

## Congenital Nephrotic Syndrome

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### Abstract

We present a case of a 2-month-old white female with Finnish-type congenital nephrotic syndrome. Her fluid electrolyte level was managed aggressively with high doses of intravenous albumin. Nutritional management included a replenishment diet of 130 to 140 cal/kg per day and protein intake of 3 to 4 g/kg/day. She responded well to therapy and is awaiting kidney transplantation.

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**Key words:** congenital, Finnish-type, kidney transplantation, nephrotic syndrome

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### Introduction

Congenital nephrotic syndrome is a rare condition affecting children from birth to the third month of life. Familial incidence has been described in this condition with an autosomal recessive mode of inheritance. This disease is seen most frequently in Finland with an incidence of 1:8000 live births. Long-term survival is dependent on the correction of nutritional deficits and renal transplantation.

### Case Presentation

A 2-month-old female presented with abdominal distention and generalized edema. The patient was born at 33 weeks gestation by cesarean section due to premature onset of labor. She presented at birth with marked abdominal distention and generalized edema. Her mother is a 31-year-old gravida 1, para 0, from England. Her father is from Lebanon. She had no known Finnish ancestry nor family history of renal illness. Maternal alpha fetoprotein was markedly elevated at 12 weeks gestation. Birth weight was 2.2 kg. Apgar scores were 9/9 at 1 and 5 minutes. The placenta weighed 940 grams.

Physical examination revealed an infant who was small for gestational age but in no apparent distress. Positive findings included a widely split sagittal fissure, wide posterior

fontanelle, and prominent forehead. She had low-set ears and bilateral periorbital edema. The abdomen was visibly distended with obvious collateral circulation. She had no organomalies. Umbilical hernia was readily visible.

Blood chemistry analysis revealed blood urea nitrogen (BUN) 25.2 mmol/L, creatinine 26 µmol/L, total protein 29 g/L, albumin 9 g/L, cholesterol 5.7 mmol/L, triglycerides 3 mmol/L, calcium 1.5 mmol/L, ICa .76 mmol/L, phosphorus 3.3 mmol/L, and carbon dioxide 20 mmol/L. Thyroid function studies were normal. Chromosome analysis was normal 46XX. Urine analysis revealed protein .20 g/L, red blood cell count 15 to 20 per high power field. Radiological studies included renal ultrasound which revealed mild bilateral echogenicity. Voiding cystourethrogram was negative.

### Discussion

Finnish type congenital nephrotic syndrome is a rare condition that presents within the first three months of life. This disease is frequently seen in Finland and familial incidence is common. Gene mapping is traced to chromosome 19. Sporadic and familial cases are described worldwide (non-Finnish type).<sup>1</sup>

Clinical manifestations of this condition include wide anterior and posterior fontanelles, generalized edema, abdominal distention, anasarca, and malnutrition. Characteristic laboratory findings include proteinuria, hypoalbuminemia, and hyperlipidemia.

Differential diagnosis includes primary and secondary causes of nephrotic syndrome sclerosis isolated or in association with male pseudohermaphroditism, Wilm's tumor, idiopathic nephrosis with minimal change disease or focal segmental glomerulosclerosis. Infections such as syphilis, cytomegalovirus, toxoplasmosis, rubella, hepatitis, nail patella syndrome, and Lowe syndrome are among the secondary causes of nephrotic syndrome.

Treatment is based on the clinical course of the patient. Long-term survival of these patients is improved by supportive and surgical management aimed to keep the patient from developing complications associated with malnutrition, loss of renal function, and immunosuppression.<sup>2-4</sup>

Children with congenital nephrotic syndrome usually die in early infancy from complications of the disease or from chronic renal failure. Anasarca is often so severe that patients require nephrectomy.

Recently, new technological advances in infant dialysis

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and aggressive nutritional and metabolic support have improved the long-term survival of patients with congenital nephrotic syndrome. Caloric intake of 120 to 130 cal/kg/day with 3 to 4 g/kg/day of protein can provide optimal growth, control edema, and reduce the number of infections. Patients undergoing renal transplant by 1 to 2 years of age carry the best long-term prognosis.<sup>3,5-7</sup>

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