Congenital Nephrotic Syndrome

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1. INTRODUCTION

Congenital nephrotic syndrome (CNS) is a rare kidney disorder characterized by heavy proteinuria, which is detectable in utero by increased alpha-fetoprotein, hypoproteinemia and marked edema (1).

Infants who develop nephrotic syndrome within the first 3 months of their life are considered to have congenital nephrotic syndrome. The most common of this syndrome is Finnish-type congenital nephrotic syndrome (1, 2, 3). Although, the incidence is reported to be 1.2 : 10,000 in Finland, it is undetermined in various other ethnic groups across the world (4). Later onset of this disease, between three months and 1 year of age, is called infantile nephrotic syndrome.

Many infants that are born with congenital nephrotic syndrome are born prematurely (between 34 and 38 weeks), with a low birth weight for gestational age. The condition is often unresponsive to conventional palliative therapies, thus, dialysis and kidney transplantation is required (3, 4, 5, 6, 7). The prognosis in congenital nephrotic syndrome is reportedly poor with death occurring within few months of life or at about 5 years of age (3, 4, 6, 8). Congenital nephrotic syndrome may be successfully controlled in some cases with early and aggressive treatment, including early kidney transplantation.

2. CASE PRESENTATION

This report presents a case of a 2 month old female infant that was referred to the Pediatric Clinic in Prishtina, Kosovo. She was the first of twins to be born at a regional hospital in Kosovo. The first observation of this baby showed marked edema and distended abdomen. The mother of this infant is 32 years old while the father is 30. This was mother’s second pregnancy. The mother’s second pregnancy was lightly distended, which was neglected at that time, while the rest seemed normal.

Anamnesis showed that this was mother’s second pregnancy. The mother had a five-year-old son, who was healthy, at home and she claimed that they never had anyone in their families with this disease. The newborn baby improved slower than the second twin, who was gaining wait, growing and developing well. Since parents were not alerted by the growing edema and the progressive abdomen distention, they were not aware about this condition and did not seek any medical consultation.

After two months since birth, the baby was hospitalized at the regional hospital due to breathing difficulties and generalized edema. She was diagnosed as bronchopneumonia, generalized edema.

After a one week treatment with antibiotics, steroids, and symptomatic therapy, she was referred to the Pediatric clinic in Pristina since she did not show any improvement. On admission her weight was 4650 g, and body temperature 40°C. Furthermore, the examination showed that this patient had generalized edema, especially on her legs, distended abdomen, ascites, palpable kidneys, blood pressure 110/80 mmHg, as well as bronchopneumonia, proved by clinical evidence and X-ray. Laboratory tests showed erythrocyte sedimentation rate was 45 mm/h, Hgb 9.7 g/dl, white blood cells 9.8, BUN 12.2 mmol/L, serum creatinine 58 µmol/L, total serum protein 35 g/L, albumins 14 g/L, serum cholesterol 10.7 mmol/L, calcium 1.1 mmol/L, Sodium 129, Potassium 4.1. Urine analysis revealed
protein (+ + +) and red blood cells, while the thyroid function was normal.

Serology for TORCH, IgM, HBsAg, HIV ELISA and VDRL were negative. Abdominal ultrasonography revealed bilateral renalomegaly with mild echogenicity. Kidney biopsy confirmed dilatation of proximal tubules and minimal proliferation of mesangial cells.

We started a therapy with antibiotics, plasma therapy, albumin infusion, and diuretics. This therapy brought little results in the beginning.

We continued the treatment daily with albumin infusion, gamma globulin replacement, ACEI and NSAIDs. In addition, the baby received high-protein diet, vitamin supplements.

After this therapy, diuresis was normalized, edema started to reduce and the patient started to show signs of temporary recovery. Parents were informed about the nature of this disease, prognosis and further medical care.

3. DISCUSSION

Infants who develop nephrotic syndrome within the first 3 months of life are considered to have congenital nephrotic syndrome. The most common cause of this syndrome is Finnish type congenital nephrotic syndrome. This is inherited as an autosomal recessive disorder, caused by mutation in the NPHS1 gene located on chromosome 19.(1)

Other causes of congenital nephrotic syndrome include congenital infection such as syphilis, toxoplasmosis, rubella, cytomegalovirus, hepatitis b and HIV infection have also been reported to cause nephrotic syndrome in the neonatal period.(1)

Clinical manifestations of this condition include generalized edema, abdominal distention, malnutrition, respiratory distress and prematurity. The clinical course is one of persistent edema, recurrent infection and progressive renal failure. Characteristic laboratory finding is proteinuria, hypercholesterolemia and hypoalbuminemia. Although, proteinuria is present at birth, the nephrotic syndrome becomes apparent within first three months.

Treatment of CNS is challenging. Corticosteroid and immunosuppressive agents are of no value. Intensive supportive management consists of diuresis, prostaglandin inhibitors, intravenous albumin infusion, along with unilateral nephrectomy to reduce proteinuria (9, 10).

Long term survival of these patients is improved by supportive and surgical management aimed to keep the patient to developing complications associated with malnutrition, loss of renal function and immunosuppressant (11, 13) undergoing renal transplantation by 1 to 2 years of age carry the best long-term prognosis (12, 14, 15).

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 those patients require nephrectomy. Recently, new technological advances in infant dialysis and aggressive nutritional and metabolic support have improved the long term survival of patients with congenital nephrotic syndrome. Caloric intake of 120-130 cal/kg/day with 3 to 4 g/kg/day of protein can provide optimal growth, control edema and reduce the number of infections (16).

In families at risk for the Finnish type of CNS antenatal diagnosis is suggested by an elevated amniotic fluid alpha-fetoprotein level and the diagnosis may be confirmed by DNA analysis.

This case was for the first time diagnosed in University Clinical Centre of Kosova. Since it is very rare disease, with proper diagnose and therapy we achieved good results in treatment during hospitalization. The patient was discharged from the hospital with recommendation that she is sent to some other medical center specialized in dialysis with capabilities for kidney transplantation.

REFERENCES