Royal College of Physicians

Professor Sir Doug Turnbull Biography

Director Wellcome Centre for Mitochondrial Research. The Wellcome Centre is focused on research to improve the lives of patients with mitochondrial disease. Professor Turnbull's focuses on understanding the molecular mechanisms underlying the neurological features in patients. With colleagues he is searching for new therapies for patients and he has been actively involved in work to prevent the transmission of mitochondrial DNA disease using an IVF technique called mitochondrial donation.

Director MRC/BBSRC Centre for Ageing and Vitality. Professor Turnbull has a major interest in understanding the basic mechanisms involved in human ageing with particular emphasis on the role of mitochondria. The MRC Centre is focused on understanding how these mechanisms are influenced by lifestyle interventions and studies aimed at promoting healthy ageing.

Lead for the NHS Highly Specialised Services for Rare Mitochondrial Services for Adults and Children. This service provides optimum care for patients with mitochondrial disease throughout the UK with Centres in Newcastle, London and Oxford. This service was built on the back of clinical and basic research and the Newcastle Centre reviews in excess of 800 patients per year. The service has developed care pathways and patient guidance that are used worldwide of the benefit of patients.

Presentation Summary

Mutations of mitochondrial DNA disease are a common cause of genetic disease. The clinical features and age of onset of these diseases is very variable making their diagnosis and management very challenging. Mitochondrial DNA is maternally inherited and preventing transmission of mutations is a very important for families. Whilst for some women current reproductive options are available to limit disease transmission, for others there is no other options apart from adoption or egg donation. The recent development of Mitochondrial Donation, a technique which transfer the nuclear DNA from the patient to a donor egg or early embryo, offers hope for these families with inherited mitochondrial DNA disease.