Royal College of Physicians

**Dr Robin Lachmann Biography**

After qualifying in 1990, I split my time between medical training, in General (Internal) Medicine and Metabolic Medicine, and research, doing a PhD on herpes simplex virus mediated gene delivery to the brain and postdoctoral work on glycosphingolipid lysosomal storage disorders. I am currently one of two consultants in the Charles Dent Metabolic Unit where we look after over 1000 adult patients with a wide range of inherited metabolic diseases. I am Chair of the Metabolic Disorders Clinical reference Group and an elected member of the RCP Council.

**Presentation Summary**

A paediatrician seeing a baby with an altered level of consciousness always thinks about a metabolic intoxication, but in adults the only intoxicants which are routinely considered are drugs and alcohol. For most physicians, inherited metabolic diseases (IMDs) are esoterica which must be learnt for exams but have no relevance to everyday practice, where metabolic acidosis is either ketoacidosis or lactic acidosis, hypoglycaemia is reactive or iatrogenic and no one even measures ammonia. However, many IMDs are treatable and more and more patients now survive into adulthood. Moreover, there are attenuated forms of these diseases which only present in adulthood. This presentation will discuss when to suspect metabolic encephalopathy, how to diagnose the underlying condition and what treatment to give to prevent permanent neurodisability.