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INFIRMARY

# SAVE THE DATE

North East England Metabolic Study Day  
hosted by Dr Mark Anderson, Consultant Paediatrician and Dr Ann Bowron,  
Consultant Clinical Scientist both from Newcastle upon Tyne Hospitals NHS  
Foundation Trust

VENUE : Education Centre, Royal Victoria Infirmary, Newcastle upon Tyne  
DATE : Wednesday March 8<sup>th</sup> 2017  
TIME : 0900 – 1630

RCPCH ACCREDITATION APPLIED FOR

The registration fee will be £25.00, to include refreshments and lunch.

To request a registration form please email Jacqui McAleer, JM Associates on  
[jmassociates1@me.com](mailto:jmassociates1@me.com)

An independent meeting with exhibition stands and support from :  
Alexion, BioMarin Europe Ltd, Orphan Europe (UK) Ltd, Sobi



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# North East England Metabolic Study Day

Wednesday March 8<sup>th</sup> 2017

Education Centre, Royal Victoria Infirmary, Newcastle upon Tyne

## PROGRAMME

<b>0900 – 0950</b>	<b>Registration, Coffee &amp; Exhibition Stands</b>	
0950 – 0955	Welcome	
0955 – 1040	Introduction to Inherited Metabolic Diseases	<i>Dr Jean Marie Saudubray (Paris)</i>
1040 – 1110	Dysmorphism as a presenting sign of genetic metabolic disease	<i>Dr Bernd Schwahn, (Willink Institute, Manchester)</i>
<b>1110 – 1130</b>	<b>Refreshments &amp; Exhibition Stands</b>	
1130 – 1200	Whole Exome Sequencing – a New approach to the Investigation of Inherited Metabolic Disease	<i>Professor Sir John Burn (Clinical Genetics, Newcastle University)</i>
1200 – 1230	Laboratory Investigation of Metabolic Disease	<i>Dr Ann Bowron, (Clinical Biochemistry, NUTH)</i>
1230 – 1300	Dietetic Management of Inherited Metabolic Diseases	<i>Sandra Adams &amp; Claire Nicol, metabolic dietitians, (Great North Children's Hospital, NUTH)</i>
<b>1300 – 1400</b>	<b>Lunch &amp; Exhibition Stands</b>	
1400 – 1430	Newborn Screening in the North East of England	<i>Dr Fiona Jenkinson (Clinical Biochemistry, NUTH)</i>
1430 – 1500	Paediatric Familial Hypercholesterolaemia	<i>Dr Mark Anderson (Great North Children's Hospital, NUTH)</i>
<b>1500 – 1520</b>	<b>Refreshments</b>	
1520 – 1550	Mitochondrial Disease: Clinical Presentation and Management	<i>Dr Bobby McFarland (Wellcome Trust Centre for Mitochondrial Research, Newcastle University)</i>
1550 – 1620	Mitochondrial Disease: Laboratory Investigation and Diagnosis	<i>Professor Rob Taylor (Wellcome Trust Centre for Mitochondrial Research, Newcastle University)</i>