PO-0787 CONGENITAL NEPHROTIC SYNDROME OF THE FINNISH TYPE – A NEW MUTATION ENTERS THE SCENE

[1]J Lorenzo, [2]I Beirão, [3]P Matos, [3]C Mota. [1]*Common Year, Centro Hospitalar Entre Douro E Vouga, Porto, Portugal;* [2]*Nephrology, Centro Hospitalar Do Porto, Porto, Portugal;* [3]*Paediatric Nephrology, Centro Hospitalar Do Porto, Porto, Portugal*

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Background Nephrin was first identified in 1998. The Congenital Nephrotic Syndrome of the Finnish type is an autosomal recessive transmitted disease caused by a mutation in the NPHS1 gene that codifies nephrin. The clinical manifestations appear in the first three months of life and progress to end stage renal failure.

Clinical case A seven weeks-old boy with normal grow and psycho-motor development was admitted to the emergency room with vomiting, diarrhoea and mild bilateral pretibial oedema. Laboratory data revealed anaemia, thrombocytosis, normal serum creatinine and urea, normal Na^+, K^+, pH and HCO_3^-, hypoalbuminemia and proteinuria (263 mg/m^2/day). The renal biopsy suggested a Congenital Nephrotic Syndrome of the Finnish type or mesangial sclerosis. The patient was treated with indomethacin and captopril for proteinuria without response. The genetic study confirmed the presence of the IVS9+4 (A >G) variant in homozygosity in the NPHS1 gene. His parents, first-degree cousins, had the same mutation in heterozygosity. The renal disease progressed to end stage renal failure at the age of four years-old. He was supported by continuous ambulatory peritoneal dialysis until the age of six, when he was successfully transplanted with a cadaveric kidney graft.

Conclusions The role of nephrin in the glomerular filtration and stability of the podocytes is unequivocally established. The Congenital Nephrotic Syndrome of the Finnish type, initially found in Finnish families, is present in other areas of the world. The identification of a new mutation in the NPHS1 gene reflects the great variability in the mutations associated with the disease.

PO-0788 'KIDNEYS IN CRISIS' A SNAPSHOT OF CONTINUOUS RENAL REPLACEMENT THERAPY IN PICU, OUR LADY'S CHILDREN'S HOSPITAL, CRUMLIN, DUBLIN

C Magner, C Dee. *PICU, Our Lady's Children's Hospital Crumlin Dublin, Dublin, Ireland*

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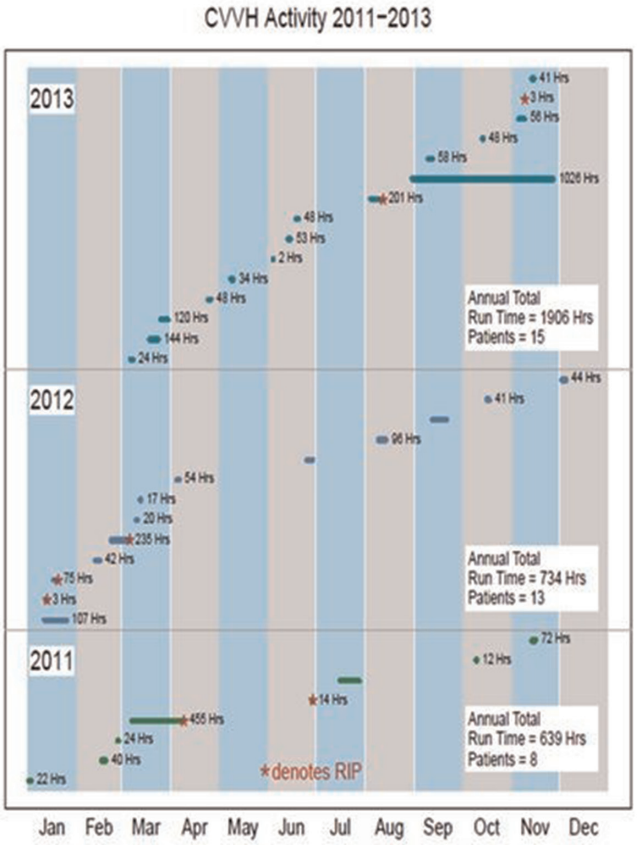
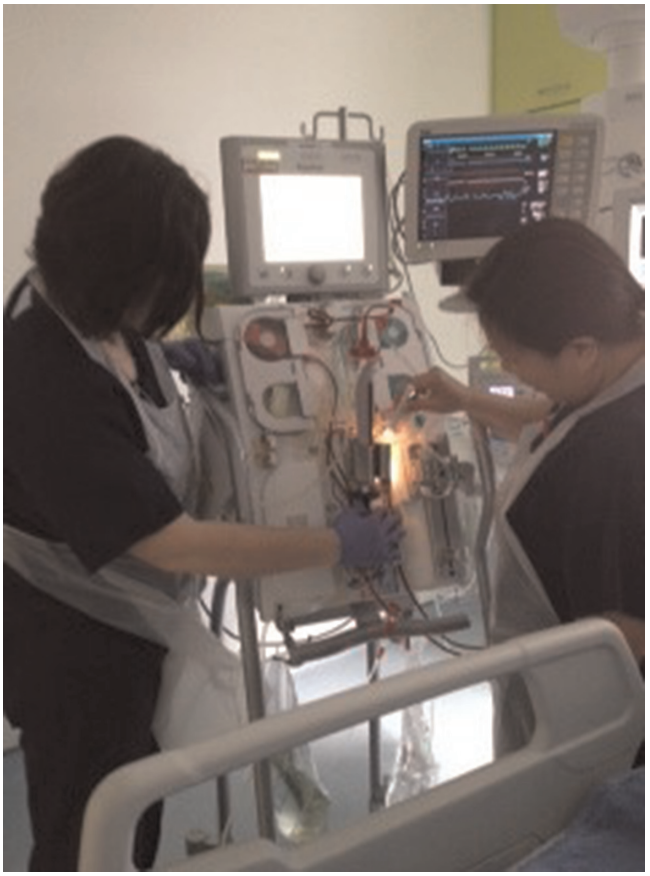
Introduction Continuous Veno Venous Haemofiltration (CVVH) is the extracorporeal renal support therapy of choice in PICU at OLCHC.[1] It is an extracorporeal blood treatment where ≤8% of patients blood is passed through a haemofilter. It removes fluid and waste products from the body in a gradual and controlled way avoiding massive fluid shifts.[2]

CVVH in PICU OLCHC CVVH in OLCHC has seen a considerable expansion over the last 5 years. There are currently 21 CVVH specialists, including a CVVH co-ordinator in post. 2–3 specialists are on duty every shift which ensures continuous service availability. CVVH specialists require 60 pump hours/year to maintain competency. Wet labs are used to facilitate training. CVVH is provided using the Aquarius® which offers a paediatric mode, a safer therapy choice in children (Nikkiso, Co, Ltd).

Results The CVVH service has progressively developed, as is evident in Figure 1. In 2011, 8 patients were supported with CVVH with a run time of 639 h, by 2013 this number increased to 15 patients and the run time had tripled.



Abstract PO-0788 Figure 1



Abstract PO-0788 Figure 2