The UCD Academic Centre on Rare Diseases (ACoRD) was awarded formal centre status in June 2013. The focus of the centre is to investigate rare genetic diseases, particularly those affecting the Irish population and the Irish Traveller population.

The Centre’s aims are focused on the study of rare genetic diseases, with a view to the identification of the mutation(s) causing the disease. Once a causal mutation(s) is established, the objective is to develop diagnostic tests for translation back into a clinical setting. Once a gene is implicated, our PIs work to further investigate the gene function and biological pathways involved in the condition. The ultimate aim is to investigate those conditions / genes which might be amenable to drug targeting or gene therapy.

Rarely in a lifetime does a scientific or medical field of research ‘come of age’. The revolution that was the ‘Human Genome Project’, coupled with the latest technological advances in genomics is set to transform the field of rare genetic diseases. An ad hoc group of UCD based clinicians, scientists, specialists in bioinformatics and cell biologists have long since recognised these developments, and the rare disease group – prior to its establishment as a centre – has achieved considerable national and international recognition.

Rather than employing the traditional approach of a large, disease-specific research group which focuses on a common disorder, our PIs focus on rare genetic disorders and utilise new tools from the genomics revolution to aid our study of common and rare disorders. We pursue an integrated approach to our work, which involves close collaboration across clinical and research teams. In Ireland there are about 285,000 individuals with a rare disease. In addition, there are approximately 60 identified recessive disorders in the Traveller population. In a pilot study we have completed data analysis on six of ten rare disorders of unknown genetic basis, affecting 25 small Irish families. Of the six studies, the disease mutation has been successfully identified for five families, of which three studies have been published to date, with four translated back into the clinical setting. This translational success demonstrates our ability to identify rare disease genes in small families.

We have recently consolidated our various working groups under one recognisable centre to harness the successful outputs and future studies for UCD. Ireland is required to submit a National Plan for Rare Diseases by 2013. A UCD academic centre on rare diseases is uniquely positioned to contribute to this plan. The centre aims to make a meaningful contribution in the progression of gene discovery to diagnostics, and ultimately in the cure or prevention of serious genetic conditions.
I am interested in the application of Genomics to genetic diseases. The main focus of my group is to make a meaningful contribution to the field of Human genetics. I am particularly interested in contributing to the progression of gene discovery to diagnostics, and ultimately to the cure or prevention of serious genetic conditions. I have been involved in establishing international collaborative approaches to the study of the genetics of Autism Spectrum Disorder (ASD) and rare genetic diseases.

My main laboratory based research is in Hereditary Spastic Paraplegia (HSP), a group of rare inherited neurodegenerative disorders. In collaboration with St Vincent’s University Hospital we have been performing genotype phenotype correlations and identifying and characterising novel causative loci. We have been studying the molecular mechanisms involved in this form of neurodegeneration. I am also interested in optimizing methods of educating medical professionals on rare genetic disorders.

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My main interest is the research and clinical application of new genetic technologies in human disease, specifically the genetics of tuberous sclerosis, the genetics of autism, and genetic diseases in the Irish Traveller population. I also have involvement in medical ethics, and am chair of the Irish National Advisory Committee on Bioethics. I was a member of the Irish Council for Bioethics, the Commission for Assisted Human Reproduction and local and national bioethics committees.

My research interest has always focused on the causation of neurological disorders in children in the broad sense and recently has focused on three areas: 1) Risk factors in neonatal hypoxic ischaemic encephalopathy 2) The molecular genetics of severe undiagnosed early onset epileptic disorders and Landau Kleffner syndrome (an older age dependent epileptic encephalopathy) 3) Movement disorders : novel genotype-phenotype associations. This research involves collaboration with researchers at UCD (SMMS) Mater and Rotunda Hospitals and internationally.

For more information about the work of UCD researchers working in the area of Rare Diseases, please visit the School’s award-winning website, available at www.ucd.ie/medicine.
10. Anim Genet 2012.


