Congenital Nephrotic Syndrome of the Finnish Type

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Dear Sir,

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Congenital nephrotic syndrome of the Finnish type (CNS) is a rare recessively inherited disorder with invariably fatal outcome in infancy [1]. CNS was initially observed in Finland, where it remains most common; however, familial and sporadic cases have been described in many countries and in a variety of ethnic groups [2]. We observed a 4-month-old white male infant admitted for evaluation and treatment of nephrotic syndrome. He was the first child of healthy and not consanguineous parents of Italian ancestry. Gestation and delivery were uncomplicated. The placenta was regarded as 'voluminous' and birth weight of the patient was 4 kg. A cousin died at 18 months of age because of nephrotic syndrome sustained by mesangial proliferative glomerulonephritis.

At 3 months of age the patient suffered from vomiting and eyelid edema. During the next 4 weeks he developed scrotal edema and was admitted to hospital. Physical examination revealed a male infant with anasarca and a poor general condition. Urinalysis showed heavy proteinuria (10 g/l). Total serum proteins were 4.16 g/100 ml and albumin 1.90 g/100 ml. Blood nitrogen was 10 mg/100 ml and creatinine 0.55 mg/100 ml, total lipids 1,190 mg/100 ml and cholesterol 292 mg/100 ml. Electrolytes and C3 were normal. VDRL was nonreactive. Histologic examination of a surgical renal biopsy revealed numerous dilated tubules with microcystic appearance prevalently at the corticomedullary junction. Glomeruli showed a wide spectrum of appearance. The majority were normal in size with mild mesangial hypercellularity accompanied by increase of the matrix (fig. 1). In addition some glomeruli showed focal to global sclerosis (fig. 2) or extracapillary cell proliferation. Fetal glomeruli were scanty. Immunofluorescence studies of glomeruli revealed irregular deposits of C3 and C1q. Electron microscopy

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Fig. 1. Tubular microcysts and mesangial proliferation. HE. × 200.

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Fig. 2. Extracapillary proliferation and segmental sclerosis. HE. × 200.

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constant nor specific [4], show clear progression [5] and are minimal or absent in biopsy specimens taken early in the disease.

This histologic heterogeneity makes the differential diagnosis with other primary and secondary forms of nephrotic syndrome in infants difficult and supports the view that CNS is not a single entity [4]. Therefore, according with Martul et al. [6], we feel that to obtain a more useful classification of the CNS, especially in cases observed outside Scandinavian countries, a better knowledge of the pathogenetic mechanism is necessary. In fact, as demonstrated in congenital syphilis [7], the simple histopathological examination of renal biopsy is insufficient.

Fig. 3. Epithelial cell hypertrophy, microvillous transformation, extensive foot process effacement and irregular thinning of the basement membrane. × 6000.

copy demonstrated that both proximal and distal tubules were cystically dilated, foot processes were diffusely fused and small microvilli were projecting into the urinary spaces from enlarged podocytes (fig. 3). Glomerular basement membranes were irregularly thinning measuring $166 \pm$ (SD) 21 nm; lamina densa measured 62 ± 17 nm. No electron-dense deposits were detectable.

The patient died at the age of 6 months; postmortem examination was not possible.

In this case, the clinical data, the histology of the renal biopsy and the fatal outcome lead to the diagnosis of CNS of the Finnish type, the second case described in Italy [3]. The main histologic changes in CNS of the Finnish type are dilated cortical tubules, mesangial proliferation, sclerosis of glomeruli, tubular atrophy and interstitial fibrosis [1]. However, these lesions are neither

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