



## Nephrosis 1, Congenital, Finnish Type

### Alternative Names

NPHS1  
Congenital Nephrotic Syndrome 1  
Nephrosis, Congenital  
Finnish Congenital Nephrosis  
CNF

### Record Category

Disease phenotype

### WHO-ICD

Diseases of the genitourinary system > Glomerular diseases

### Incidence per 100,000 Live Births

6-10

### OMIM Number

256300

### Mode of Inheritance

Autosomal Recessive

### Gene Map Locus

19q13.1

### Description

Finnish congenital nephrosis is a very rare autosomal recessive form of nephrotic syndrome. It is seen more commonly in families of Finnish descent, with an incidence of 1:10,000 births, but can affect every race, with a considerably lower frequency. It is a distinct clinical entity involving massive proteinuria, prematurity, large placenta, hypoproteinemia and marked edema. The typical histological findings of Finnish congenital nephrosis kidneys are dilated proximal tubules, mesangial hypercellularity, and glomerular fibrosis and sclerosis. This progressive disease leads to death in the first two years of life; the only curative therapy is bilateral nephrectomy followed by renal transplantation.

### Molecular Genetics

Finnish congenital nephrosis has been found to be caused by mutation in the NPHS1 gene, spanning 26 kb and contains 29 exons. The NPHS1 is mapped to chromosome 19q31.1 between marker D19S1175 and the APLP1 gene. NPHS1 encodes a podocyte-specific type 1 membrane protein, nephrin, which belongs to the large immunoglobulin (Ig)-like superfamily. The protein has 1,241 amino acid residues, and extracellular part consisting of eight Ig motifs followed by a fibronectin type III domain, a short transmembrane region and a cytoplasmic C-terminal part. Over 50 different mutations, including deletions, insertions, nonsense, missense, splice site and promoter mutations, have been identified both in Finnish and non-Finnish patients with congenital nephrotic syndrome.

### Epidemiology in the Arab World

#### Jordan

Hamed and Shomaf (2001) reviewed the clinical characteristics, pathologic findings, and results of medical management in 30 infants who presented to Jordan University Hospital with congenital nephrotic syndrome in the years 1989 to 1999. Most patients (80%) had parents who were consanguineous. Most patients (80%) were born premature, with an average gestational age of 36 weeks. Most infants (77%) presented the nephrotic syndrome in the first three months of life and 26 (87%) had significant growth retardation. Twenty-five verified episodes of serious bacterial infections occurred in 18 patients. Antibiotic therapy however was successful in all these episodes. Light microscopy of the renal biopsies was consistent with the Finnish type of congenital nephrosis in most patients (83%). Chronic renal insufficiency developed in 17, and five of them needed chronic peritoneal dialysis. All patients died before the age of 5 years. Most deaths occurred at an average age of 15 months (range 1-60). Hamed and Shomaf (2001) concluded that the Finnish type of congenital nephrotic



syndrome is the most common type of congenital nephrotic syndrome in Jordan.

### **Oman**

Rajab et al. (2005) undertook a study to estimate the prevalence of commonly diagnosed autosomal recessive diseases in Oman from a hospital-based register in years 1993 to 2002. The study revealed that congenital nephrotic syndrome (Finnish type) was diagnosed in 25 patients, with an observed incidence of 1 in 20,000 births.

### **Saudi Arabia**

Abdurrahman et al. (1989) evaluated 16 Saudi children with juvenile nephrotic syndrome over a 5-year period. This sample represented 17% of the 92 cases of childhood nephrotic syndrome seen during the period. Onset of the nephrotic syndrome was less than or equal to 3 months of age in four patients. Ten of the patients developed renal failure. Eight patients died, seven of them by 1 year of age. Two patients who were given renal transplants had functioning grafts without recurrence of the disease. Renal biopsy in 12 patients showed congenital nephrotic syndrome of the Finnish type (4 cases). In 1990, Abdurrahman et al. described the clinicopathological features in 119 Arab children from Saudi Arabia with the nephrotic syndrome. The clinical and laboratory data were similar to those described in other parts of the world. Onset of the nephrotic syndrome was at less than 1 year of age in 17 patients (14.3%). There were nine deaths, all in patients with end-stage renal disease: six of the deaths occurred in infants. Abdurrahman et al. (1990) concluded that the pattern of childhood nephrotic syndrome in Saudi Arabia is different from that in tropical countries.

### **Sudan**

Elshibly et al. (1987) reported a 5-month-old Sudanese boy with a probable diagnosis of Congenital Nephrotic Syndrome. The case had features which are neither typical of the Finnish type nor of other hereditary renal diseases. Histologically, the most striking changes were in the glomerular basement membranes which showed patchy thinning, thick segments (with reduplication) and occasional low spikes. Tubules were well preserved, and no foam cells were seen. Electron microscopy showed extensive fusion of foot processes with podocyte microvilli. In parts the glomerular basement membrane showed irregular thickening and splitting, and incorporation of podocyte cytoplasm into the membrane. In other areas, there was marked thinning

of the basement membrane. Immunological features included a high level of IgA and IgG.

### **United Arab Emirates**

Abou-Chaaban et al. (1997) studied the pattern of pediatric renal diseases among children in the Dubai Emirate during the period from 1991 to 1996. In this period, a total of 712 pediatric patients, including 230 nationals of the United Arab Emirates, were seen with various renal problems. Of a total of 13 patients with congenital nephrotic syndrome, three were nationals from the United Arab Emirates. These patients either expired, within the first two weeks of life, or were on conservative treatment awaiting cadaveric donor renal transplantation. Abou-Chaaban et al. (1997) noted that consanguineous parents were a striking feature among their patients with congenital nephritic syndrome.

### **References**

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### **Related CTGA Records**

N/A

### **External Links**

- <http://www.emedicine.com/ped/topic1564.htm>  
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<http://www.orpha.net/data/patho/GB/uk-CNF.pdf>

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