antilles. The increase is particularly due to migrants who arrive in the Netherlands during adulthood, as they are not traced through the current neonatal screening programmes. High birth rates among these migrant populations are also considered an important factor in explaining the increase in the number of patients.

Experts have noted that the population at risk of SCD increased from 0.5 to 1.7 million people over the last two decades in the Netherlands,<sup>262</sup> a trend that is likely to continue according to patient organisations. The patient group Organisation for Sickle Cell Anemia Relief (OSCAR) estimates that between 2005 and 2015 more than 100 children will be born with thalassaemia.<sup>263</sup> There are no data on the life expectancy of affected patients, which is generally estimated between 45 and 55 years old.

In the Netherlands, there is no central registry for haemoglobinopathy patients. Medical centres in Rotterdam, Amsterdam and The Hague have their own databases.

The neonatal screening programme testing for SCD was launched on 1 January 2007. Before this, only a few patients were followed from birth. Therefore, there are currently no reliable death rates for the Dutch SCD population.<sup>264</sup>

Although the *Centrum Volksgezondheid Toekomst Verkenningen* (cVTV - Centre for Public Health Forecasting) of the RIVM provides public information on the costs linked to illnesses,<sup>265</sup> there is no precise data available related to haemoglobinopathies.

<sup>258</sup> Beta-thalassaemie: Informatie in het kader van de neonatale hieprikscreening, RIVM, 2012. [www.rivm.nl/dsresource?type=pdf&objectid=rivmp:75736&versionid=&subjectname=](http://www.rivm.nl/dsresource?type=pdf&objectid=rivmp:75736&versionid=&subjectname=) <sup>259</sup> Alfa-thalassaemie: Informatie in het kader van de neonatale hieprikscreening, RIVM, 2012. [www.rivm.nl/dsresource?type=pdf&objectid=rivmp:75735&versionid=&subjectname=](http://www.rivm.nl/dsresource?type=pdf&objectid=rivmp:75735&versionid=&subjectname=) <sup>260</sup> Sikkkelcelziekte (SZ): Informatie in het kader van de neonatale hieprikscreening, RIVM, 2012. <http://www.rivm.nl/dsresource?objectid=rivmp:75734&type=org&disposition=inline> <sup>261</sup> Stichting OSCAR Nederland, 'Diagnose', <http://www.oscarnederland.nl/node/140> <sup>262</sup> The advisory report 'Neonatal screening' from the Health Council of The Netherlands, P.A. Bolhuis, G.C. Page-Christiaens, *Ned Tijdschr Geneesk*, 2005, Dec 17;149(51):2857-60 - as referred to in PhD thesis by X.W. van den Tweel (2009), 'Measuring complications of sickle cell disease', University of Amsterdam (UVA), p. 29. <http://dare.uva.nl/document/134112> <sup>263</sup> Stichting OSCAR Nederland, Thalassaemie, <http://www.oscarnederland.nl/node/57> <sup>264</sup> Measuring complications of sickle cell disease (PhD thesis), X.W. van den Tweel, 2009, University of Amsterdam (UVA), p. 11, <http://dare.uva.nl/document/134111> <sup>265</sup> Cost of Illness in the Netherlands, RIVM, <http://www.kostenvanziekten.nl/systeem/service-menu-rechts/homepage-engels/>

## Country Factsheet: The Netherlands

RELEVANT POLICY  
INSTITUTIONS

POLICY FOCUS

The Ministry of Health does not shape disease-specific policies, but is responsible for general guidance aimed at ensuring that healthcare is affordable, of high quality and accessible for every individual.

The national working party for haemoglobinopathy practitioners (*Landelijke Werkgroep Hemoglobinopathie Behandelaren*) defines guidelines, protocols and indicators for SCD and thalassaemia.

It cooperates with the specialised Dutch government agency *Rijksinstituut voor Volksgezondheid en Milieu* (RIVM) – the National Institute for Public Health and the Environment. It also cooperates with the *Nederlandse Organisatie voor Toegepast Natuurwetenschappelijk Onderzoek* (TNO – Netherlands Organization for Applied Scientific Research) and *Nederlandse Organisatie voor Gezondheidsonderzoek en Zorginnovatie* (ZonMw – Netherlands Organisation for Health Research and Development) to shape policy and take various initiatives.

The TNO is an organisation that collects and analyses data derived from the neonatal screening programme whereas the ZonMw finances healthcare research and stimulates the use of developed knowledge so as to improve care and health.

In the Netherlands, haemoglobinopathies are defined as rare diseases.<sup>266</sup>

On 29 February 2012, following the 2009 Recommendation by the EU Council of Health Ministers,<sup>267</sup> the Dutch Minister of Health sent a letter to the House of Representatives updating them on the Dutch strategy for rare diseases. The attachment to the letter enters into more detail about the strategy for the upcoming years. The Netherlands has thus already fulfilled its obligation to set out a strategy for rare diseases. The aim of the government is that this strategy and the policies derived from it should improve the situation of patients with rare diseases.<sup>268</sup>

Towards the end of 2011, a dedicated online resource was set up to systematically collect information about healthcare, research, education and the availability of treatment for people affected by a rare disease. The information will then be used to create a Dutch National Plan on Rare Diseases.<sup>269</sup> The aim of the government is to create a plan that is specific, measurable, acceptable, realistic and time-bound.<sup>270</sup> An updated draft plan,<sup>271</sup> dated January 2013, foresees long-term actions for those people who are either already suffering from rare diseases or may become a patient in the future (personally or in their immediate family environment). There is no specific focus or reference to migrants or haemoglobin disorders in particular.

Patients and experts stress the value of and need for greater cooperation and exchange of positive practices amongst European countries. Experts also note that relevant policies should be shaped by cultural and societal factors present in the population groups at risk.

<sup>266</sup> Wat is een zeldzame ziekte?, Stichting Zeldzame Ziekten Fonds, <http://www.zzf.nl/wat.php> | <sup>267</sup> Council Recommendation of 8 June 2009 on an action in the field of rare diseases, Official Journal of the European Union, 8 June 2009, <http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:C:2009:151:0007:0010:EN:PDF> | <sup>268</sup> Letter from Minister E. Schippers of Health, Welfare and Sports to the Chair of the House of Representatives about the Dutch Strategy for Rare Diseases. The Hague, dated 29 February 2012. | <sup>269</sup> Nationaal Plan Zeldzame Ziekten, Raamwerk Nationaal Plan in wording, <http://www.npzz.nl/>. | <sup>270</sup> Nationaal Plan Zeldzame Ziekten, Raamwerk Nationaal Plan in wording, op. cit. | <sup>271</sup> Concept Nationaal Plan Zeldzame Ziekten, [http://www.npzz.nl/wp-content/uploads/2012/08/2e-versie\\_29\\_01-20131.pdf](http://www.npzz.nl/wp-content/uploads/2012/08/2e-versie_29_01-20131.pdf)

## Country Factsheet: The Netherlands

### PREVENTION AND SCREENING

The Netherlands is considered to be a leading country in the implementation of broad neonatal screening programmes.

However, there is growing consensus amongst experts that a greater focus should be put on prevention and preconception screening of haemoglobinopathies.<sup>272</sup> This would require greater cooperation from health insurers.<sup>273</sup> Since 2007, both SCD and thalassaemias ( $\beta$  and  $\alpha$ ) are part of the broader neonatal screening programme for hereditary diseases, developed by the National Institute for Public Health and the Environment (RIVM) (*Hielprijk voor pasgeborenen*<sup>274</sup> – heel prick test for newborn babies).<sup>275</sup> Screening is voluntary and free of charge for patients.<sup>276</sup> The *Centrum voor Bevolkingsonderzoek* (CvB – Centre for Population Research), which operates under RIVM, is responsible for the overall coordination of the programme.<sup>277</sup> 99.8% of all parents get their children screened.<sup>278</sup> Experts and public authorities believe the neonatal screening programme is providing excellent results and could be shared as a positive experience with other countries.

Since 2011, *NEOnatale Registratie Afwijkende Hielprikscreening* (NEORAH – Neonatal Registry for Atypical Heel Prick Screening),<sup>279</sup> run by RIVM, gathers data (name, date of birth, address and result of the screening) on newly diagnosed children with a view to strengthening coordination between screening, care, follow up and research efforts.<sup>280</sup>

The neonatal screening programme is funded via the Dutch medical expenses act (Algemene Wet Bijzondere Ziektekosten, AWBZ). In 2012, the costs of the programme amounted to EUR 15 million.

Referrals for further diagnosis and treatment are paid by health insurers. A study to estimate the prevalence of children with SCD in the Netherlands concluded that the number of children with SCD is much higher than previously estimated, and that the majority of these children seem not to be examined regularly by a paediatrician. Children born abroad (27% of new cases) do not benefit from neonatal screening and are at high risk of life-threatening complications before SCD is diagnosed. As this introduces disparities in healthcare, healthcare professionals believe the introduction of adequate measures should be considered.<sup>281</sup>

In the Netherlands, preconception screening for SCD,  $\beta$ -thalassaemia and  $\alpha$ -thalassaemia is available for the population considered at risk of being carriers (e.g. those who have a family member suffering from a haemoglobinopathy condition, originate from Africa and/or Asia, and/or from southern European countries).<sup>282</sup> However, it is not covered by the general healthcare system. Screening is not provided systematically to patients at risk, who often have to request it.<sup>283</sup> One initiative that is currently being discussed is wider and standardised screening for haemoglobinopathies amongst parents from at-risk groups. However, it is not currently clear which groups would be considered 'at risk' as part of this initiative.

Since 2007, doctors increasingly inform pregnant women of the possibility of prenatal screening.<sup>284</sup> Experts stress, however, that there are still challenges linked to poor awareness amongst a number of healthcare professionals, as well as professionals' personal beliefs and views.<sup>285 286</sup> Awareness of prenatal screening amongst the population at risk of being carriers is considered to be higher than amongst the general public.<sup>287</sup>

<sup>272</sup> Neonatale screening op sikkelcelziekte, K.J.J. Wierenga, *Ned Tijdschr Geneesk*, 1997, 141:184–7, <http://www.ntvg.nl/publicatie/neonatale-screening-op-sikkelcelziekte/volledig> <sup>273</sup> Neonatale screening op sikkelcelziekte, op. cit. <sup>274</sup> Hielprijk, RIVM, <http://www.rivm.nl/Onderwerpen/Onderwerpen/H/Hielprijk> <sup>275</sup> The heel prick test for newborn babies, General information for parents, RIVM, 2011 <http://www.rivm.nl/dsresource?objectid=rivmp:75162&type=org&disposition=inline> <sup>276</sup> Veelgestelde vragen over de hielprijk, RIVM, [http://www.rivm.nl/Onderwerpen/Onderwerpen/H/Hielprijk/Veelgestelde\\_vragen\\_over\\_de\\_hielprijk](http://www.rivm.nl/Onderwerpen/Onderwerpen/H/Hielprijk/Veelgestelde_vragen_over_de_hielprijk) <sup>277</sup> Leren van de burenen. Beleid publieke gezondheid internationaal bezien: roken, alcohol, overgewicht, depressie, gezondheidsachterstanden, jeugd, screening, E.A. van der Wilk, J.M. Melse, J.M. den Broeder, P.W. Achterberg (eindred.), RIVM, rapportnr.270051010. Houten: Bohn Stafleu Van Loghum, 2007, p.17. <http://www.rivm.nl/bibliotheek/rapporten/270051010.pdf> <sup>278</sup> Leren van de burenen. Beleid publieke gezondheid internationaal bezien: roken, alcohol, overgewicht, depressie, gezondheidsachterstanden, jeugd, screening, op. cit., p.230–232. <sup>279</sup> NEORAH (NEOnatale Registratie Afwijkende Hielprikscreening), RIVM, <http://www.zorggegevens.nl/preventie/neonatale-registratie-afwijkende-hielprikscreening/> <sup>280</sup> NEORAH (NEOnatale Registratie Afwijkende Hielprikscreening), op. cit. <sup>281</sup> One-third of the new paediatric patients with sickle cell disease in The Netherlands are immigrants and do not benefit from neonatal screening, M. Peters, K. Fijnvandraat, X. W. van den Tweel, et al, *ADC* online, 2010. <sup>282</sup> Breder screenen op hemoglobopathie, M.C. Cornel, dr. S. Detmar, dr. A.M.C. Plass, D. Moerman, prof. dr. A.J. Waarlo, M. De Kinderen, dr. P.C. Giordano, *November 2009*, nr. 45, vol.3. <http://medischcontactartsennet.nl/Nieuws-26/archief-6/Tijdschriftartikel/68006/Breder-screenen-op-hemoglobopathie.htm> <sup>283</sup> OSCAR, <http://www.oscarnederland.nl/node/38> <sup>284</sup> Leren van de burenen. Beleid publieke gezondheid internationaal bezien: roken, alcohol, overgewicht, depressie, gezondheidsachterstanden, jeugd, screening, op. cit., p.17. <sup>285</sup> Leren van de burenen. Beleid publieke gezondheid internationaal bezien: roken, alcohol, overgewicht, depressie, gezondheidsachterstanden, jeugd, screening, op. cit., p.237. <sup>286</sup> 'Sikkelcelziekte met huisartsenprotocol aanpakken', 26 February 2010 <http://www.mw.nl/caribiana/article/sikkelcelziekte-met-huisartsenprotocol-aanpakken> <sup>287</sup> Breder screenen op hemoglobopathie, op. cit.

## Country Factsheet: The Netherlands

### TREATMENT AND CARE

Specialised haematologists are responsible for patient care and the provision of information to parents of children diagnosed with a haemoglobin disorder. Paediatricians are often in charge of the follow up care, in coordination with haematologists.

Around 50% of patients in the Netherlands receive care at the main two or three medical centres. Experts express concerns, however, that specialised centres are not always involved with the care and treatment of patients. This results in an increased number of complications and deaths which could be prevented. The National guidelines for the treatment of SCD and thalassaemia, written and published by the National Working Party for Haemoglobinopathy practitioners,<sup>288</sup> include no reference to cultural competency or sensitivity to different ethnicities, religions or cultural backgrounds.

Follow up care during the first years of life also involves screening of linked health complications, such as heart, liver and kidney conditions, and assessment of eventual need for bone marrow transplantation.<sup>289</sup> Prevention of infections and pain crisis is also a focus of the treatment of SCD.<sup>290</sup> Experts believe that the impact of migration flows and the linked increase in the number of haemoglobinopathy patients is posing additional challenges to the healthcare system.<sup>291 292</sup> They highlight the need to provide care through multidisciplinary teams, focused on the prevention, screening and care of haemoglobinopathies and their secondary complications. These teams would also provide psychosocial and social support to patients, taking account of cultural and language specificities of the population at risk.<sup>293</sup> Patient groups stress the need to improve the education of parents with children suffering from haemoglobinopathies. Patients and healthcare professionals are currently exploring how coordination between patients, families and medical staff can be better organised in order to help parents and patients to manage their diseases effectively.

Patients can access innovative care and screening techniques such as T2\* to measure heart and liver damage linked to iron overload, and four or seven Tesla MRI scans to trace micro strokes. Other treatments include: Arterial Spin Labeling (ASL) MRI to measure blood perfusion, transcranial Doppler ultrasound, stem cell transplants, iron chelation treatments and, following successful experience in Italy, bone marrow transplantation.<sup>294 295 296</sup> Ongoing research focuses on the neurocognitive and behavioural impacts of haemoglobinopathies and how the quality of life for children with SCD can be improved.

Awareness of haemoglobin disorders among the general public and also medical professionals is considered to be low. Experts have highlighted that the low awareness results in health complications that could be prevented. Patients and healthcare professionals run awareness campaigns and disseminate information on thalassaemia and SCD amongst the general public, the medical community and patients.<sup>297 298 299 300</sup> The Dutch Ministry of Health supports the dissemination of information on hereditary diseases, including thalassaemia and SCD.<sup>301</sup> The main sequelae and complications include cardiovascular problems, liver and kidney complications, pulmonary hypertension as well as chronic pain in SCD patients.<sup>302</sup>

Patients regret that the level of quality care is inconsistent across the country; and stress the need for improved coordination amongst medical centres, individual doctors and patients, in order to ensure an effective holistic approach to the management of these diseases. Patients have called for nationwide quality and healthcare management indicators. Daily health management of the disease, helping patients overcome difficulties towards social integration and outreach to groups of the at risk population with language and cultural specificities are other key challenges stressed by patients.<sup>303</sup>

<sup>288</sup> National Working Group of Haemoglobinopathies Practitioners, [www.hemoglobinopathie.nl](http://www.hemoglobinopathie.nl) | <sup>289</sup> Erfelijkheid.nl, Thalassemie, <http://erfelijkheid.nl/node/71> | <sup>290</sup> Erfelijkheid.nl, Sikkelcelziekte, <http://erfelijkheid.nl/node/388> | <sup>291</sup> Available from <http://statline.cbs.nl/StatWeb>, as referred in Measuring complications of sickle cell disease (PhD Thesis), X.W. van den Tweel, University of Amsterdam, (UVA), p. 11, <http://dare.uva.nl/document/134111> | <sup>292</sup> NFU, Erfelijke hemolytische anemie/ Hemoglobinopathieën, <http://www.nfu.nl/trf/index.php?id=8&specialisme=2&trf=106> | <sup>293</sup> Sikkelcelziekte en thalassemie in Den Haag: een perspectief voor een Haags Hemoglobinopathie Centrum?, Kerkhoffs et al., 2007, p. 16., [http://www.awpgnzh.nl/assets/awp/upload/2007\\_nr1\\_05-sikkelcelziekte\\_1318257206.pdf](http://www.awpgnzh.nl/assets/awp/upload/2007_nr1_05-sikkelcelziekte_1318257206.pdf) | <sup>294</sup> Nederlandse Vereniging voor Hematologie, Sikkelcelziekte, <http://www.hematologienederland.nl/sikkelcelziekte#behandeling> | <sup>295</sup> Nederlandse Vereniging voor Hematologie, Thalassemie, <http://www.hematologienederland.nl/thalassemie#symptomen> | <sup>296</sup> Nederlandse Vereniging voor Hematologie, Thalassemie, op.cit. | <sup>297</sup> Nederlandse Vereniging voor Hematologie, Sikkelcelziekte, <http://www.hematologienederland.nl/sikkelcelziekte> | <sup>298</sup> Oscar Nederland, home, <http://www.oscarnederland.nl/node/6> | <sup>299</sup> Hemoglobinopathies and Red cell Diagnostics, Human and Clinical Genetics, Leiden University Medical Center, A selection from the recent years of scientific output (2005-1996), Updated January 2006. [http://www.hbpinfo.com/view.php3?view=\\_12](http://www.hbpinfo.com/view.php3?view=_12) | <sup>300</sup> Hemoglobinopathies Laboratory, <http://www.hbpinfo.com/> | <sup>301</sup> Erfelijkheid.nl, home, <http://www.erfelijkheid.nl/node/491> | <sup>302</sup> Sikkelcelziekte en thalassemie in Den Haag: een perspectief voor een Haags Hemoglobinopathie Centrum?, op. cit. | <sup>303</sup> Project Sikkelcel Ziekte, In opdracht van OSCAR, AMC en de Hogeschool van Amsterdam, Amsterdam School of Nursing.

## Country Factsheet: The Netherlands

### POLICY OUTLOOK

#### EXPECTED POLICY DEVELOPMENTS AND TIMELINE

Plans to develop a Dutch National Plan on Rare Diseases that is specific, measurable, acceptable, realistic and time-bound.<sup>304</sup>

It is yet not known whether there will be specific references to migrants.

#### INTERVIEWEES' SUGGESTED POLICY ACTIONS

- Establishment of national database on haemoglobinopathies
- Expansion and full implementation of preconception and prenatal screening
- Greater coordination/centralisation of care
- Greater focus on a holistic approach towards care and treatment
- Education and awareness amongst the public and patients, including genetic counselling when appropriate
- European guidelines on stem cell transplantation

**“THERE ARE BIG DIFFERENCES AMONGST DOCTORS WHEN IT COMES TO AWARENESS OF SICKLE CELL DISEASE AND THALASSAEMIA AND ADEQUATE CARE AND TREATMENT. A FAIR NUMBER OF SICKLE CELL PATIENTS RECEIVE INSUFFICIENT AND INADEQUATE CARE RESULTING IN SERIOUS PAIN, HEALTH PROBLEMS OR EVEN DEATH, WHICH COULD BE PREVENTED. IN BOTH CASES, EARLY DETECTION AND INTERVENTION MAKE ALL THE DIFFERENCE.”**

**“IN THE NETHERLANDS, A HOLISTIC APPROACH BASED ON PATIENT-CENTRED CARE IS CRUCIAL TO IMPROVE PATIENTS’ LIVES AND OUTCOMES. BEYOND HOSPITAL CARE, PATIENT AND GENERAL EDUCATION MEASURES AS WELL AS OTHER SUPPORT MEASURES TO IMPROVE HOME CARE AND SELF-MANAGEMENT OF THE DISEASE SHOULD BE URGENTLY ADDRESSED WITH THE ACTIVE INVOLVEMENT OF PHYSICIANS AND HEALTHCARE SERVICES.”**

Soroya Beacher, Chairman of the OSCAR Patient Organisation in the Netherlands

<sup>304</sup> Nationaal Plan Zeldzame Ziekten, Raamwerk Nationaal Plan in wording, <http://www.npzz.nl/>



## Country Factsheet: Spain

### KEY FACTS AND FIGURES



According to healthcare professionals the estimated number of patients in Spain affected with major haemoglobinopathy is over 600. Around 80% are sickle cell disease (SCD) patients and at least 40 new cases are expected to be diagnosed every year, most of them amongst patients originating from Sub-Saharan Africa and the Maghreb.<sup>310</sup>

The number of  $\beta$  thalassaemia carriers varies significantly across regions; the average prevalence is estimated to be 0.5%, with prevalence ranging from zero in some regions of the north of Spain to five in the Canary Islands.

Recent studies have analysed SCD prevalence in Spain and the impact of migratory flows from Sub-Saharan Africa.<sup>305</sup> There is consensus amongst the healthcare community that the prevalence of haemoglobinopathies in Spain, especially SCD, is increasing exponentially as a result of recent African immigration. However, haemoglobinopathy carriers can also be found amongst the autochthonous population in the South of Spain, namely the area of Cadiz (Andalusia), with a carrier prevalence of 1 out of 415 inhabitants.

Catalonia and Madrid are the regions with the highest estimated SCD incidence per year, over 2.1 and 1.6 cases per 10,000 newborns, respectively.<sup>306 310</sup>

There is no official national registry on haemoglobinopathies covering both Thalassaemia and SCD patients. Two relevant pilot initiatives on clinical registries for haemoglobinopathies are on-going. A paediatric registry of SCD patients is maintained by the Spanish Society of Paediatric Haematology and Oncology including updated data of patients (age 0–18 years) since 2000.<sup>307</sup> Currently, 45 centres distributed across Spain are reporting to the registry. In 2012, 497 patients affected by SCD were registered.<sup>315</sup>

A second registry is also promoted by the Spanish red cell pathology group attached to the Spanish Society of haematology and haemotherapy. It covers both thalassaemic and SCD patients, adults and children.

Data in 2012 reported a total number of 316 patients, amongst which 96 were thalassaemic patients, 200 suffered SCD and the remaining suffered different forms of haemoglobinopathy combinations. A potential limitation with the paediatric registry is that duplications are not removed, however they are considered minimal.

Other available data on haemoglobinopathy patients in Spain are partial and limited to specific geographical regions.<sup>308 309</sup> In 2005, the Ministry of Health set up the Spanish Rare Diseases Registry. In 2008, the *Red Epidemiológica de Investigación de Enfermedades Raras* (REPIER - Spanish Epidemiological Network of Rare Disease Research) concluded that existing epidemiology registries in Spain failed to meet the standard criteria for data collection.<sup>310</sup>

<sup>305</sup> Neonatal haemoglobinopathy screening in Spain, María del Mar Mañú Pereira and Joan Luis Vives Corrons, *Journal of Clinical Pathology*, 2009, 62, 22–25, p.22 | <sup>306</sup> Evaluation of systematic neonatal screening for sickle cell diseases in Madrid three years after its introduction, E. Cela de Julián et al, *Anales de Pediatría*, 2008, 131, 5–9 | <sup>307</sup> Ref. poster communication "National Registry of SCD patients". Cela, E et al. V Congress of the National Society of Paediatric Haematology and Oncology", Pamplona, Spain 2012.) | <sup>308</sup> Neonatal haemoglobinopathy screening in Spain, op. cit., p.22 | <sup>309</sup> 2011 Report on the State of the Art of Rare Disease Activities in Europe of the European Union Committee of Experts on Rare Diseases - Part III: Activities in EU Member States and Other European Countries, eds S. Aymé and C. Rodwell, July 2011, p.138 | <sup>310</sup> 2011 Report on the State of the Art of Rare Disease Activities in Europe of the European Union Committee of Experts on Rare Diseases - Part III: Activities in EU Member States and Other European Countries, op. cit., p.138

## KEY FACTS AND FIGURES

In 2012, the *Instituto de Investigación en Enfermedades Raras del Instituto de Salud Carlos III* (Institute of Rare Diseases Research - Health Institute Carlos III, attached to the Ministry of Economy and Competitiveness) initiated a Collaborative Research Joint Project entitled 'Spanish Rare Disease Registries Research Network – SpainRDR'. This project is part of the International Rare Diseases Research Consortium – IRDiRC for the establishment of a national registry and harmonised methodology for gathering and processing data on rare diseases.<sup>311</sup> Disaggregated data on migrants' cultural and religious background and/or country of origin however is not available according to the current regulatory framework.

There is no official available data on the estimated costs related to haemoglobinopathies. There is ongoing research to build appropriate models to quantify the socio-economic costs linked to a selected group of ten rare diseases.<sup>312</sup>

Although haemoglobin disorders are not included, experts believe that the results and conclusions may potentially be extended to haemoglobinopathies in order to allow for an estimation of the associated socio-economic burden.

## POLICY FOCUS

The *Ministerio de Sanidad, Servicios Sociales e Igualdad* (Ministry of Health, Social Services and Equality)<sup>313</sup> in Spain is responsible for shaping national health and social affairs policy.

Together with the regional governments, the Ministry sets healthcare minimum standards and policy objectives for the entire country. These are developed and implemented by each of the 17 autonomous regions (ARs) – including two autonomous cities located in the North of Africa – according to their local needs and priorities. Spain is one of the few EU countries where there is free, universal access to healthcare across the country. However, more recently, due to the severe austerity measures, the government has limited the access to healthcare for migrants in illegal situation and their families, hindering access to adequate screening, treatment and care.

Haemoglobinopathies are addressed under the policy framework for rare diseases in Spain, and the policy approach and impact vary widely across the different ARs.<sup>314</sup> The Spanish government adopted in 2009 the *Estrategia en Enfermedades Raras del Sistema Nacional de Salud* (the National Rare Disease Strategy).<sup>315 316</sup> This strategy sets out the general framework for the prevention and care of rare diseases. Regional authorities are responsible for further developing and implementing this framework. The national strategy addresses seven main areas for action: information, prevention and early detection, healthcare coordination, treatment, social services, research and training.<sup>317</sup> It also sets up a number of general and specific objectives, with their respective recommendations and monitoring indicators. Until now, there has been no specific focus on haemoglobinopathies. Healthcare professionals believe that the strategy helped formalise the existing procedures already established on an unofficial basis in some regions across the country. However, it did not contribute to introducing innovative healthcare management approaches.

<sup>311</sup> 2011 Report on the State of the Art of Rare Disease Activities in Europe of the European Union Committee of Experts on Rare Diseases - Part III: Activities in EU Member States and Other European Countries, op. cit., p.138 | <sup>312</sup> Burqol Rare Diseases project (funded by the European Commission), <http://www.burqol-rd.com> | <sup>313</sup> Ministry of Health and Social Policy, <http://www.msc.es/home.htm> | <sup>314</sup> Report on the neonatal screening programmes situation in Spain, Committee for Public Health of the Interterritorial Council of the National Health System, 2007 | <sup>315</sup> Rare Disease Strategy of the Spanish National Health System, Ministry of Health and Social Policy, [http://ec.europa.eu/health/ph\\_threats/non\\_com/docs/rare\\_strategy\\_spain\\_en.pdf](http://ec.europa.eu/health/ph_threats/non_com/docs/rare_strategy_spain_en.pdf) | <sup>316</sup> 2011 Report on the State of the Art of Rare Disease Activities in Europe of the European Union Committee of Experts on Rare Diseases - Part III: Activities in EU Member States and Other European Countries, op. cit., p.136 | <sup>317</sup> 2011 Report on the State of the Art of Rare Disease Activities in Europe of the European Union Committee of Experts on Rare Diseases - Part III: Activities in EU Member States and Other European Countries, op. cit.

## POLICY FOCUS

A first evaluation on the implementation and the impact of the Rare Disease Strategy began in 2012. Healthcare professionals have expressed concerns about the evaluation exercise, and feel that the conclusions do not reflect an accurate assessment of the haemoglobinopathies situation and challenges which need to be addressed in the future.

Patients and healthcare professional representatives were involved in the development of the National Strategy for Rare Diseases.<sup>318</sup> However, haemoglobinopathy stakeholders regret the lack of focus on these diseases, despite the fact they are the most common rare disorders.

**“DESPITE POSITIVE PROGRESS IN THE LAST YEARS, A NUMBER OF ISSUES NEED TO BE URGENTLY ADDRESSED. AN INCREASINGLY COORDINATED POLICY APPROACH ACROSS THE COUNTRY, AWARENESS AND KNOWLEDGE AMONGST THE MEDICAL COMMUNITY, AND THE ESTABLISHMENT OF A SPECIALISED REFERENCE CENTRE ON HAEMOGLOBINOPATHIES ARE ESSENTIAL STEPS IN THE WAY FORWARD IN SPAIN.”**

Joan Lluís Vives-Corróns, Hospital Clinic de Barcelona

## PREVENTION AND SCREENING

### There is no national official preventive programme for haemoglobinopathies.

Official universal systematic screening programmes on haemoglobinopathies for neonates are practised in some regions: Extremadura since 2002, Madrid since 2003, Basque Country since 2011 and Valencia 2012.

Since 2009 Catalonia, Sevilla (Andalusia), Murcia, Castilla Leon, Galicia and Baleares Islands have performed non official pilot programmes for the neonatal screening of haemoglobinopathies. Catalonia has not yet established an official programme for neonatal screening of SCD despite being the Spanish region with the highest African immigration flow and SCD incidence.<sup>319 320</sup>

Since 2013, the Catalan Network for the Diagnosis and Follow-up of Major Haemoglobinopathies (CATGLOBIN), sponsored by the “Marato Foundation” and supported by the Catalan Agency for Public Health, is running a pilot universal neonatal screening project, with a view to establishing a consolidated programme linked to the official regional neonatal screening programme for inherited metabolic disorders. Pilot studies and research have demonstrated that universal screening should be standard practice for regions with a high annual birth rate and SCD prevalence, such as Catalonia and Madrid.<sup>321</sup>

Screening of target at-risk ethnic groups, including African immigrants, varies widely across the different regions and hospitals.<sup>322</sup> There are no standardised practices or criteria to define individuals at risk. Targeted screening is generally carried out on patients with African origins or relevant apparent physical traits.

<sup>318</sup> 2011 Report on the State of the Art of Rare Disease Activities in Europe of the European Union Committee of Experts on Rare Diseases - Part III: Activities in EU Member States and Other European Countries, op. cit., p.136 | <sup>319</sup> Neonatal haemoglobinopathy screening in Spain, op. cit., p.24 | <sup>320</sup> Neonatal haemoglobinopathy screening in Spain, op. cit., p.24 | <sup>321</sup> Neonatal haemoglobinopathy screening in Spain, op. cit., p.25 | <sup>322</sup> Neonatal haemoglobinopathy screening in Spain, op. cit., p.23

## PREVENTION AND SCREENING

Although there is no official prenatal programme for prevention of major haemoglobinopathies, prenatal screening in the clinical centres is recommended to individuals at risk.

Healthcare professionals say that access to innovative screening techniques remains a major challenge for patients in Spain. They generally perceive wide inequalities in terms of access to quality, innovative healthcare across the different regions.

Awareness and education initiatives are generally run by patient organisations.<sup>323</sup> Patients recognise, however, that campaigns have a limited impact due to scarce resources and call on the government to provide appropriate financial support.

## TREATMENT AND CARE

In 2002, the Spanish Association of primary care paediatricians published a 'Guideline for clinical care of the migrant child' emphasising social, education and integration aspects, however haemoglobinopathies were not well covered.<sup>324</sup>

The same year, the Spanish Society of Paediatric Haematology and Oncology published a guideline for the diagnosis and clinical follow-up of the patients affected by SCD. It also emphasised the need to register the new cases annually. This guideline is widely followed by paediatricians in the whole country.<sup>325</sup>

Experts and patients believe that decentralisation in Spain hinders appropriate and equal access to diagnosis, treatment and care. Until 2012, the Spanish social security system covered the costs relating to diagnosis and treatment of patients with major haemoglobinopathies. However, recent health policy measures in some Spanish regions have obliged patients to cover part of the costs related to healthcare.

Access to specialised and innovative care, as well as the level of patient reimbursement, are considered heterogeneous. For example, patients stress that only a few regional health services provide free of charge pre-implantation genetic diagnosis (PGD).

Experts and patients consider that this should be urgently addressed and improved, through comprehensive nationwide guidelines, greater access to medical equipment, specialised training and education for healthcare professionals. Stakeholders also express deep concerns over the negative impact of current cost containment measures on the quality of healthcare delivery, as well as concerns over the restrictions in access to health care. The Royal decree of 2012, which entered in force in April 2013 limiting access to health care for migrants in illegal situation, as well as some categories of citizens, will also have consequences on the appropriate management of haemoglobinopathies in these populations.

<sup>323</sup> Asociación Española de Lucha contra las Hemoglobinopatías y Talasemias, <http://www.alheta.com> | <sup>324</sup> "Guideline for clinical care of the migrant child", <http://www.aepap.org/inmigrante/area11.pdf> | <sup>325</sup> Guideline for the diagnosis and clinical follow-up of the patients affected by SCD, <http://www.svnp.es/Documen/protodrepanocitosis.htm>

## TREATMENT AND CARE

The *Centro de Referencia Estatal de Atención a Personas con Enfermedades Raras y sus Familias* (CREER - National Centre for Rare Disease Patients) was established in 2009 in Burgos as the main reference centre to ensure quality, specialised care and greater expert coordination through education and information activities targeted at patients and healthcare professionals.<sup>326 327</sup> Healthcare professionals regret that the centre lacks the necessary competences and resources to function as a real centre of reference as its activities are confined to providing information and specialised social support services for patients and caregivers.

The Ministry, in collaboration with the regional governments, experts and professional organisations, is working to set up reference centres for rare diseases and other diseases whose treatment and care require advanced specialisation and technology. In December 2012, the official site of the *Ministerio de Salud, Servicios Sociales e Igualdad* (Ministry of Health, Social Services and Equality) reported that a total of 166 national reference centres were already recognised for 45 pathologies. Most of them are related to the application of advanced technologies, namely transplantation procedures or surgery for specific pathologies, including a few rare diseases. At the same time, the autonomous communities are initiating plans to design regional centres of expertise for rare diseases. At the time of writing, a detailed list of centres including services offered and pathologies covered was not available.

Migration into Spain is considered a recent trend, in comparison with other European countries. Experts consider that because of their particularly vulnerable social and economic situation, migrants do not tend to exercise their rights to healthcare or demand support services when needed. A past pilot programme in Mataró (Catalonia) looked into the integration of mediators in medical teams working with SCD patients amongst the migrant community.

The results showed increased adherence of the patients to the treatment and follow up visits. The project was stopped due to lack of funding. However, it showed that language and cultural barriers should be addressed through targeted measures, including information on the disease and its management in the language of origin of the patient.

Suggested measures amongst stakeholders in order to improve care include: uniform and homogenised care standards and policies at national level; creating centres of reference and expertise, supported with appropriate resources and coordination; introducing early detection programmes at national level; and universal screening programmes for those geographical regions with a higher prevalence of haemoglobinopathies.

Improving the awareness and adequate training of primary healthcare professionals is also identified as an important means to facilitate early diagnosis, timely referral to specialists and appropriate long-term management of these diseases.

## POLICY OUTLOOK

## EXPECTED POLICY DEVELOPMENTS AND TIMELINE

New common registry and a harmonised methodology for gathering and processing data on rare diseases, by the Institute of Rare Diseases Research (Institute of Health Carlos III).

<sup>326</sup> Spanish Official Journal, Royal Decree 1302/2006 of 10 November 2006. 11 November 2006. <http://www.msc.es/profesionales/CentrosDeReferencia/docs/RdCsur.pdf> | <sup>327</sup> State reference centre for the treatment of patients with rare diseases and their families, [http://www.creenfermedadesraras.es/creer\\_01/index.htm](http://www.creenfermedadesraras.es/creer_01/index.htm)

## Country Factsheet: Sweden

### KEY FACTS AND FIGURES



National Board of Health and Welfare data from 2004 and 2009 indicate that there are approximately 150 patients affected by haemoglobinopathies in Sweden (approximately 100 sickle cell disease [SCD] patients and 50 thalassaemia patients).<sup>328 329</sup>

The number of patients affected by haemoglobin disorders is considered to have increased since 2009, although there are no specific data on how many new haemoglobinopathy patients are diagnosed every year. The country of origin is generally considered a relevant prevalence factor for people with both SCD and thalassaemia.<sup>330 331</sup> Healthcare professionals note that new diagnoses are linked to migration flows.

There are no comprehensive data on the life expectancy of haemoglobinopathy patients in Sweden. It is nevertheless acknowledged that life expectancy of patients with haemoglobinopathies has improved significantly as a result of new and improved treatment and care.

There is no specific registry for haemoglobinopathies in Sweden, the main reason being that the disease concerns such a small number of patients according to government officials. Healthcare professionals are taking initiatives to implement a national registry for SCD and thalassaemia. The registry is currently not supported by the government. Registries can apply for funding from the National Healthcare Quality Registries once they fulfill certain criteria.<sup>332</sup>

The National Board of Health and Welfare, the main public authority in charge of the healthcare system under the Ministry of Health, defines haemoglobinopathies as rare diseases that lead to numerous disabilities.<sup>333</sup>

The National Board gathers relevant statistical information on the support and services for people with disabilities,<sup>334</sup> and care provided by the local authorities to people with disabilities and the elderly. At the time of this research, a database was being developed by the National Board to centralise information on small and/or lesser-known groups of disabled people and to map the support and services these groups may need. The Swedish Information Centre for Rare Diseases, founded by the National Board, is responsible for the Swedish Rare Disease Database. It supports and compiles information gathered by leading experts across the country on each rare disease (including SCD and thalassaemia), which is reviewed by the scientific advisory board before publication. Patients from disability organisations are also involved.<sup>335</sup>

The National Board of Health and Welfare provides general information on SCD and thalassaemia related symptoms, sequelae and diseases. Pain is considered one of the major symptoms of sickle cell disorders, also often leading to under- or misdiagnosis of affected patients.<sup>336 337</sup> Liver problems, cirrhosis, heart failure or arrhythmia, and endocrine disorders are considered amongst the most common sequelae.<sup>338 339</sup>

There are no available data or evidence on the estimated costs linked to the care of haemoglobinopathy patients in Sweden. Healthcare professionals note, however, that hospital care related costs represent the major cost linked to the care of sickle cell disorders, whereas medicinal treatment tends to be a major cost in the case of thalassaemia patients.

<sup>328</sup> National Board of Health and Welfare, <http://www.socialstyrelsen.se/statistik/statistikefteramne/personermedfunktionsnedsattning-vardochomsorg> | <sup>329</sup> National Board of Health and Welfare, <http://www.socialstyrelsen.se/ovanligadiagnoser/sicklecellsanemi> (Publication date 2004-06-22) | <sup>330</sup> National Board of Health and Welfare, <http://www.socialstyrelsen.se/ovanligadiagnoser/sicklecellsanemi> (Publication date 2004-06-22) | <sup>331</sup> Läkartidningen Nr 1-2, 1996, Volym 93 <http://larkiv.lakartidningen.se/1996/temp/pda11544.pdf> | <sup>332</sup> Swedish Municipalities and Regions (Sveriges Kommuner och Landsting), [http://www.kvalitetsregister.se/om\\_kvalitetsregister/quality\\_registries](http://www.kvalitetsregister.se/om_kvalitetsregister/quality_registries) | <sup>333</sup> Rare Diseases Sweden: [http://www.sallsyntadiagnoser.se/Om\\_oss/Vanliga\\_fragor/](http://www.sallsyntadiagnoser.se/Om_oss/Vanliga_fragor/) | <sup>334</sup> National Board of Health and Welfare: <http://www.socialstyrelsen.se/statistik/statistikefteramne/personermedfunktionsnedsattning-stodochservice> | <sup>335</sup> University of Gothenburg, the Sahlgrenska Academy, [http://www.sahlgrenska.gu.se/english/sgc\\_eng/?languageId=100001&contentId=-1&disableRedirect=true&returnUrl=http%3A%2F%2Fwww.sahlgrenska.gu.se%2Fovanligadiagnoser%2F](http://www.sahlgrenska.gu.se/english/sgc_eng/?languageId=100001&contentId=-1&disableRedirect=true&returnUrl=http%3A%2F%2Fwww.sahlgrenska.gu.se%2Fovanligadiagnoser%2F) (Updated 2010-06-05) | <sup>336</sup> National Board of Health and Welfare: [http://www.socialstyrelsen.se/ovanligadiagnoser/sicklecellsanemi#anchor\\_4](http://www.socialstyrelsen.se/ovanligadiagnoser/sicklecellsanemi#anchor_4) (Publication date 2004-06-22) | <sup>337</sup> National Board of Health and Welfare: <http://www.socialstyrelsen.se/ovanligadiagnoser/thalassemi> (Publication date 2004-06-22) | <sup>338</sup> National Board of Health and Welfare: <http://www.socialstyrelsen.se/ovanligadiagnoser/thalassemi> | <sup>339</sup> Mini Care Program for thalassaemia: [www.orebroll.se/vph/vardprogram.htm](http://www.orebroll.se/vph/vardprogram.htm) (Publication date 2009-06-01)



## Country Factsheet: Sweden

### POLICY FOCUS

### PREVENTION AND SCREENING

In Sweden, ensuring equal access to quality health and care for the entire population is a major goal of the healthcare system.<sup>340</sup>

Full healthcare coverage is extended to people applying for a residence permit as a refugee, people granted a residence permit, held in detention or with a temporary residence permit.<sup>341</sup> Asylum seekers are also entitled to a free health assessment, as well as to emergency medical and dental care.<sup>342</sup>

As regards migrants' health, the main focus for the government is the Cross-Border Healthcare Directive, where Sweden has been one of the main drivers. However, the Directive only applies to people (EU or non) legally residing in EU member states and with health insurance, so does not cover all types of migrants or even non-insured EU citizens. In 2012, the minority government and the Green party agreed to cooperate in the area of healthcare for hidden and paperless refugees. It is not yet clear, however, what the result of this cooperation will be. Generally there has been a movement towards granting access to migrants in the area of healthcare, note government officials.

Sweden did not have (at the time of this report) a dedicated national plan on rare diseases in place, but the National Board of Health and Welfare has been tasked by the Government to present relevant information for such a plan. The information is expected to be presented in October 2012. It was, at the time of writing, not yet known what the materials would include or if they will result in a national plan for rare diseases.<sup>343</sup> The National Board of Health and Welfare carries out this task in cooperation with the Karolinska Institute and the National Operation of Rare Disorders ([www.nfsd.se](http://www.nfsd.se)).<sup>344</sup>

Healthcare professionals regret the fact that awareness of haemoglobinopathies and linked health risks, sequelae and social impact is generally limited to a reduced number of specialists. Awareness amongst the medical community and the general public is poor or nonexistent. The Swedish Information Centre for Rare Diseases is responsible for raising general awareness of rare diseases. However, research shows no results for campaigns specifically on haemoglobinopathies.

There are no special screening programmes for SCD and thalassaemia.

Prenatal screening is generally offered to expecting parents with family records of haemoglobinopathies. In the case of an affected foetus, parents may consider whether or not to terminate the pregnancy. There are no special screening programmes or clear models or screening protocols. There are, however, ongoing discussions amongst medical professionals and maternity clinics on how to provide targeted screening to the population at risk of being disease carriers, in order to prevent pregnancies with affected fetuses.

The care programmes for both SCD and thalassaemia developed by healthcare professional organisations include a focus on screening indicators for diagnosis of health complications. These include transcranial Doppler screening in children with SCD and magnetic resonance screening for thalassaemia. Generally, healthcare professionals believe that patients have access to the latest, most innovative screening techniques. However, they also noted that this may not always be the case across the whole country nor for all people residing within Sweden.

<sup>340</sup> Sweden's Parliament, The Healthcare Law, 1982, [http://www.riksdagen.se/sv/Dokument-Lagar/Lagar/Svenskforfattningssamling/Halso--och-sjukvardslag-1982\\_sfs-1982-763/?bet=1982:763](http://www.riksdagen.se/sv/Dokument-Lagar/Lagar/Svenskforfattningssamling/Halso--och-sjukvardslag-1982_sfs-1982-763/?bet=1982:763)

<sup>341</sup> Sweden's Parliament, The Healthcare Law for Asylum seekers, 2008, [http://www.riksdagen.se/sv/Dokument-Lagar/Lagar/Svenskforfattningssamling/Lag-2008344-om-halso--och-s\\_sfs-2008-344/?bet=2008:344](http://www.riksdagen.se/sv/Dokument-Lagar/Lagar/Svenskforfattningssamling/Lag-2008344-om-halso--och-s_sfs-2008-344/?bet=2008:344)

<sup>342</sup> Sweden's Parliament, Regulation (1996:1357) About State Reimbursement for Healthcare for Asylum Seekers, 1996, [http://www.riksdagen.se/sv/Dokument-Lagar/Lagar/Svenskforfattningssamling/Forordning-19961357-om-stat\\_sfs-1996-1357/](http://www.riksdagen.se/sv/Dokument-Lagar/Lagar/Svenskforfattningssamling/Forordning-19961357-om-stat_sfs-1996-1357/)

<sup>343</sup> Interview, desk officer, Ministry of Social Affairs.

<sup>344</sup> Karolinska Institutet: <http://ki.se/ki/jsp/polopoly.jsp?d=36310&a=115614&l=sv>

## Country Factsheet: Sweden

### TREATMENT AND CARE

### POLICY OUTLOOK

#### Patients with haemoglobinopathies in Sweden are taken care of by specialist physicians.<sup>345</sup>

Care is free of charge after patients have reached their annual standard consultation contribution, set at a maximum of SEK 1,100/€127 per patient per year. Such a system benefits individuals covered by the Swedish social security system.

The Swedish Paediatric Society has developed two programmes for the care and treatment of haemoglobinopathies, which serve as guidelines for healthcare professionals: the Care Programme for Sickle Cell Disease and the Mini Care Programme for Thalassaemia. The SCD care programme includes diagnostics, treatment and treatment of complications as well as what information should be provided to parents and their child. The aim of the mini care programme for thalassaemia is to provide recommendations around the most common questions that health care professionals may have when treating children with thalassaemia.

Experts believe that patients in Sweden have access to the highest level of innovative care. Sharing of knowledge and implementation of state of the art treatments is effective. There are, however, concerns as to variations across the country, with some regions lagging behind.

Physicians are generally patients' primary source of information about haemoglobin disorders. Experts stress the importance of patient education in order to ensure optimal management of these complicated diseases. Increased awareness and education amongst both patients and medical staff about the diseases, implications and care needs are major challenges that need to be addressed in order to improve care and patients' quality of life. In the case of thalassaemia, experts consider that patient non-compliance with treatment remains a major obstacle to optimal disease management.

#### EXPECTED POLICY DEVELOPMENTS AND TIMELINE

Both the SCD and thalassaemia care programmes are expected to be updated within the upcoming years (uncertain timing).

Adoption and full implementation of Swedish Plan on Rare Diseases and Rare Disease Registry.

#### INTERVIEWEES' SUGGESTED POLICY ACTIONS

- Targeted education and awareness amongst patients and professionals

<sup>345</sup> National Board of Health and Welfare: <http://www.socialstyrelsen.se/ovanligadiagnoser/sicklecellsanemi> (Publication date 2004-06-22) <http://www.socialstyrelsen.se/ovanligadiagnoser/thalassemi> (Publication date 2004-06-22)

## Country Factsheet: United Kingdom

### KEY FACTS AND FIGURES



Experts estimate that there are about 800 patients with thalassaemia and 15,000 with sickle cell disease (SCD) in the UK. A large number are under 19 years of age.

Data from the National Haemoglobinopathy Registry (NHR) from 2012 report 4,595 haemoglobinopathy patients in the UK.<sup>346 347</sup> The NHR was run between 2009 and 2012, following past initiatives to study the number of children affected by thalassaemia amongst Cypriot migrants in the London area,<sup>348</sup> and to share clinical and research knowledge on thalassaemia across the UK<sup>349 350</sup> 4,595 patients were registered, with an average increase of more than 1,500 additional patients registered per year.<sup>351 352</sup> The registry, however, operated on a voluntary basis and therefore does not provide complete data on all patients across the country, note healthcare professionals. An envisaged measure to enhance uptake would be to make it mandatory for physicians to put new cases on the registry.

Despite the progressive reduction in the number of deaths amongst haemoglobinopathy patients, data from the NHS Sickle Cell & Thalassaemia Screening Programme show a current rising trend in the number of patients.

On average, 360 newborns<sup>353</sup> are identified as carriers of a haemoglobin disorder.<sup>354</sup> Between 2009 and 2010, 1.4% (total number 9,732) of newborn babies in the UK were identified as haemoglobinopathies carriers.<sup>355</sup>

The number of carrier fetuses identified in the prenatal screening programme has increased significantly, with a peak in 2008 to 2009 when over 22,000 fetuses were identified as haemoglobinopathies carriers, amongst 650,000 screened pregnant women.<sup>356</sup>

Experts explain that this trend is due to the impact of migration flows in the UK. However, different trends apply to SCD and thalassaemia prevalence. While SCD is considered to have been a rapidly growing problem in the last decade due to strong immigration flows from African, Caribbean and Pacific states, thalassaemia patients have decreased as a result of effective prevention and screening policies in the countries of origin (e.g. Cyprus) and adherence to screening programmes amongst the migrant population living in the UK. According to consulted healthcare sources, screening programmes are considered more successful amongst groups of the population with higher education, social and economic status (e.g. migrants from Cyprus or India).

There are no recent data on the costs linked to care and treatment of haemoglobin disorders.<sup>357</sup> However experts explain that the costs connected to the treatment, care and medication of haemoglobinopathy patients is estimated to add up to 52 million GBP (30 million GBP for treatment and care and 22 million GBP for medication).

SCD is one of the most common inherited conditions in the UK; around 300 babies are born each year with SCD compared to 20–30 babies with thalassaemia.<sup>358</sup>

Almost 60% (2,673 patients) of the haemoglobinopathies registered in the NHR have sickle cell anaemia (Hb SS).<sup>359</sup> According to consulted healthcare sources, the growth of the number of patients suffering from haemoglobin disorders is estimated to be around 1.5 to 1.8% per year. This estimated growth rate does not take into account patient migration.

<sup>346</sup> The following diagnoses are counted in the National Haemoglobinopathy Registry: Beta thalassaemia intermedia, Beta thalassaemia major, Beta thalassaemia/Hb E disease, Haemoglobin H disease, Hb S/D Punjab, Hb S/E, Hb S/HPFH, Hb S/D Arab, Hb SC Disease, HbS/Lepore, Other, Sickle Cell Anaemia (Hb SS), SickleB+ thal, SickleB0thal. <sup>347</sup> National Haemoglobinopathy Registry Information Service, Number of Patients Registered by Diagnosis, 2012, available from [http://www.nhr.nhs.uk/docs/pdfs/InfoNo\\_Patients\\_By\\_Diagnosis.pdf](http://www.nhr.nhs.uk/docs/pdfs/InfoNo_Patients_By_Diagnosis.pdf) <sup>348</sup> Improved survival of thalassaemia major in the UK and relation to T2\* cardiovascular magnetic resonance, op. cit. <sup>349</sup> Improved survival of thalassaemia major in the UK and relation to T2\* cardiovascular magnetic resonance, op. cit. <sup>350</sup> National Haemoglobinopathy Registry Working for improved Treatment Services (NHR newsletter June 2009) in: <http://www.nhr.nhs.uk/docs/newsletters/NHR%20Newsletter%20final.pdf> <sup>351</sup> Lords Hansard text for 18 Mar, 2010/18, Mar 2010, pt 0003, <http://www.publications.parliament.uk/pa/ld200910/ldhansrd/text/100318w0003.htm> <sup>352</sup> Lords Hansard text for 18 Mar, op. cit. <sup>353</sup> The average calculation is based on the following periods and the respective number of newborns identified as carriers of a haemoglobin disorder: In the period 2007/8: 359 newborns, in the period 2008/9: 360 newborns, in the period 2009/10: 361 newborns <sup>354</sup> Cumulated data from NHS Sickle Cell and Thalassaemia Screening Programme Laboratory Data Report 2007–2008: Development. Towards a Quality Report, 2009; NHS Sickle Cell and Thalassaemia Screening Programme. Data Report, 2008/09. Informing Policy and Improving Outcome, 2011; NHS Sickle Cell and Thalassaemia Screening Programme Data Report, 2009/10. Performance Analysis and Quality Improvement, 2012 <sup>355</sup> National Health Service Screening Programmes Sickle Cell and Thalassaemia, Annual Report 2010–11, p.8. <sup>356</sup> NHS Sickle Cell and Thalassaemia Screening Programme. Data Report, 2008/09. Informing Policy and Improving Outcome, 2011. <sup>357</sup> Lifetime treatment costs of beta-thalassaemia major, J. Karmon et al., in Clin Lab Haematol, Decemehr 1999, 21(6):377–85. <sup>358</sup> Specialised services national definitions set (SSNDS) Definition No. 38 Specialised Haemoglobinopathy Services (all ages) (3rd Edition), available from [http://www.specialisedservices.nhs.uk/library/26/Specialised\\_Haemoglobinopathy\\_Services\\_all\\_ages.pdf](http://www.specialisedservices.nhs.uk/library/26/Specialised_Haemoglobinopathy_Services_all_ages.pdf) <sup>359</sup> National Haemoglobinopathy Registry Information Service, Number of Patients Registered by Diagnosis, op. cit.

## KEY FACTS AND FIGURES

Research has shown that 240,000 people are symptom-free SCD carriers and over 12,500 people suffer from SCD. The highest prevalence of SCD is found amongst Black Caribbeans, Black Africans and Black British.<sup>360</sup>

SCD is considered a rising cause of mortality and morbidity in England and consequently an important focus for the National Health Service.<sup>361</sup> The rate of hospital admissions related to health complications in SCD patients has risen from 21.2 in 2001 to 2002, to 33.5 in 2009 to 2010, which is equivalent to a rise of over 50%.<sup>362</sup> Studies have shown that hospitalisations amongst SCD patients can be reduced and prevented through improved ambulatory care of patients.<sup>363</sup>

Thalassaemias are less common than SCD in the UK; 214,000 people are considered carriers of the thalassaemia gene variant; 700 people suffer from a more or less severe form of thalassaemia. Thalassaemia is particularly prevalent amongst Cypriots, Indians, Pakistanis, Bangladeshis, Chinese, and other groups of population with Asian origin.<sup>364</sup>

The most common causes of death amongst thalassaemia patients includes infections, bone marrow transplantation complications, and cardiac disease due to iron overload. Data from 2000 to 2003 showed that the total number of deaths has decreased to 4.3 per 1,000 patients per year, which represents a decrease of more than 60%. This is explained mainly by the management and treatment of iron overload and linked heart problems amongst patients (-62%,  $p < 0.05$ ).<sup>365 366</sup>

## POLICY FOCUS

### The Parliamentary Group on Sickle Cell and Thalassaemia gathers together a group of 30 peers from the House of Lords and cross-party MPs who share an interest in sickle cell and thalassaemia.

The purpose of the parliamentary group is “to reduce the health inequalities that are faced by sickle cell and thalassaemia patients in the UK by improving standards of care and by addressing other critical issues, as recommended by the key stakeholders.”<sup>367</sup>

The group aims to raise awareness relating to the conditions and needs of patients amongst parliamentary colleagues, the government, health professionals, and the broader community. It has called for greater coordination in the NHS, at a local and regional level, greater choice and flexibility for patients, as well as appropriate training for health professionals about SCD and thalassaemia. The group also calls for higher awareness in schools and social services of SCD and thalassaemia, better engagement of the Government with the voluntary sector to provide patient support and community education, and the exemption of patients with long-term conditions, including those with SCD and thalassaemia from prescription charges.<sup>368</sup> These measures are currently in place in Wales, Northern Ireland and Scotland.

Patients, however, feel that stronger, targeted policy action is needed. Patient groups play an active role in raising public awareness on the health risks, sequelae and social impact derived from haemoglobinopathies.<sup>369 370</sup> Particular campaigns focus on promoting integration and support for patients at the workplace<sup>371</sup> and at school.<sup>372</sup>

<sup>360</sup> Screening for Sickle Cell and Thalassaemia, Streetly A. et al, 2011. <sup>361</sup> Trends in hospital admissions for sickle cell disease in England, AlJuburi G et al., 2001/02–2009/10, in : Journal of Public Health, June 2012. <sup>362</sup> Trends in hospital admissions for sickle cell disease in England, op. cit. <sup>363</sup> Trends in hospital admissions for sickle cell disease in England, op. cit. <sup>364</sup> Screening for Sickle Cell and Thalassaemia, op.cit <sup>365</sup> Improved survival of thalassaemia major in the UK and relation to T2\* cardiovascular magnetic resonance, B. Modell, M. Khan, M. Darlison, M.A. Westwood, D. Ingram, D.J. Pennell, J Cardiovasc Magn Reson. 2008, 10:42. <sup>366</sup> Improved survival of thalassaemia major in the UK and relation to T2\* cardiovascular magnetic resonance, op. cit. <sup>367</sup> UK Parliament Register Of All-Party Groups [as at 15 June 2012], <http://www.publications.parliament.uk/pa/cm/cmallparty/register/sickle-cell-and-thalassaemia.htm>. <sup>368</sup> All-Party Parliamentary Group on Sickle Cell and Thalassaemia, Sickle Cell Disease and Thalassaemia: A Health Check, p. 4, <http://www.ukts.org/pdfs/awareness/appg.pdf>. <sup>369</sup> Website of UK Thalassaemia Society (UKTS), <http://www.ukts.org/> <sup>370</sup> Website of UK Sickle Cell Society, <http://www.sicklecellsociety.org/> <sup>371</sup> UK Thalassaemia Society - Careers Advice website, <http://www.ukts.org/careers.html?i6s3s1> <sup>372</sup> UK Thalassaemia Society information leaflet for teachers, [http://www.ukts.org/pdfs/awareness/ukts\\_%20school\\_leaflet.pdf](http://www.ukts.org/pdfs/awareness/ukts_%20school_leaflet.pdf)

## POLICY FOCUS

The Department of Health is currently preparing the national rare disease plan, which is expected to create specialised centres for haemoglobinopathies care.<sup>373</sup>

Responsibilities for the NHR and the organisation of the provision of services for haemoglobinopathy services have now passed from the Department of Health to Specialised Commissioning Groups. Specialised commissioning is the means by which Primary Care Trusts (PCTs) work together to plan, buy and manage services which treat patients with rare conditions. Specialised Commissioning Groups are responsible for the commissioning arrangements for specialised services. Specialised services are limited and generally provided by fewer than 50 hospitals.<sup>374</sup>

At the time of writing of this report, the commissioning was organised on a regional basis but plans were already under consideration to transfer commissioning to the national level in order to cope with the immense inconsistency in treatment and care within the country, e.g. regional disparities.

## PREVENTION AND SCREENING

**Antenatal haemoglobinopathy screening is routinely recommended to all pregnant women by weeks 8–10 of pregnancy.**

Despite this, evidence shows that in most cases prenatal diagnosis is still done too late to allow parents to make an informed, timely choice about the pregnancy.

The antenatal care guideline of the National Institute for Health and Clinical Excellence (NICE)<sup>375</sup> recommends strengthening care and diagnosis. The NHS provides for a Family Origin Questionnaire. More recently, research has highlighted that taking account only of “ancestors’ origins or ethnicity” to identify potential carriers of haemoglobinopathies can result in under- or misdiagnosis. It is well known that the real risk factor for being a carrier of these disorders is linked to the exposure of one’s ancestors to environmental factors, namely malaria. Malaria exerts selective pressure, favoring the survival of people who are more resistant to the parasite, which includes haemoglobinopathy carriers. Recent data from newborn screening in England suggests that newborns who were allocated to the UK Census category “White British” may have carrier rates around 1 in 500.<sup>376</sup>

In England if the population has a sickle cell/thalassaemia carrier rate higher than 1 in 1,000, then they are defined as an at-risk group. Therefore, based on these screening thresholds, White British are a high risk group for carrying genes associated with sickle cell/thalassaemia. This is the reason why some experts consider that neonatal screening programmes that assess patients’ family history of exposure to the risk factors will be more effective than those relying solely on the concept

<sup>373</sup> Rare Diseases Consultation, [http://consultations.dh.gov.uk/rare-diseases/rare\\_diseases\\_consultation](http://consultations.dh.gov.uk/rare-diseases/rare_diseases_consultation) | <sup>374</sup> Specialised services national definitions set (SSNDS) Definition No. 38 Specialised Haemoglobinopathy Services (all ages) (3rd Edition), [http://www.specialisedservices.nhs.uk/library/26/Specialised\\_Haemoglobinopathy\\_Services\\_all\\_ages.pdf](http://www.specialisedservices.nhs.uk/library/26/Specialised_Haemoglobinopathy_Services_all_ages.pdf) | <sup>375</sup> Antenatal care Routine care for the healthy pregnant woman in, NICE, <http://www.nice.org.uk/nicemedia/pdf/CG062NICEguideline.pdf> | <sup>376</sup> Territory, ancestry and descent: the politics of sickle cell disease. *Sociology* 45 (6): 963–976. Carter, B and Dyson, SM (2011). Not thinking ethnicity: a critique of the ethnicity paradigm in an over-ethnicised sociology. *Journal for the Theory of Social Behaviour* 40 (1): 1–18. Presentation at Thalassaemia International Federation Third Pan European Congress, 25–26 October 2012, Dyson, SM. | <sup>377</sup> Lessons for Intermediate and Low Prevalence Areas in England from the Ethnicity Questions and Antenatal Screening for Sickle Cell/Thalassaemia (EQUANS) Study. *Diversity in Health and Social Care* 4 (2): 123–35. Dyson, SM; Chambers, K; Gawler, S; Hubbard, S; Jivanji, V; Sutton, F; and Squire, P (2007) and based on the presentation given by Simon Dyson at Thalassaemia International Federation Third Pan European Congress, 25–26 October 2012. | <sup>378</sup> Observation and Interview Findings from the Ethnicity Questions and Antenatal Screening for Sickle Cell/Thalassaemia (EQUANS) Study. *Critical Public Health* 17 (1): 31–43. Dyson, SM; Cochran, F; Culley, LA; Dyson, SE; Kennefick, A; Kirkham, M; Morris, P; Sutton, F; and Squire, P (2007). And Presentation at Thalassaemia International Federation Third Pan European Congress, 25–26 October 2012, Dyson, SM. | <sup>379</sup> Ethnicity Screening Question and Ante-Natal Screening for Sickle Cell and Thalassaemia (EQUANS) in England: A randomized controlled trial of two questionnaires. *Ethnicity and Health* 11 (2): 169–189; Dyson, SM; Culley, LA; Gill, C; Hubbard, S; Kennefick, A; Morris, P; Rees, D; Sutton, F; Squire, P (2006). And Presentation at Thalassaemia International Federation Third Pan European Congress, 25–26 October 2012, Dyson, SM. | <sup>380</sup> Screening for Sickle Cell and Thalassaemia, Streetly A. et al., 2011 Update. | <sup>381</sup> The Care Quality Commission (CQC) is the independent regulator of healthcare, adult social care and primary dental care services in England, see annual report 2012, [http://www.cqc.org.uk/sites/default/files/media/documents/cqc\\_annualreport\\_2012\\_tagged.pdf](http://www.cqc.org.uk/sites/default/files/media/documents/cqc_annualreport_2012_tagged.pdf) | <sup>382</sup> Screening for Sickle Cell and Thalassaemia, op. cit. | <sup>383</sup> Screening for Sickle Cell and Thalassaemia, op. cit. | <sup>384</sup> Screening for Sickle Cell and Thalassaemia, op. cit. | <sup>385</sup> Screening for Sickle Cell and Thalassaemia, op. cit.



## PREVENTION AND SCREENING

of ethnicity or race. They conclude that “this example underscores the need for people living with sickle cell and thalassaemia to be accorded appropriate health services in Northern Europe”, because “sickle cell and thalassaemia are health issues, not ethnic minority issues”.<sup>377</sup> Screening has traditionally been carried out on the basis of ‘presumed ethnicity’, linked to skin colour and ‘commonsense’, with significant divergences in the practices by nurses and midwives. These criteria are considered to have failed to assess the risk of carrying genes associated with SCD and thalassaemia.<sup>378</sup> Busy schedules and low knowledge amongst nurses and midwives are reported as outstanding obstacles to accomplish adequate antenatal screening.<sup>379</sup>

Since 2006, newborn screening programmes for SCD and thalassaemia have been universally implemented. These programmes aim to detect affected newborns and ensure that early treatment can be instigated to reduce mortality and health complications.<sup>380</sup> The Care Quality Commission<sup>381</sup> and its predecessor, the Health Care Commission, also highlighted the importance of early access to testing in relation to quality maternity care.<sup>382</sup>

Current research focuses on genetic and prenatal non-invasive diagnosis.<sup>383 384 385</sup>

**“THERE IS A STRONG NEED TO PUT GREATER FOCUS AND MORE RESOURCES INTO PSYCHOLOGICAL CARE OF HAEMOGLOBINOPATHY PATIENTS IN THE UK.”**

Elaine Miller, UK Thalassaemia Society

## TREATMENT AND CARE

**The UK Thalassaemia Society (UKTS) has published standards for the Clinical Care of Children and Adults with Thalassaemia in the UK.**<sup>386</sup>

The guidelines address the management of thalassaemia services, core management standards, prevention and management of complications. Core management standards include: the initial management of the newly-diagnosed infant; the decision to start regular transfusions; red cell transfusion; iron load monitoring and treatment; psychosocial issues; acute clinical presentation in the treated patient; referral for consideration of bone marrow transplantation; surgery including splenectomy; transition from paediatric care and management of adults.

The NHS has also published standards and guidelines for clinical care of SCD during childhood.<sup>387</sup> The guidelines address issues such as outpatient care, transition to adult service, management of pain at home and in hospital and acute complications, psychological management and specific treatment. Guidelines further recommend that information is provided to patients and their families “in a culturally sensitive manner, respecting their dignity and individuality”. The cost of treatment is fully covered by the NHS and patients are entitled to at least a yearly check up with a haematologist in a centre specialised<sup>388</sup> in haemoglobinopathies. Professionals further explain that patients can adapt or review their treatment plan. According to healthcare professionals, the treatment and care situation for haemoglobinopathy patients in the UK is considered good compared to other European countries, although experts note differences in the level of quality care between highly populated and rural areas.

Health professionals note that with the increased life expectancy for patients with  $\beta$ -thalassaemia major, there is increasing expert focus on the complications of the disease and its treatment in later life, including bone disease and skeletal complications, such as osteoporosis, bone fractures and pain. Professionals point out that studies have concluded that despite available social support and access to care in the UK, the quality of life for children with thalassaemia major remains poor.

<sup>386</sup> Standards for the Clinical Care of Children and Adults with Thalassaemia in the UK, UKTS, 2008, <http://www.ukts.org/pdfs/awareness/standards/ukts-standards-2008.pdf> <sup>387</sup> Sickle cell disease in childhood standards and guidelines for clinical care, 2010, 2nd edition, <http://sct.screening.nhs.uk/getdata.php?id=11164> | <sup>388</sup> NHR Information Service, Number of Patients Registered at each Centre, [http://www.nhr.nhs.uk/docs/pdfs/InfoNo\\_Patients\\_By\\_Centre.pdf](http://www.nhr.nhs.uk/docs/pdfs/InfoNo_Patients_By_Centre.pdf)



## Country Factsheet: United Kingdom

### POLICY OUTLOOK

#### EXPECTED POLICY DEVELOPMENTS AND TIMELINE

The UK Department of Health held a public stakeholder consultation on the rare disease plan from February until May 2012. The results had not yet been published at the time of writing. However, experts expect that the rare disease plan will be adopted by the end of 2013.

According to government officials, one main challenge for the organisation of specialised haemoglobinopathy services is the lack of understanding of migration flows, the demographic changes connected to migration and their impact on the provision of services, e.g. where and how exactly services have to be provided.

**“OVER THE LAST 2 YEARS THERE HAS BEEN HUGE PROGRESS ON RAISING AWARENESS OF THE IMPORTANCE OF STRONG COMMISSIONING ARRANGEMENTS FOR SPECIALISED HAEMOGLOBINOPATHY SERVICES.**

**THE PUBLICATION OF THE NATIONAL HAEMOGLOBINOPATHY PROJECT IN 2011 AND THE FURTHER WORK OF THE NATIONAL CLINICAL REFERENCE GROUP SHOULD RESULT IN CONSISTENT COMMISSIONING DOCUMENTS BEING INCLUDED IN ACUTE SPECIALISED CONTRACTS FROM APRIL 2013. THESE WILL ALLOW US TO DESCRIBE NATIONALLY THE STANDARDS OF CARE PATIENTS CAN EXPECT OF EVERY PROVIDER.”**

Jon Currington, Acting Head of Strategy and Planning,  
Midlands & East Specialised Commissioning Group

#### INTERVIEWEES' SUGGESTED POLICY ACTIONS

- In the context of the current economic situation and cost containment measures, experts feel that there is not much room for further spending on the development of existing programmes on haemoglobinopathies. The situation of haemoglobinopathy patients in UK is already considered to be one of the best in Europe
- Patient organisations claim however that more spending for psychological care is absolutely crucial
- Healthcare professionals stress the importance of promoting specialised training and professional development opportunities in order to address challenges related to staff shortages
- Government officials feel that the reorganisation of commissioning offers opportunities to decrease costs and optimise healthcare resources in order to improve treatment and care for haemoglobinopathy patients

**“THE PROFILE OF GENETIC DISEASES HAS TO BE PRESENT ON THE POLITICAL AGENDA AND POLICY MAKERS SHOULD BE AWARE THAT SCD IS THE MOST COMMON GENETIC DISEASE IN THE UK.**

**WE NEED BETTER CAREER DEVELOPMENT OPPORTUNITIES FOR YOUNG DOCTORS AND RESEARCH SUPPORTED BY ADEQUATE FUNDING.**

**WE ALSO NEED A COMMISSIONING PROCESS FOR HAEMOGLOBINOPATHIES IN THE UK THAT ALLOWS PATIENTS ACCESS TO ADVANCES IN MEDICAL TREATMENT IN A TIMELY MANNER**

**EXPERT CLINICIANS NEED THE DISCRETION TO PRESCRIBE LICENSED DRUGS IN A MANNER CONSISTENT WITH INTERNATIONAL GUIDELINES FOR THE MANAGEMENT OF HAEMOGLOBINOPATHIES.”**

*John Porter, Department of Haematology, University College London*

## **Key Findings and Conclusions**

- **Prevalence and Burden of Haemoglobinopathies**
- **Policy Focus**
- **Prevention and Diagnosis**
- **Treatment and Care**
- **Haemoglobinopathies Report Summary of Indicators**



## Key Findings and Conclusions

## Prevalence and Burden of Haemoglobinopathies

01

**DESPITE BEING ORIGINALLY ENDEMIC IN THE MEDITERRANEAN, AFRICAN AND ASIAN REGIONS, HAEMOGLOBINOPATHIES ARE TODAY THE MOST COMMON GENETIC DISORDERS IN EUROPE.**

In countries in which haemoglobinopathies are endemic, they are not considered a rare disease (e.g. Cyprus).

02

**HAEMOGLOBINOPATHIES ARE CHRONIC LIFE-RESTRICTING AND LIFE-THREATENING CONDITIONS.**

The main causes of related deaths are linked to secondary sequelae such as heart failure due to iron overload, organ damage (notably liver) and serious infections. Thanks to both targeted prevention measures and treatment, life expectancy has nevertheless increased considerably and most haemoglobinopathy patients live from 45 years (average in Belgium and Cyprus) to 50 or more years (France, Spain and the Netherlands).

03

**HAEMOGLOBIN DISORDERS ARE PRESENT IN ALL EUROPEAN COUNTRIES,** with the number of patients varying from an estimated 150 cases in Sweden to approximately 10,500 in France.

04

**THERE ARE POOR DATA ON THE PRECISE PREVALENCE, OVERALL BURDEN AND TRENDS OF HAEMOGLOBINOPATHIES.**

This is linked to the general lack, or poor implementation of comprehensive data collection and analysis systems.

05

**EXPERTS HAVE LONG STRESSED THE IMPACT OF MOBILITY AND MIGRATION FLOWS ON THE NUMBER OF HAEMOGLOBINOPATHY PATIENTS IN MANY EUROPEAN COUNTRIES.**

However, accurate and comprehensive data on these important contributing factors are still missing in most countries. Data collection and analysis systems, if in place, do not comprehensively address relevant migration/ethnic origins of patients. Moreover, in some countries, patients' data protection and non-discrimination legislation pose additional challenges. International organisations, stakeholders and the EU have, however, called on national governments to allow for the disaggregation of data in order to effectively assess migrants' access to healthcare.

06

**GIVEN THE LACK OF COMPARABLE AND CONSISTENT DATA ON THE BURDEN AND TREND OF HAEMOGLOBINOPATHIES,** policy makers are unable to effectively assess and address the current and future challenges linked to these diseases.

07

**THERE IS STILL REPORTED RESISTANCE TO, AND DIFFICULTIES IN INTEGRATING DIFFERENT POLICY AREAS** into a single, holistic policy approach to health and migration.

08

**COST ESTIMATES ON THE FINANCIAL BURDEN OF NON-PREVENTION, LATE, INCORRECT OR NON DIAGNOSIS AND POOR QUALITY OF CARE OF HAEMOGLOBIN DISORDERS, AS WELL AS COSTS RELATED TO MEDICATION, TRANSPORTATION AND PRODUCTIVITY LOSS ARE GENERALLY NON-EXISTENT.** When cost estimates do exist, they are not systematically or comprehensively recorded.

### Main Needs and Improvements

ESTABLISH DEDICATED REGISTRIES/ DATABASES TO ENSURE ADEQUATE AND CONSISTENT COLLECTION AND ANALYSIS OF DATA, INCLUDING DISAGGREGATED DATA ON PATIENTS' RELIGIOUS BACKGROUND (TO UNDERSTAND THEIR BELIEFS ON KEY ISSUES SUCH AS PRENATAL SCREENING, ETC.), COUNTRY OF ORIGIN AND ETHNICITY.

## Key Findings and Conclusions

## Policy Focus

01

DESPITE INTERNATIONAL RECOGNITION AND CONSENSUS FOR GOVERNMENTS TO ADEQUATELY ADDRESS MIGRANT HEALTH AND HAEMOGLOBIN DISORDERS (E.G. 2006 WHO RECOMMENDATIONS AND WHO 2008 RESOLUTION ON HEALTH OF MIGRANTS ADOPTED BY ALL MEMBER STATES AT THE WHO ANNUAL WHA - WORLD HEALTH ASSEMBLY), TARGETED, COMPREHENSIVE POLICY MEASURES ARE NOT PRESENT ACROSS THE EU COUNTRIES. THERE ARE ONLY A FEW EXCEPTIONS.

02

ONLY CYPRUS, GREECE (BOTH ENDEMIC COUNTRIES) AND FRANCE HAVE SPECIFIC POLICY STRATEGIES OR PROGRAMMES IN PLACE TO ADDRESS HAEMOGLOBINOPATHIES, including education and awareness components as well as preventive, screening and holistic management measures.

03

SPECIFIC PLANS AND MEASURES THAT TAKE ACCOUNT OF CULTURAL PRACTICES AND THE GENERAL SOCIO-ECONOMIC CONTEXT OF AFFECTED OR AT-RISK POPULATIONS ARE FOUND ONLY OCCASIONALLY.

However, the measures are not duly implemented and do not include systematic tools to regularly measure the policy impact on the prevalence and the quality of care afforded to affected patients.

04

ALTHOUGH HAEMOGLOBIN DISORDERS ARE THE MOST COMMON GENETIC DISEASES, THEY REMAIN A SECONDARY PRIORITY FOR ACTION

and there is no particular reference or focus within rare diseases strategies or migrant health initiatives. Most countries provide a fragmented and vague policy framework for haemoglobinopathies under their national rare diseases plans, if a framework exists at all. Policy measures targeting migrants' access to healthcare remain exceptional, and policies addressing migrants' access specifically to rare diseases or haemoglobinopathies prevention and care are even rarer.

05

GENERALLY, THE EXISTENCE AND EFFECTIVE IMPLEMENTATION OF RELEVANT POLICY MEASURES DIVERGE ACROSS DIFFERENT REGIONS OF THE COVERED EU COUNTRIES. INEQUALITIES IN ACCESS TO HEALTHCARE FOR MIGRANTS AND MOBILE POPULATIONS REMAIN A CHALLENGE.

06

RELUCTANCE FROM GOVERNMENTS TO TAKE TARGETED ACTION TO ADDRESS HAEMOGLOBIN DISORDERS EFFECTIVELY MAY BE ATTRIBUTED TO: THE UNDERESTIMATION OF THE BURDEN OF THESE DISEASES AND INADEQUATE DISEASE MANAGEMENT;

cost-containment measures in healthcare budgets; and ethical, legal and social concerns of at-risk groups/patients towards screening and prevention of genetic diseases. Structured policy dialogue between the government and the relevant healthcare community as well as patients is limited and in most of the countries, inexistent.

### Main Needs and Improvements

- ENSURE HOLISTIC, LONG-TERM AND QUALITY CARE FOR ALL HAEMOGLOBINOPATHY PATIENTS REGARDLESS OF STATUS OR ORIGIN
- DEVELOP, ADOPT AND IMPLEMENT CULTURALLY COMPETENT DEDICATED, LONG-TERM PLAN AT NATIONAL LEVEL AS A MEANS TO IMPROVE PREVENTION AND DELIVERY OF CARE
- ESTABLISH REFERENCE CENTRES FOR HAEMOGLOBINOPATHIES

## Key Findings and Conclusions

## Prevention and Diagnosis

01

### SCREENING AND DIAGNOSIS.

Screening generally targets those considered at risk of being disease carriers (including with relevant migrant/ethnic origin). Screening and diagnosis practices vary widely across within the 10 countries covered by this report. In most cases, screening is not structured and comprehensively implemented within countries. Instead, screening is dependent on the knowledge and education of both healthcare professionals and patients and, therefore, screening tends to be carried out on a voluntary basis when doctors propose it and the patients agree to do it. There is low awareness and information about experts and centres of expertise in haemoglobinopathies throughout Europe.

02

### PRE-NATAL AND NEONATAL SCREENING.

Ethical, legal and social concerns, particularly related to pre-natal screening, and linked challenges continue to pose difficulties to implement adequate screening and in particular pre-natal screening. Although neonatal screening has proven to be one of the most effective prevention tool for severe forms of SCD, its practice, even though generally more widely available as compared to prenatal screening, remains inconsistent and limited across and within member states.

03

### HEALTHCARE PROFESSIONAL AWARENESS.

Prevention and diagnosis guidelines and protocols, when existing, are driven by specialist medical associations. However, awareness and knowledge of haemoglobin disorders amongst primary physicians and other specialists (other than haematologists) is considered low. Although they are the most common genetic disorders, education on haemoglobinopathies typically does not figure in general medicine curricula.

04

### CRITICAL FACTORS.

Language barriers and poor multicultural competences of healthcare staff and support teams are considered critical factors leading to under- or misdiagnosis, lack of patient adherence with treatment and low patient safety.

05

### LOW PUBLIC AWARENESS

Generally, there is low awareness amongst the general population about the risks of being a carrier, affected birth rates, associated health risks and sequelae and how to prevent and manage them effectively. There is a need for improved awareness raising activities specially targeted at populations at risk, including children.

06

### CURRENT BARRIERS.

Difficulties in reaching out to affected patients/potential carriers and in ensuring access to healthcare, linked to their mobility/migrant origin and cultural specificities are still considered one of the main barriers.

07

### LIMITED CAMPAIGNS.

Targeted education and awareness campaigns to prevent haemoglobin disorders amongst the general population are only implemented in a limited number of countries (Cyprus, Belgium). In most cases campaigns are mainly driven by patient support organisations (France, Netherlands, Spain and UK). Exceptionally, campaigns are shaped and run with the involvement of the broader community.

## Main Needs and Improvements

- INCREASE AWARENESS OF GENERAL PUBLIC AND OF 'AT-RISK' POPULATIONS
- PROVIDE TARGETED EDUCATION TO RAISE AWARENESS OF DELIVERY OF CARE AMONGST HEALTHCARE PROFESSIONALS
- FACILITATE ACCESS TO GENETIC COUNSELLING AS WELL AS PRECONCEPTION AND PRENATAL SCREENING FOR AT-RISK COUPLES
- DEVELOP NATION-WIDE PROTOCOLS TO ENSURE CONSISTENT SCREENING PRACTICES THAT PAY SPECIAL ATTENTION TO DIVERSITY (MOBILE AND MIGRANT AT RISK GROUPS)



## Key Findings and Conclusions

## Treatment and Care

## Social Support

01

### HETEROGENEITY IN CARE AND TREATMENT.

Care of patients with haemoglobin disorders is generally delivered by specialist doctors at hospital units free of charge, or it may be either fully or partly reimbursed by healthcare systems. However, timely, regular access to specialist care, innovative treatment and techniques for screening of sequelae is heterogeneous across and within countries. Healthcare cuts and specialist shortages are outlined as some of the main reasons for this heterogeneity.

02

### SPECIALISED CARE AND FOLLOW UP OF SEQUELAE.

Specialised care and follow up of sequelae is considered a crucial component in the effective management of the diseases and health complications and reducing mortality. Despite this, the approach to care is generally fragmented and coordination between specialists from different relevant disciplines is often unstructured, or is limited.

03

### GUIDELINES AND PROTOCOLS.

Guidelines and protocols are generally driven by specialist medical associations. However, professional awareness and effective implementation across the country appears to be inconsistent.

01

### PATIENT SUPPORT.

Psychological and professional support to help patients improve their quality of life and integrate into the labour market is considered scarce, fragmented and is often a forgotten component in the management of these diseases.

02

### FINANCIAL SUPPORT.

Financial support to affected patients is often provided through general support measures for people with disabilities when/if the patients qualify under national rules. However, certain additional costs linked to care are often not covered by general support schemes (e.g. regular displacements to specialised hospital units).

## Main Needs and Improvements

- INTRODUCE HAEMOGLOBINOPATHIES AND CULTURAL COMPETENCY IN CROSS-SPECIALTY PHYSICIANS' EDUCATION AND TRAINING AND INCREASE RECRUITMENT OF SPECIALISED PRACTITIONERS
- ENSURE UNIFORM ACCESS TO TREATMENT AND SPECIALISED CARE AS WELL AS COMPREHENSIVE FOLLOW-UP ACROSS DIFFERENT REGIONS AND COUNTRIES
- INCREASE FINANCIAL SUPPORT FOR RESEARCH INTO HAEMOGLOBINOPATHIES AND INNOVATIVE TECHNIQUES SUCH AS GENE THERAPIES AND STEM CELL TRANSPLANTATION
- DEVELOP EU-WIDE GUIDANCE AND FRAMEWORK ON STEM CELL TRANSPLANTATION

## Main Needs and Improvements

- IMPROVE PATIENT ACCESS TO CULTURALLY COMPETENT PSYCHOLOGICAL SUPPORT
- IMPROVE PATIENT ACCESS TO SOCIAL AND PROFESSIONAL SUPPORT

## Key Findings and Conclusions

## Haemoglobinopathies Report Summary of Indicators

COUNTRY	BELGIUM	CYPRUS	FRANCE	GERMANY	GREECE	ITALY	THE NETHERLANDS	SPAIN	SWEDEN	UK
PERCEIVED INFLUENCE OF MIGRATION ON EPIDEMIOLOGY	high influence	low influence traditionally, but increasing	high influence	high influence	low influence: disorders linked to indigenous population	low influence: disorders linked to indigenous population	high influence	high influence	high influence	high influence
NATIONAL /LOCAL THALASSAEMIA REGISTRY	no	national	national	no	national (on haemoglobinopathies)	regional (on haemoglobinopathies)	local	national scientific registry not official	no	yes
NATIONAL /LOCAL SCD REGISTRY	national	no	local	no	national (on haemoglobinopathies)	regional (on haemoglobinopathies)	local	national paediatric scientific registry	no	yes
STRUCTURED NATIONAL NEONATAL SCREENING PROGRAMME	local	no	national, targeted to the at risk population	no (trial Berlin)	yes	local/regional	yes	regional	no	yes
STRUCTURED NATIONAL ANTENATAL* SCREENING PROGRAMME	no rules	yes by the church (for the Greek community) and by civil authorities (for the Turkish community)	available	no	yes	local/regional	yes	no rules	no	yes
NATIONAL /REGIONAL RARE DISEASE PLAN EXPECTED BY	yes, work in progress	yes, approval planned in 2013	yes	yes, work in progress 2013	yes	yes, work in progress 2013	yes, work in progress 2013	yes	yes, work in progress 2013	yes, work in progress 2013
HAEMOGLOBINOPATHIES FOCUSED PLANS OR FOCUS IN RARE DISEASE PLANS	no	yes	yes	N/A	yes	N/A	N/A	no	N/A	yes
REIMBURSEMENT /FREE OF CHARGE TREATMENT	yes	yes	yes	partly	yes (for insured patients and asylum seekers)	yes (depending on the region)	yes	in general yes, some regions partially	yes	yes
COUNTRY AWARENESS	low	very good	low	very low	high to some extent only	generally low, high in specific regions	low	low	very low	relatively high

\* Before birth, including preconception and during pregnancy.

## Policy Recommendations

BASED ON THE KEY FINDINGS AND CONCLUSIONS OF THIS REPORT, POLICY ACTIONS AT EU AND NATIONAL LEVEL IN A NUMBER OF AREAS ARE URGENTLY REQUIRED.

## EU Policy Recommendations

EU INSTITUTIONS SHOULD:

01

**TAKE APPROPRIATE MEASURES TO ADDRESS COMPREHENSIVELY ACROSS THE EU GROWING CHALLENGES POSED BY HAEMOGLOBINOPATHIES, MIGRATION, MOBILITY AND TODAY'S EUROPEAN MULTICULTURAL SOCIETIES TO HEALTHCARE SYSTEMS, INCLUDING HEALTH INEQUALITIES.**

A practical measure would be to allocate explicit responsibility for these issues to a specific unit at the European Commission. The unit could be created and/or adapted to involve services/representatives from various relevant Commission Directorate Generals, such as DG Health and Consumers; DG Employment, Social Affairs and Inclusion; and DG Home Affairs. The European Union should also foster the identification, evaluation and sharing of best practices, and information on existing reference centres and expertise, across EU Member States as well as provide adequate support and coordination for the development of targeted national policies, awareness and education campaigns. The European Union Committee of Experts on Rare Diseases (EUCERD) provides a suitable platform to lead part of this work. Through a dedicated Working Group, EUCERD should look at how this report's recommendations can be best promoted and integrated into clinical practice across EU member states. This would include the identification, as per the criteria and work set by ENERCA in its White Book (in press 2013), of expert centres for haemoglobinopathies across Europe; targeted awareness and dissemination measures (in particular in Eastern countries); and supporting measures for the establishment of newborn screening programmes in countries where there is poor or no epidemiology information on haemoglobinopathies. These steps should be aligned with and embedded into other relevant initiatives, such as the European Programme of Newborn Screening and EU migration policies.

02

**PROVIDE FURTHER EVIDENCE AND SURVEILLANCE SYSTEMS TO ASSESS AND ADDRESS EFFECTIVELY HAEMOGLOBINOPATHIES AND, MORE GENERALLY, MIGRATION FLOWS AND THE SPECIFIC NEEDS OF TODAY'S MULTICULTURAL SOCIETIES.**

This should be done through the development and effective implementation of data collection and analysis systems, which provide for cross-country comparable data and standard indicators. These should cover haemoglobin disorders prevalence, linked mortality and health sequelae, as well as, with appropriate data protection measures, key patient data on migrant/ethnic origin to understand the epidemiology trends and therefore allow policy makers to shape and advance responsive, effective and evidence-based policy measures.

03

**ENCOURAGE AND SUPPORT EU MEMBER STATES IN IMPROVING CARE FOR ALL HAEMOGLOBINOPATHY PATIENTS,**

by establishing and improving centres of reference or expertise on haemoglobin disorders, identifying and supporting experts in the field as well as adapting healthcare delivery systems based on patient-centred and multicultural approaches which respond to the specific needs of haemoglobinopathy patients as well as migrant and ethnic minority groups. These specific needs include challenges linked to lack of information, cultural and linguistic barriers and socioeconomic situations of the affected populations, which are crucial to ensure quality prevention, diagnosis and care of haemoglobinopathies.

04

**ENCOURAGE MEMBER STATES TO DRAW INCREASED POLICY ATTENTION TO HAEMOGLOBINOPATHIES**

by adopting targeted programmes and addressing haemoglobin disorders in the framework of National Rare Diseases Plans and other policy measures to enhance migrants' access to appropriate healthcare.

05

**INCREASE STANDARDS OF CARE AND PREVENTION BY SUPPORTING THE DEVELOPMENT OF EU-WIDE GUIDELINES**

on holistic prevention and care of haemoglobin disorders, covering a range of issues involved, such as prevention and genetic counselling, diagnosis and care, multidisciplinary and culturally competent quality healthcare delivery standards.

06

**ACTIVELY SUPPORT EU MEMBER STATES IN ENHANCING HEALTHCARE PROFESSIONAL EDUCATION**

and training on haemoglobinopathies and multicultural care competences and addressing the challenges posed by healthcare staff shortages.

07

**PRIORITISE FUNDING FOR RESEARCH ON HAEMOGLOBINOPATHIES CARE**

(e.g. gene therapy) as well as dedicated policies in response to current and future migration and mobility flows, and centres of references on haemoglobin disorders.

## National Policy Recommendations

NATIONAL GOVERNMENTS SHOULD:

01

### DEVELOP AND IMPLEMENT TARGETED PROGRAMMES TO ADDRESS HAEMOGLOBINOPATHIES.

These programmes should aim to provide a tailored framework and holistic approach to the prevention and management of haemoglobinopathies, including appropriate data collection and analysis, awareness and education measures, establishment of centres of reference or expertise, prevention, quality diagnosis, long-term care, psychological, social and professional support to all haemoglobinopathy patients.

02

### ACCORDING TO THE SPECIFIC NATIONAL NEEDS, PROVIDE FOR ADEQUATE SPECIFIC MEASURES

to address haemoglobin disorders in the framework of National Rare Diseases Plans as well as policy measures to enhance migrants' appropriate access to health services.

03

### DEVELOP AND EFFECTIVELY IMPLEMENT DATA COLLECTION AND ANALYSIS SYSTEMS,

which provide for cross-country comparable data and standard indicators. These systems should cover haemoglobin disorders prevalence, linked mortality and health sequelae, as well as, with appropriate data protection measures, key patient data on migrant/ethnic origin to understand the epidemiology trends and therefore allow policy makers to shape responsive, effective policy measures.

04

### ESTABLISH CENTRES OF REFERENCE OR EXPERTISE,

and adapt and improve medical services to embed patient-centred and multicultural approaches along the patient pathways. These approaches would respond to the specific needs of haemoglobinopathy patients, ethnic minority groups and, where appropriate, second and third generations of migrants. The approaches would include specific measures to provide for adequate prevention and long-term, multidisciplinary care, psychological, social and professional support. Targeted measures should be designed to address relevant challenges linked to lack of information, cultural and linguistic barriers and socioeconomic situations of affected populations. For example, integration of staff of different ethnic origins within healthcare services, as well as translation and cultural mediation services.

05

### SUPPORT THE DEVELOPMENT AND IMPLEMENTATION OF GUIDELINES

and standards of care and prevention of haemoglobin disorders and linked sequelae. This includes prevention, genetic counselling, diagnosis and care, and multidisciplinary and multicultural quality healthcare delivery standards.

06

### SUPPORT THE EMPOWERMENT AND PARTICIPATION OF PATIENTS AND THE HEALTHCARE COMMUNITY

in shaping adequate policies that respond to the specific needs of haemoglobinopathy patients and multicultural healthcare delivery models. Civil society organisations representing migrant and ethnic minorities should also be supported.

07

### IMPLEMENT AND SUPPORT EDUCATION AND AWARENESS MEASURES TARGETED AT THE GENERAL PUBLIC, HAEMOGLOBIN PATIENTS AND POTENTIAL CARRIERS, INCLUDING DIFFERENT MIGRANT AND ETHNIC MINORITY GROUPS.

These measures should address general awareness of haemoglobin disorders, prevention and diagnosis, care, relevant available healthcare services and support, as well as ethical, legal and social issues linked to haemoglobinopathies. Effective information and education measures should be tailored to the different range of ethnic minority groups (including translated materials and/or picture/diagram-supported materials). Easily accessible information materials (e.g. through pamphlets or short movies) should be developed, specially targeted to couples and children and describing the most important aspects of haemoglobinopathies diagnosis and prevention.

08

### IMPLEMENT AND ACTIVELY SUPPORT TARGETED HEALTHCARE PROFESSIONAL EDUCATION AND TRAINING PROGRAMMES WITH A PARTICULAR FOCUS ON THE PREVENTION, DIAGNOSIS AND MANAGEMENT OF HAEMOGLOBINOPATHIES.

These should include multicultural care competences, to encourage and help healthcare professionals understand patients' history and background as part of quality prevention, diagnosis and care.

09

### ADOPT SPECIFIC MEASURES AIMED AT ADDRESSING HEALTHCARE STAFF SHORTAGES and linked challenges, to ensure patients' access to quality healthcare.

10

### FUND RESEARCH ON HAEMOGLOBINOPATHIES (e.g. gene therapy), as well as dedicated policies and centres of references on haemoglobin disorders.

## List of Consulted Organisations

COUNTRY	ORGANISATION
Belgium	<ul style="list-style-type: none"> <li>• HÔPITAL ERASME, BRUSSELS</li> <li>• ACTION DRÉPANOCYTOSE</li> <li>• INSTITUT NATIONAL D'ASSURANCE MALADIE-INVALIDITÉ (INAMI)</li> </ul>
Cyprus	<ul style="list-style-type: none"> <li>• CYPRUS THALASSAEMIA CENTRE</li> <li>• THALASSAEMIA INTERNATIONAL FEDERATION</li> <li>• PANCYPRIAN THALASSAEMIA ASSOCIATION</li> </ul>
France	<ul style="list-style-type: none"> <li>• CENTRE DE COMPÉTENCE, LABORATOIRE D'HÉMATOLOGIE, CHU DE MONTPELLIER</li> <li>• ASSOCIATION POUR L'INFORMATION ET LA PRÉVENTION DE LA DRÉPANOCYTOSE (A.P.I.P.D.)</li> <li>• MINISTÈRE DES AFFAIRES SOCIALES ET DE LA SANTÉ</li> </ul>
Germany	<ul style="list-style-type: none"> <li>• UNIVERSITY HOSPITAL ULM</li> <li>• UNIVERSITÄTSKLINIKUM HAMBURG-EPPENDORF</li> <li>• HELP FOR THALASSEMIA WITHOUT BORDERS</li> </ul>
Greece	<ul style="list-style-type: none"> <li>• LAIKO GENERAL HOSPITAL, ATHENS</li> <li>• GREEK THALASSEMIA FEDERATION</li> <li>• CENTRAL HEALTH COMMITTEE FOR THALASSEMIA</li> </ul>
Italy	<ul style="list-style-type: none"> <li>• UNIVERSITÀ DI VERONA</li> <li>• ASSOCIAZIONE TALASSEMICI E DREPANOCITI LOMBARDI (ATDL)</li> <li>• CENTRO DI COORDINAMENTO PER LE MALATTIE RARE (REGIONE LOMBARDIA)</li> </ul>
The Netherlands	<ul style="list-style-type: none"> <li>• PAEDIATRIC HAEMATOLOGIST, AMSTERDAM MEDICAL CENTRE</li> <li>• ERASMUS MEDICAL CENTRE, ROTTERDAM</li> <li>• OSCAR</li> <li>• MINISTRY OF HEALTH</li> </ul>
Spain	<ul style="list-style-type: none"> <li>• INSTITUTO DE INVESTIGACIÓN EN ENFERMEDADES RARAS, INSTITUTO DE SALUD CARLOS III</li> <li>• RED CELL PATHOLOGY UNIT, HOSPITAL CLÍNIC, UNIVERSITY OF BARCELONA</li> <li>• ASOCIACIÓN ESPAÑOLA DE LUCHA CONTRA LAS HEMOGLOBINOPATIAS Y TALASEMIAS (ALHETA)</li> </ul>
Sweden	<ul style="list-style-type: none"> <li>• AKADEMISKA SJUKHUSET, UPPSALA</li> <li>• SÖDERSJUKHUSET, STOCKHOLM</li> <li>• MINISTRY OF SOCIAL AFFAIRS</li> </ul>
United Kingdom	<ul style="list-style-type: none"> <li>• DEPARTMENT OF HAEMATOLOGY, UNIVERSITY COLLEGE LONDON</li> <li>• UK THALASSAEMIA SOCIETY (UKTS)</li> <li>• MIDLANDS &amp; EAST SPECIALISED COMMISSIONING GROUP</li> </ul>





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