NIHR Rare Disease Day Cambridge Evening Lecture

01 February 2018

There will be talks from CUH researchers working on childhood neurogenic diseases, mitochondrial disease and a CUH study investigating excessive growth to parts of the body.

The lecture will give people the opportunity to meet people from the rare disease community, hear the latest updates in rare disease and to talk to fellow patients and researchers.

The event will be in the clinical school at Cambridge University Hospitals (not far from the main entrance), 17.30-20.00. All are welcome to attend, book your free tickets or more information here.

Speakers:

- Professor David Rowitch, Professor of Paediatrics and Head of Department, University of Cambridge “Early detection and advanced therapy for childhood neurogenetic diseases in the UK”

- Professor Patrick Chinnery, Head of Department for Clinical Neurosciences “Mitochondrial Disease”

- David Rose, CRDN Volunteer and GOSH speaker living with ultra-rare Occipital Horn Syndrome “My Patient Journey and What Research Means to Me”

- Dr Victoria ER Parker, Consultant in Endocrinolgy and member of the Segmental Overgrowth Study at WT-MRC Institute of Metabolic Science (IMS), Addenbrooke’s Hospital presenting the results of their clinical trial of sirolimus in PIK3CA related overgrowth

- Professor Fiona Karet, Prof of Nephrology and Consultant in Renal Medicine at the School of Clinical Medicine, Cambridge whose team’s research explores kidney malfunction and mutations of genes in rare kidney conditions such as Gitleman Syndrome. They also study common inherited kidney disorders such as polycystic kidney disease.