About the RKD and vasculitis Registry.  This is an integrated resource across Tallaght Hospital, St James’s Hospital and Trinity College, and in collaboration with other units in the country. It is funded by Science Foundation Ireland, Dublin Centre for Clinical Research and Trinity College Medical Day. Kidney failure as a whole is a major focus of the Irish healthcare system, accounting for some 10% of the healthcare budget; individually, about 1/3 of these patients with kidney failure have developed this because they suffer from a “rare kidney disease”. People with rare diseases (those with a prevalence <5 in 10,000) are served poorly by health providers. As most doctors see few cases of a given rare disease in their career, significant delays in diagnosis are usual, with patients receiving treatment for other conditions before a unifying diagnosis is made. Even then, care is fragmented and poorly co-ordinated. 80% of rare diseases are genetic in origin affecting multiple organ systems, requiring input from multiple specialties on multiple hospital sites, which often lack the specific expertise to deal with these unusual conditions.

Research into rare diseases is challenging. Even large centres will see very few patients with a given rare disease, so it is virtually impossible to develop a sufficiently large cohort for meaningful study. To overcome this problem, it is necessary to study a large number of these patients and assess the long term effects of various treatments.

To share expertise and resources, networked collaboration between units is essential. For a country the size of Ireland, a national network is of the right size and allows study of epidemiology across the whole country. Such a network would revolve around a robust patient registry, with capture of detailed longitudinal clinical data across multiple units and linked collection of biological samples. Our goal is to provide the infrastructure to allow study of these rare diseases.

C3 glomerulopathy, a novel disorder of the complement pathway

This is a recently introduced umbrella term to describe a group of conditions characterised by kidney (glomerular) damage with significant deposition of the complement component C3. The variability that has been observed in kidney biopsy findings, clinical outcomes and genetic abnormalities between patients with different forms of C3 glomerulopathy is poorly understood. In collaboration with the Centre for Complement & Inflammation Research in Imperial College, Prof Peter Conlon, Dr Michelle O’Shaughnessy and Dr Lynn Redehan aim to screen identified Irish patients for genetic variations which may be responsible for the patient’s kidney impairment. Samples deriving from this project are being archived in the RKD biobank.

International registry of systemic vasculitis: UKVAS

The UKVAS registry is a component of UKIVAS, the Vasculitis Rare Disease Working Group of the UK and Ireland. It’s aims are to create a database of patients with systemic vasculitis attending centres across the British Isles and to archive longitudinal clinical data to build up a picture of clinical course over time. The long term vision is to link these data to carefully processed clinical samples, something that is already occurring in Ireland. Prof Little is co-founder and co-chair of this Kidney Research UK funded UK initiative.

Vasculitis affects about 50 per million population per year, as opposed to affecting a handful of people in the country, so is considered a “common-rare disease”. It causes severe multi-organ dysfunctions, including irreversible kidney failure, lung haemorrhage, stroke and sino-nasal destruction.

International vasculitis network

UKVAs

We intend implementing and developing the UKVAS registry in Ireland. Although the RKD registry will focus on the Irish population, the systemic vasculitis component of this will be designed to link seamlessly with the UKVAS registry. Irish patients will thus be included in a registry containing in excess of 2000 patients.

www.ukvas.org

Genetic kidney diseases: centralised study of “Rare-Rare” disorders

In those with “rare kidney disease”, some may have several affected first degree family members, but the diagnosis remains unknown or the condition undescribed. Such families may be the only cases in the country. This type of “rare-rare” kidney disease can only be studied through an infrastructure dedicated to genetic investigation of sporadic cases through a centralised clinic, which is set up to network with other centres around the world, thereby developing sufficiently large kindred sets to permit identification of the causative genetic mutation. This service will be provided through the Rare Kidney Disease Clinic at Tallaght hospital, and via the RKD registry, under the guidance of Dr Peter Lavin.
The study is designed as a National Registry for “Rare Kidney Diseases” and will include patients with systemic vasculitides, hereditary kidney disease and other rare diseases. The criterion for inclusion is a point prevalence of <5/10,000. The Registry aims to compile sufficiently large cohorts to facilitate:

- Testing of new therapies and conduct of phase II/III interventional studies by allowing easy identification of a suitable cohort which would not otherwise be possible
- Monitoring of expensive biologic use
- Study of genetic factors, identification of susceptibility genes and proteins contributing to disease.
- Service delivery research informing development of a care model that improves on the current fragmented approach.
- Addressing fundamental questions about epidemiology
- Rapid assessment of the clinical utility of new biomarkers for development in clinical trials as surrogate end-points.
- Characterisation of ‘difficult to define’ disease subgroups, including ANCA negative vasculitis and polyarteritis overlap syndromes.

Aims of the RKD and Vasculitis registry:

Since Sep 2012 we have registered over 100 patients onto the registry, well ahead of target. Samples are processed using at each site and archived centrally at the TCD biobank. Patient recruitment occurs via the Rare Kidney Disease research clinic in Tallaght Hospital, and at the Nephrology, Immunology and Rheumatology clinics detailed below left. Current active sites are Tallaght, St James’s and Beaumont Hospitals. St Vincent’s and Cork Hospitals are awaiting ethical approval, and Galway University Hospital is seeking research nurse funding.

Sites Involved, Recruitment & Sample Collection

We would like to Thank our Funding Organisations:

Principal Contacts

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