

Congenital Nephrotic Syndrome

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SUMMARY

The Congenital nephrotic syndrome is rare in pediatric population and associated with significant mortality. This syndrome is characterized by massive proteinuria, a large placenta and marked edema. We report an unusual case of congenital nephrotic syndrome in a two month old baby boy who presented with marked edema.

Keyword: Congenital Nephrotic syndrome, TORCH infection, Hyperlipidemia.

INTRODUCTION

The congenital nephrotic syndrome is a rare autosomal recessive disorder that is most common in population of Scandinavian descent¹. Clinical features include marked edema, heavy proteinuria and hypoalbuminemia². There is no role of corticosteroids however patient may improve with oral indomethacin and ACE inhibitors. The definitive treatment is renal transplant³.

CASE REPORT

A 2 month old baby boy presented to pediatric ward with 20 days history of marked edema. He was afebrile, heart rate of 100beats/min, respiratory rate of 35 breaths/min and blood pressure of 70/50 mmHg. Baby was irritable and markedly edematous. Birth history revealed a large placenta. He was born to the consanguineous parents. There was no history of rash in mother during pregnancy. Complete blood count revealed leukocytosis 20, 000/ mm³. Urine analysis showed proteinuria 3+ and granular cast 15-20/HPF. Blood urea was 21mg/dl, serum creatinine, albumin, cholesterol were 0.5mg/dl, 0.66mg/dl, and 300mg/dl, respectively. The baby was managed with oral indomethacin, ACE inhibitors captopril and intravenous 20% Albumin (salt free).

DISCUSSION

The congenital nephrotic syndrome is a rare autosomal recessive disorder having marked edema, heavy proteinuria, hypoalbuminemia, hyperlipidemia. It presents at birth or during first 3 months of age. Additional clinical features include prematurity, respiratory distress and separation of cranial sutures^{4,5}. There are few documented pediatric cases in Finland⁶. There are two types of Congenital nephrotic syndrome; hereditary and acquired.

Hereditary type includes Finnish type, Diffuse mesangial sclerosis and Familial focal segmental glomerulosclerosis^{7,8}. Acquired types includes Congenital infections (Toxoplasmosis, Cytomegalovirus, syphilis), Systemic Lupus Erythematosus, Other syndrome (Denys-drash syndrome, Epidermolysis bullosa, Nail patella syndrome)⁹.

The natural history of the disease is one of the persistent edema, recurrent infections and progressive renal failure with death by the age of 5 years¹⁰. Congenital nephrotic syndrome may be caused by mutations in one of the 2 genes NPHS 1 and NPHS 2 which encode the nephrin and podocin respectively¹¹. Complication includes: renal failure, hypothyroidism, infections, and growth restriction¹².

Our patient Abdullah had marked oedema with heavy proteinuria, hypoalbuminemia and hyperlipidemia with normal renal functions and negative TORCH's titre. So we labeled it as congenital nephrotic syndrome and planned renal biopsy to know the exact type. Meanwhile patient was put on tablet indomethacin and ACE inhibitor captopril alongwith intravenous albumin. Parents were explained the nature and prognosis of the disease. Patient improved with our management and was discharged on follow up.

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