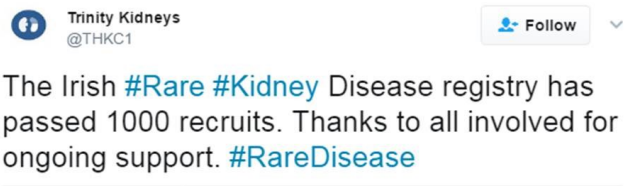
The Rare Kidney Disease Registry and Biobank

Newsletter - Spring 2017

The RKD and Vasculitis Registry and Biobank continues to provide an integrated resource to facilitate vital research. Working closely with hospitals and research centres across the country, the service brings together a large cohort of patient data and biological samples. This information is used by both national and international research groups to achieve greater understanding of rare kidney disease, which in turn leads to better patient care and treatment .

2017 - A Milestone Year for RKD

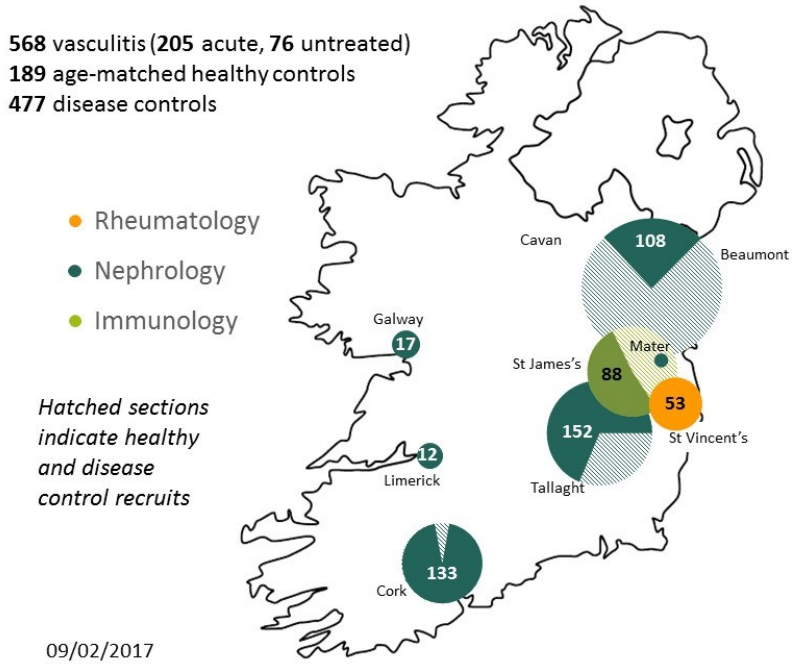
Since it's inception in 2012, the RKD Registry and Biobank has grown to be one of the largest such repositories of clinical data and biological samples in the world. This year marked the recruitment of the 1000th patient and this has continued past 1300. The link between the vasculitis and familial kidney disease genomics groups under the RKD umbrella has strengthened throughout 2016.



Recruitment to Date

The Meath Foundation-funded national recruitment strategy, ably supported by a highly effective Whatsapp group, has dramatically enhanced recruitment such that most new diagnoses of systemic vasculitis in major Irish centres are now being recruited, with most sampled before treatment.

The map below depicts the current number of recruits to the RKD Registry and Biobank (many have multiple longitudinal samples at different stages of the disease).

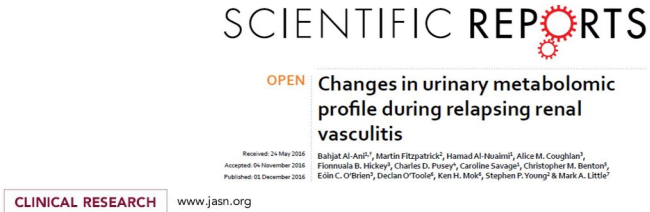


Key Research Supported in 2017

- ANCA vasculitis cyclophosphamide pharmacogenomics, in collaboration with Dr A Vaglio University Hospital of Parma, Italy.
- Urinary metabolomic biomarkers in Vasculitis - a collaboration with Agilent Technologies and Dr Ken Mok (TCD) using high throughput mass spectroscopy techniques.
- Genome Wide Association Studies (GWAS) in collaboration with the European Vasculitis Genetics Consortium:
  - PR3 ANCA vasculitis
  - MPO ANCA vasculitis
  - Eosinophilic granulomatosis with polyangiitis
  - Rituximab response
- C3 Glomerulopathy in collaboration with Prof Peter Conlon (Beaumont) and Imperial College, London.
- Familial Spastic Paraparesis and Chronic Kidney Disease in collaboration with Duke University, North Carolina.
- Familial Kidney Disease Genomics - an ongoing collaboration with Prof Peter Conlon and Dr Dervla Connaughton (Beaumont Hospital), and Dr Friedhelm Hildebrandt (Harvard Medical School, Boston Children's Hospital).
- Mechanisms of immunothrombosis in ANCA Vasculitis with Dr Roger Preston (RCSI)

Impactful research

Studies supported by the RKD Registry and Biobank continue to generate important new knowledge in the field of rare kidney disease research. The establishment of a biomarker discovery pipeline has led to the licensing of the urine sCD163



Urinary Soluble CD163 in Active Renal Vasculitis

Vincent P. O'Reilly,* Limy Wong,* Claire Kennedy,* Louise A. Elliot,† Shane O'Meachair,‡ Alice Marie Coughlan,* Eoin C. O'Brien,* Michelle M. Ryan,* Diego Sandoval,* Emma Connolly,* Gerjan J. Dekkema,§ Jiaying Lau,* Wayel H. Abdulahad,¶ Jan-Stephan F. Sanders,§ Peter Heeringa,¶ Colm Buckley,** Cathal O'Brien,††‡‡ Stephen Finn,††‡‡ Clemens D. Cohen,§§ Maja T. Lindemeyer,§§ Fionnuala B. Hickey,* Paul V. O'Hara,* Conleth Feighery,† Sarah M. Moran,* George Mellotte,* Michael R. Clarkson,‖‖ Anthony J. Dorman,** Patrick T. Murray,¶¶ and Mark A. Little*

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Welcomes and Farewells

The past year saw the arrival of Dr Barbara Fazekas and Dr Dearbhaile Dooley to support the Biobank. We are extremely grateful to Barbara for her contribution to co-ordinating sample delivery and processing when required.

We also saw the departure of Alice Coughlan. We thank her for her invaluable contribution and wish her well with her new post.

RITA

Rare Immunodeficiency auToinflammatory Autoimmune

European Reference Network

Vilnius, Lithuania was the location for the unveiling of the successful **European Reference Network** applicants on 9th March 2017. These networks have the aim of harmonising care for patients with rare diseases, hitherto left to fight their way through health services ill-equipped to deal with their needs. The **Vasculitis Ireland Network** designated centre of expertise was represented by Prof Mark Little, who is a co-coordinator of the Rare Immune Disorders ERN, "RITA".

Underpinned by a €1m grant from EU-CHAFEA, this is focused on rare immunodeficiency, autoinflammatory and autoimmune disorders, the autoimmune strand having been developed and managed by Prof Little. The inaugural committee meeting in Vilnius provided the opportunity to begin shaping how the chosen 23 European centres of expertise in rare immune disorders will cooperate to facilitate development of patient centric registries, common care pathways, transnational telemedicine solutions and new collaborative research opportunities. The RKD registry is a cornerstone of the Irish approach to this rare disease; RITA provides an opportunity for mainstreaming of RKD into the HSE.

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Thanks

To all patients and control individuals who have so kindly taken part. To all consultants for generously allowing access to their patients. To registrars Paul O'Hara, Claire Kennedy, Sarah Moran, Dervla Connaughton, Adrian Whelan, Heather Gunning, Tammy Wanigasekera, Limy Wong, Dearbhla Kelly, Yvelynne Kelly, Radzi Rodzlan, Conor Judge, Amy Hudson, Susie Murray, Louise Ryan and Mark Canney who help us recruit extremely valuable acute patients. To John McCourt and Conor Judge for database support. And to Joe McPartlin and the TCD Biobank for facilitating biobank freezer storage.