



ACB SOUTHERN REGION AUTUMN MEETING



INHERITED METABOLIC RENAL DISEASE AND MEMBERS' PAPERS

MONDAY 16TH DECEMBER 2013

Roberts Lecture Theatre 421, Roberts Building, University College London, Torrington Place,
London, WC1E 7JE (www.ucl.ac.uk/maps?locationID=16)

- 9:00 – 9:30 Registration
- 9:30 – 10:15 Renal tubular physiology
Dr Stephen Walsh, Senior Lecturer in Experimental Medicine and Honorary Consultant Nephrologist, UCL Centre for Nephrology
- 10:15 – 11:00 Cystinosis in children
Dr William van't Hoff, Consultant Paediatric Nephrologist, Great Ormond Street Hospital for Children
- 11:00 – 11:30 Tea
- 11:30 – 13:00 Members' papers
- Evaluation of next generation sequencing for the genetic diagnosis of the Primary Hyperoxalurias (*Eleanor Bagg*)
 - Diffuse large B-cell lymphoma presenting with AKI and hypercalcaemia (*Emma Ashley*)
 - A case of ovarian cancer...or is it? (*Jennifer Hersey*)
 - Molybdenum cofactor deficiency – rapid diagnosis is essential (*Joe Bailey*)
 - An unusual case of hypomagnesaemia (*Becky Batchelor*)
 - Chilling out and flying high: aberrant results due to an unexpected precipitate (*Laura Hewitt*)
- 13:00 – 14:00 Lunch
- 14:00 – 14:45 Cystinuria – treating the disease and the patient
Dr Shabbir Moochhala, Consultant Nephrologist, UCL Centre for Nephrology
- 14:45 – 15:30 FTIR spectroscopy as a possible diagnostic tool: application to cystinuria and future prospects
Prof. Peter Rich, Professor of Bioenergetics, University College London
- 15.30 – 16:00 Tea
- 16:00 – 16:45 Primary hyperoxaluria
Dr Gill Rumsby, Consultant Biochemist, University College London Hospitals
- 16:45 – 17:00 Discussion and Members' papers awards

Cost: £25 (ACB members), £15 (retired ACB members), £40 (non-members)
This meeting includes refreshments and lunch. 5 CPD points approved

For further information contact nick.flynn@nhs.net, katie.hadfield@uclh.nhs.uk or
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Meeting sponsored by Orphan Europe.