

Finnish type congenital nephrotic syndrome in a Sri Lankan neonate

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Congenital nephrotic syndrome is a rare condition presenting as oedema in the newborn period. It is often not entertained as a differential diagnosis of neonatal oedema because it has been mainly described among those of Finnish descent. We report a case of histologically proven Finnish type nephrotic syndrome in a Sri Lankan neonate.

Case report

A three week old infant, born at term, weighing 3.15 kg, following an uncomplicated pregnancy (placental size not known), developed lethargy, rapid breathing and poor feeding. He was treated for septicaemia at the local hospital and was transferred to the Lady Ridgeway Children's Hospital with haematuria and abdominal distension. He was the second child of non-consanguineous parents whose first pregnancy had ended in an unexplained intrauterine death at eight months.

On examination, the baby appeared well thrived but hypothermic and pale and had prominent cutis marmorata with a prolonged capillary refill. The abdomen was distended and both kidneys were ballotable. There was moderate hepatosplenomegaly.

Investigations showed a moderate proteinuria (2+) and a field full of red cells with a few pus cells, in the urine. A marked neutrophilia, with a left shift, was seen in the blood picture. The platelet count was $130 \times 10^9/L$. Urine and blood cultures were sterile. The serum creatinine was 3.8 mg/dl. Arterial blood gases showed a metabolic acidosis. Bilateral hydronephrotic kidneys, with diffusely increased echogenicity and poor corticomedullary demarcation, were found on ultrasonic examination. There was no evidence of Wilms tumour or renal vein thrombosis.

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Over the next two weeks, the proteinuria increased and the total plasma protein reduced to 5.9 g/dl and the serum albumin to 1.1 g/dl. The serum cholesterol level was 418 mmol/l. Serum immunoglobulins were within the normal range.

A clinical diagnosis of congenital nephrotic syndrome was made in spite of it being rare among non Finnish populations. Despite aggressive symptomatic management, steroids, captopril and broad spectrum antibiotic cover, he developed gross oedema, ascites, pleural effusions and renal failure. He succumbed at 56 days of life.

At necropsy examination the kidneys were enlarged (right 56x45x25mm, left 65x45x30mm). There was reduced cortical thickness, indistinct corticomedullary demarcation, cystic changes in the paranchyma of the left kidney and prominent pelvicalyceal systems. The ureters and bladder were normal.

On microscopy, expansion of Bowman capsule, increased mesangial cellularity and reduced capillary luminal patency were present in most glomeruli. A few (1%) glomeruli showed crescent formation. The glomerular capillary basement membranes were not thickened. Cystically dilated proximal and distal convoluted tubules were present in some areas. There was no tubular atrophy. The appearance was in keeping with Finnish type congenital nephrotic syndrome. The thymus was atrophic. There was diffuse severe macro and microvesicular steatosis in the liver.

Discussion

Congenital nephrotic syndrome is a rare cause of oedema in the newborn period. Excess weight gain may precede the onset of clinically detectable oedema, as occurred in our patient, who appeared relatively well thrived on admission in spite of ongoing systemic illness for three weeks.

The Finnish type congenital nephrotic syndrome is an autosomal recessive condition due to a mutation in the NPHS1 gene. This causes a reduction in

'nephrin', the key protein component of the glomerular filter¹. Two mutations (Fin-major and Fin-minor) have been described¹. Accurate diagnosis will enable dialysis and transplantation and also identifies families at risk. Antenatal diagnosis, in couples with a positive family history, is done by analysis for the presence of NPHS1 mutation.

Diagnosis of this entity requires a high index of clinical suspicion because there is a misconception that it is only seen in those of Finnish descent. Non-Finnish cases have been described, including one patient in Sri Lanka, although not histologically confirmed as in our report³.

Congenital nephrotic syndrome has a very poor prognosis irrespective of the resources available. It requires urgent treatment with daily albumin infusions and antibiotic coverage. Captopril and indomethacin are used to reduce urinary protein loss. Instead of classical bilateral nephrectomy recent

studies have shown unilateral nephrectomy and dialysis to be an efficacious alternative management strategy which may allow better survival². In the few cases that have survived early dialysis and later transplantation has taken place.

References

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