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

1) Lorenzo, 2) Beirão, 3) Matos, 4) Mota. 1Common Year, Centro Hospitalar Entre Douro E Vouga, Porto, Portugal; 2Nephrology, Centro Hospitalar Do Porto, Porto, Portugal; 3Paediatric Nephrology, Centro Hospitalar Do Porto, Porto, Portugal

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 HCO₃⁻, hypoalbuminemia and proteinuria (263 mg/m²/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.