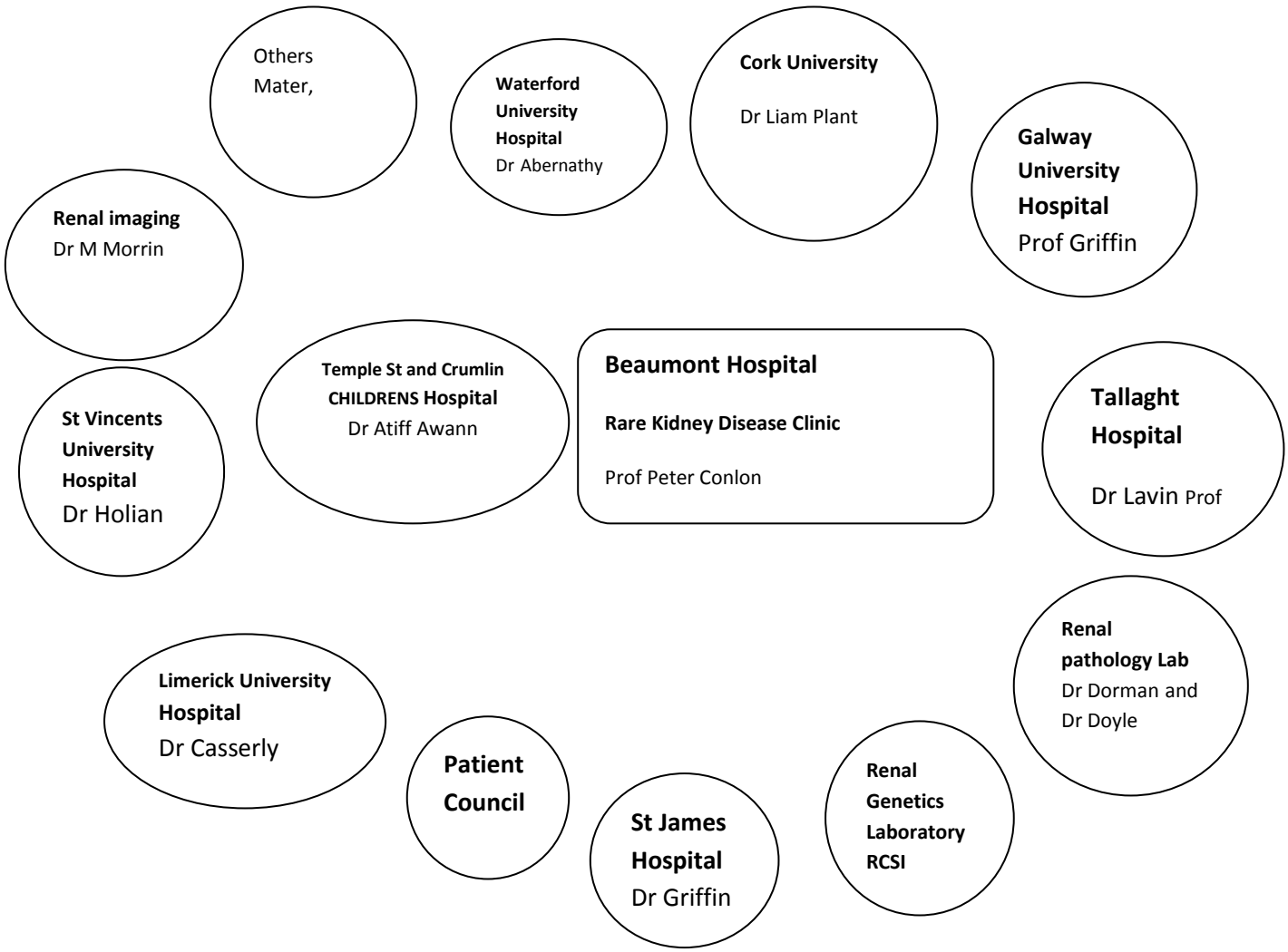
Irish Rare kidney Disease Network (IRKDN)



MISION

- Provision of care to patients with Rare Kidney Disease based on best available medical evidence through collaboration within Ireland and Europe
- Making available clinical trials for rare kidney disease to Irish patients where available
- Collaboration with other centres in Europe treating rare kidney disease
- Education of Irish nephrologists on rare Kidney Disease.
- Ensuring a seamless transition of children from children's hospital with rare kidney disease to adult centres with sharing of knowledge of rare paediatric kidney disease with adult centres
- The provision of precise molecular diagnosis of patients with rare kidney disease

- The provision of therapeutic plan based on understanding of molecular diagnosis where available
- Development of rare disease specific registries within national renal It platform (Emed)

Structure

Beaumont Hospital will act as National rare Kidney Disease Coordinating centre working in conjunction with a network of Renal unit across the country including .

- Dr Atiff Awann Temple St Children's Hospital Crumlin Childrens Hospital Soon to become National Childrens Hospital at St James'
- Dr Peter Lavin Tallaght Hospital
- Prof Mark Little Trinity Health representing Vasculitis Network
- Prof Yvonne O'Meara Mater Hospital and University College Dublin
- Dr John Holian, St Vincent's University Hospital Dublin
- Dr Liz Abbernathy Waterford Regional Hospital Waterford
- Prof Liam Plant , Cork University Hospital Cork
- Dr Liam Casserly, Limerick University Hospital, Limerick
- Prof Donal Reddin, Galway University Hospital Galway

Each member of the network will develop a presence on ORPHANNET https://www.orpha.net/consor4.01/www/cgi-bin/Clinics_Search.php?lng=FR&data_id=122949&Centres%20experts=Family-Kidney-Disease-Clinic&title=Family-Kidney-Disease-Clinic&search=Clinics_Search_Simple

Describing the disease and therapeutic areas where they are experts in. Where expertise in Ireland is lacking we will engage through facilities at Beaumont to obtain expertise from specialists centres in Europe.

Irish Rare Kidney Disease Network Board

A board to govern the Irish Rare Kidney Disease network will be developed to compose of each of the above representatives and in addition two patient representatives. The board will meet 3 times a year to review progress of the rare kidney disease network and to provide input into operational issues and future developments

EMED as a tool to provide comprehensive registry of rare kidney disease

Ireland has a national shared medical record (Emed) on which all clinical records for all patients with medical renal disease is maintained. This system is available in the clinic rooms and dialysis units across the country and contains medical records on more than 20000 patients with kidney disease. Emed provides an ideal platform for the further development of rare disease specific registries

Teaching Objectives

- Deliver a annual course of lectures to Specialist Registrars in training and Nurses in training on rare kidney disease.
- Contribute to the Royal college of Surgeons in Ireland medical school module on rare disease on aspects related to rare kidney disease.
- Promote rare kidney disease as a subject for MD and PhD students in Ireland
- Provide education to patients and parents about rare kidney disease both threouh our patient contacts and through the Irish Kidney Association (patient advocacy association)

Services available at constituent members of rare Kidney Disease network

Beaumont Hospital Rare Kidney Disease Centre

Rare Kidney Disease that we have Expertise in managing

See appendix 1

Services available to Patients with Rare Kidney Disease at Beaumont Hospital

- Comprehensive team of six consultant Nephrologists with extensive experience of all aspects of kidney disease including rare kidney disease together with a full team of nursing and paramedical staff able to deal with patients presenting with kidney disease

- Comprehensive team of 10 consultant Urologists/ transplant surgeons. This team of highly experienced consultant urologist are capable of dealing with all of the common and rare urological conditions and delivers a renal transplant service of almost 200 live and deceased donor kidney transplants annually
- Acute and chronic dialysis service/. The unit deliver dialysis therapy to in excess of 300 dialysis patients on an annual basis. The unit has access to acute inpatient, outpatient chronic dialysis home haemodialysis and peritoneal dialysis facilities. The unit has a comprehensive team in nursing, technical and other staff to run these facilities
- In patient wards The unit has access to more than 50 inpatient beds to deliver inpatient care of patients with rare kidney disease including a 24 hour a day , 365 day a year service.

- Renal pathology service. The department has access to a fully equipped renal pathology laboratory able to deliver light, immunoflouresence and electro micrographic examination of kidney biopsies. All biopsies are reviewed by a multi-disciplinary team on a weekly basis. The two renal Pathologists are Prof Tony Dorman and Dr Brendan Doyle

- Renal Radiology Service: Beaumont hospital has all of the necessary radiology modalities to deal with rare kidney disease including renal Ultrasound, CT scanning, nuclear medicine, Magnetic resonance imaging, all aspects of interventional radiology, These services are led by Prof Martina Morin

- Biochemistry Laboratory. Beaumont hospital has a fully equipped biochemistry laboratory capable of rapidly turning around all of the necessary blood and urine biochemistry tests for the evaluation of patients with kidney disease
- Immunology and haematology Laboratory; The hospital has access to a renal immunology laboratory that can deliver rapid turnaround of renal immunology tests including ANCA , Anti-GBM, antiPLRA2, compliment levels and protein electrophoresis.

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Appendix 1: Rare Kidney Disease

Renal or urinary tract malformation					
	Non-syndromic renal or urinary tract malformation				
		Multicystic dysplastic kidney			
			Unilateral multicystic dysplastic kidney		
			Bilateral multicystic dysplastic kidney		
		Exstrophy-epispadias complex			
			Epispadias		
			Cloacal exstrophy		
			Bladder exstrophy		
		Congenital hydronephrosis			
		Renal tubular dysgenesis			
			Renal tubular dysgenesis due to twin-twin transfusion		
			Drug-related renal tubular dysgenesis		
			Renal tubular dysgenesis of genetic origin		
		Medullary sponge kidney			
		Oligomeganephronia			
		Duplication of urethra			
		Congenital primary megaureter			
			Primary megaureter, adult-onset form		
			Congenital primary megaureter, obstructed form		
			Congenital primary megaureter, refluxing form		
			Congenital primary megaureter, nonrefluxing and unobstructed form		
		Renal hypoplasia			
			Renal hypoplasia, unilateral		
			Renal hypoplasia, bilateral		
		Renal dysplasia			
			Renal dysplasia, unilateral		
			Renal dysplasia, bilateral		
		Congenital megacalycosis			
			Unilateral congenital megacalycosis		
			Congenital bilateral megacalycosis		
		Megacystis-megaureter syndrome			
		Renal agenesis			
			Renal agenesis, bilateral		
			Renal agenesis, unilateral		
		Fetal lower urinary tract			

		obstruction			
			Prune belly syndrome		
			Atresia of urethra		
			Posterior urethral valve		
			Anterior urethral valve		
		Congenital urachal anomaly			
			Urachal cyst		
			Patent urachus		
			Urachal sinus		
			Urachal diverticulum		
	Syndromic renal or urinary tract malformation				
		BOR syndrome			
		Cystic hamartoma of lung and kidney			
		Megacystis-microcolon-intestinal hypoperistalsis syndrome			
		Renal coloboma syndrome			
		Ochoa syndrome			
		NPHP3-related Meckel-like syndrome			
Glomerular disease					
	Basement membrane disease				
		Alport syndrome			
			X-linked Alport syndrome		
			Autosomal dominant Alport syndrome		
			Autosomal recessive Alport syndrome		
		X-linked diffuse leiomyomatosis-Alport syndrome			
		HANAC syndrome			
		Nephrotic syndrome-deafness-pretibial epidermolysis bullosa syndrome			
		Junctional epidermolysis bullosa with respiratory and renal involvement			
	Secondary glomerular disease				
		Anti-glomerular basement membrane disease			
		Collagen type III glomerulopathy			
		Hereditary amyloidosis with primary renal involement			
			AApoAI amyloidosis		
			ALys amyloidosis		

			AFib amyloidosis		
			AApoAII amyloidosis		
		Immunotactoid or fibrillary glomerulopathy			
		Pauci-immune glomerulonephritis			
		AApoAIV amyloidosis			
		AH amyloidosis			
		IgG4-related kidney disease			
	Primary glomerular disease				
		Nail-patella syndrome			
		Schimke immuno-osseous dysplasia			
		Galloway-Mowat syndrome			
		Nail-patella-like renal disease			
		Pierson syndrome			
		Denys-Drash syndrome			
		Frasier syndrome			
		Primary membranoproliferative glomerulonephritis			
			Immunoglobulin-mediated membranoproliferative glomerulonephritis		
			Non-immunoglobulin-mediated membranoproliferative glomerulonephritis		
				Dense deposit disease	
				C3 glomerulonephritis	
		Congenital membranous nephropathy due to maternal anti-neutral endopeptidase alloimmunization			
		Fibronectin glomerulopathy			
		Autosomal dominant intermediate Charcot-Marie-Tooth disease type E			
		Congenital and infantile nephrotic syndrome			
			Congenital nephrotic syndrome, Finnish type		
			Familial steroid-resistant nephrotic syndrome with sensorineural deafness		
			LAMB2-related infantile-onset nephrotic syndrome		
			Familial steroid-resistant nephrotic syndrome with adrenal insufficiency		
		Idiopathic membranous glomerulonephritis			
		Action myoclonus-renal failure syndrome			

		MYH9-related disease			
		Rapidly progressive glomerulonephritis			
		Lipoprotein glomerulopathy			
		Idiopathic nephrotic syndrome			
			Familial idiopathic steroid-resistant nephrotic syndrome		
				Familial idiopathic steroid-resistant nephrotic syndrome with focal segmental hyalinosis	
				Familial idiopathic steroid-resistant nephrotic syndrome with diffuse mesangial proliferation	
				Familial idiopathic steroid-resistant nephrotic syndrome with minimal changes	
				Familial idiopathic steroid-resistant nephrotic syndrome with diffuse mesangial sclerosis	
			Idiopathic steroid-sensitive nephrotic syndrome		
				Idiopathic steroid-sensitive nephrotic syndrome with focal segmental hyalinosis	
				Idiopathic steroid-sensitive nephrotic syndrome with minimal change	
				Idiopathic steroid-sensitive nephrotic syndrome with diffuse mesangial proliferation	
			Sporadic idiopathic steroid-resistant nephrotic syndrome		
				Sporadic idiopathic steroid-resistant nephrotic syndrome with focal segmental hyalinosis	
				Sporadic idiopathic steroid-resistant nephrotic syndrome with diffuse mesangial sclerosis	
				Sporadic idiopathic steroid-resistant nephrotic syndrome with minimal changes	
				Sporadic idiopathic steroid-resistant nephrotic syndrome with diffuse mesangial proliferation	
				Sporadic idiopathic steroid-resistant nephrotic syndrome with collapsing glomerulopathy	

Thrombotic microangiopathy					
	Atypical hemolytic-uremic syndrome				
		Atypical hemolytic-uremic syndrome with C3 anomaly			
		Atypical hemolytic-uremic syndrome with MCP/CD46 anomaly			
		Atypical hemolytic-uremic syndrome with B factor anomaly			
		Atypical hemolytic-uremic syndrome with H factor anomaly			
		Atypical hemolytic-uremic syndrome with I factor anomaly			
		Atypical hemolytic-uremic syndrome with anti-factor H antibodies			
		Atypical hemolytic-uremic syndrome with thrombomodulin anomaly			
		Atypical hemolytic-uremic syndrome with DGKE deficiency			
	Methylcobalamin deficiency type cblG				
	Thrombotic thrombocytopenic purpura				
		Congenital thrombotic thrombocytopenic purpura			
		Acquired thrombotic thrombocytopenic purpura			
	Typical hemolytic-uremic syndrome				
Familial cystic renal disease					
	Autosomal recessive polycystic kidney disease				
	Tuberous sclerosis complex				
	Autosomal dominant polycystic kidney disease				
	Hepatic fibrosis-renal cysts-intellectual disability syndrome				
	Adult familial nephronophthisis-spastic quadriparesia syndrome				
	Neonatal diabetes-congenital hypothyroidism-congenital glaucoma-hepatic fibrosis-polycystic kidneys syndrome				
	Autosomal dominant polycystic kidney disease type 1 with tuberous sclerosis				
	Familial juvenile hyperuricemic nephropathy type 1				
	Renal-hepatic-pancreatic				

	dysplasia				
	Karyomegalic interstitial nephritis				
	Ventriculomegaly-cystic kidney disease				
Nephropathy secondary to a storage or other metabolic disease					
Rare renal tubular disease					
	Oculocerebrorenal syndrome of Lowe				
	Cystinuria				
		Cystinuria type A			
		Cystinuria type B			
	Bartter syndrome				
		Infantile Bartter syndrome with sensorineural deafness			
		Antenatal Bartter syndrome			
		Classic Bartter syndrome			
		Bartter syndrome with hypocalcemia			
	Joubert syndrome with oculorenal defect				
	Gitelman syndrome				
	Alström syndrome				
	Nephrogenic diabetes insipidus-intracranial calcification syndrome				
	Senior-Loken syndrome				
	Nephronophthisis				
		Late-onset nephronophthisis			
		Infantile nephronophthisis			
		Juvenile nephronophthisis			
	Idiopathic hypercalciuria				
	Dent disease				
		Dent disease type 1			
		Dent disease type 2			
	Nephrogenic diabetes insipidus				
	Primary Fanconi syndrome				
	Autosomal dominant tubulointerstitial kidney disease				
		MUC1-related autosomal dominant tubulointerstitial kidney disease			
		UMOD-related autosomal dominant tubulointerstitial kidney disease			
		REN-related autosomal dominant tubulointerstitial kidney disease			
	Familial primary hypomagnesemia				
		Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis			

			Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis with severe ocular involvement		
			Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis without severe ocular involvement		
		Familial primary hypomagnesemia with hypocalcuria			
			Autosomal dominant primary hypomagnesemia with hypocalciuria		
		Familial primary hypomagnesemia with normocalcuria			
			Primary hypomagnesemia with secondary hypocalcemia		
			Familial primary hypomagnesemia with normocalciuria and normocalcemia		
			Isolated autosomal dominant hypomagnesemia, Glaudemans type		
	Senior-Boichis syndrome				
	Acquired monoclonal Ig light chain-associated Fanconi syndrome				
	Tubulointerstitial nephritis and uveitis syndrome				
	Nephrogenic syndrome of inappropriate antidiuresis				
	Hereditary renal hypouricemia				
	Pseudohypoparathyroidism				
		Pseudohypoparathyroidism with Albright hereditary osteodystrophy			
			Pseudohypoparathyroidism type 1A		
			Pseudohypoparathyroidism type 1C		
			Pseudopseudohypoparathyroidism		
		Pseudohypoparathyroidism without Albright hereditary osteodystrophy			
			Pseudohypoparathyroidism type 1B		
			Pseudohypoparathyroidism type 2		
	RHYNS syndrome				
	Hypotonia-cystinuria type 1 syndrome				
		Hypotonia-cystinuria syndrome			
		2p21 microdeletion syndrome			
		Atypical hypotonia-cystinuria syndrome			

	Dominant hypophosphatemia with nephrolithiasis or osteoporosis				
	Primary renal tubular acidosis				
		Osteopetrosis with renal tubular acidosis			
		Distal renal tubular acidosis			
			Autosomal dominant distal renal tubular acidosis		
			Distal renal tubular acidosis with anemia		
			Autosomal recessive distal renal tubular acidosis		
		Proximal renal tubular acidosis			
			Autosomal recessive proximal renal tubular acidosis		
			Autosomal dominant proximal renal tubular acidosis		
		Hypertrophic cardiomyopathy and renal tubular disease due to mitochondrial DNA mutation			
	Hyperuricemia-pulmonary hypertension-renal failure-alkalosis syndrome				
	Pseudohypoaldosteronism				
		Pseudohypoaldosteronism type 2			
			Pseudohypoaldosteronism type 2A		
			Pseudohypoaldosteronism type 2B		
			Pseudohypoaldosteronism type 2C		
			Pseudohypoaldosteronism type 2D		
			Pseudohypoaldosteronism type 2E		
		Pseudohypoaldosteronism type 1			
			Renal pseudohypoaldosteronism type 1		
			Generalized pseudohypoaldosteronism type 1		
		Transient pseudohypoaldosteronism			
	IgG4-related kidney disease				
	Psychomotor regression-oculomotor apraxia-movement disorder-nephropathy syndrome				
Rare cause of hypertension					
	Adrenocortical carcinoma				
	Congenital renal artery stenosis				
	Genetic hypertension				
		Williams syndrome			
		Catecholamine-producing tumor			
			Von Hippel-Lindau disease		
			Hereditary pheochromocytoma-paraganglioma		
		Familial hyperaldosteronism			

		type I			
		Liddle syndrome			
		Brachydactyly-arterial hypertension syndrome			
		Familial hyperthyroidism due to mutations in TSH receptor			
		Pseudohypoaldosteronism type 2			
			Pseudohypoaldosteronism type 2A		
			Pseudohypoaldosteronism type 2B		
			Pseudohypoaldosteronism type 2C		
			Pseudohypoaldosteronism type 2D		
			Pseudohypoaldosteronism type 2E		
		Pseudoxanthoma elasticum			
		Apparent mineralocorticoid excess			
		Autosomal dominant progressive nephropathy with hypertension			
		Hypertension due to gain-of-function mutations in the mineralocorticoid receptor			
		Familial gestational hyperthyroidism			