CASE REPORT

"A stitch in time saves nine": managing congenital nephrotic syndrome in resource-limited circumstances

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SUMMARY

We report a case of congenital nephrotic syndrome. Our patient presented with sepsis-like illness and acute kidney injury during the neonatal period. Subsequent urine analysis showed persistent heavy proteinuria with concomitant hypoalbuminaemia and clinical anasarca. Screening for congenital infections was negative. Diuresis was initially induced with daily albumin infusion. Subsequently, administration of furosemide combined with amiloride was adopted as albumin-sparing strategies. Both angiotensin-converting enzyme inhibitor and non-steroidal anti-inflammatory drugs were started after 4 weeks of life. Such treatment strategies resulted in a significant reduction in the need for albumin infusion, thus allowing better preservation of the venous estate and improving quality of life with a reduced need for inpatient care.

INTRODUCTION

Congenital nephrotic syndrome (CNS) is a rare disorder that presents with heavy proteinuria, hypoalbuminaemia and oedema in the first 3 months of life. Incidence is 0.5 per 100 000 live births.1 This disease is more prevalent in some parts of the world, owing to the genetic makeup of the population. Overall, disease outcomes in CNS are poor, with most children requiring kidney transplant. Herein, we discuss the challenges in the management of an infant with CNS and review the literature on conservative measures in the management.

CASE REPORT

A 3-week-old immigrant baby girl presented with sepsis-like illness. She had decreased physical activity and reduced oral intake. She was born full-term at 38 weeks with a birth weight of 2.5 kg. Antenatally, in-utero growth parameters were normal. Parents were non-consanguineous with no family history of concern. On clinical examination, she had no dysmorphic features. Her anterior fontanelle was sunken with dry mucous membrane. Her measured body weight was 2.2 kg. The cardiorespiratory and abdominal examinations were unremarkable. She had normal female genitalia. Her blood investigations showed hyponatremia and hypokalaemia (serum sodium of 129 mmol/L and potassium 2.9mmol/L) with elevated urea 5.2 mmol/L and creatinine 62 umol/L. Her haemoglobin measured 149 g/L, white cell counts $22.7x10^9$/L and platelet $527x10^9$/L. She had severe hypoalbuminemia with serum albumin of 8 g/L. Her corrected calcium 3.27 mmol/L, phosphate 1.5 mmol/L and magnesium 0.9 mmol/L. Venous blood gas revealed severe metabolic acidosis (pH 7.01, pCO_2 16.9mmHg, HCO_3 7.8mmol/L and base excess of -26mmol/L). Urinalysis showed heavy proteinuria (urine protein 4+). Urine protein:creatinine index was 10983 mg/mmol. Screening for congenital infections including toxoplasmosis, rubella, cytomegalovirus, herpes, hepatitis B and human immunodeficiency virus (HIV) were all negative. Karyotype was 46 XX. Soon after admission, we observed progressive generalised body swelling accentuated by crystalloids used during initial fluid resuscitation. Initially, she had to be given albumin infusion twice a day via a peripherally inserted central venous catheter to facilitate diuresis. Unfortunately, her condition was complicated with ESBL Klebsiella pneumoniae catheter-related blood stream infection. A combination of furosemide and amiloride to control the anasarca was instituted. Upon normalisation of her serum creatinine, both captopril and indomethacin were started.

These strategies allowed sustainable weaning measures of albumin infusions, with no resurgence of oedema. Her caloric intake was optimised to 130 kcal/kg/day. The albumin level remained stable in the range between 14 and 16 g/L. An increment of creatinine by 20% from her baseline was accepted. As expected, she required thyroxine and mineral supplements. Our management protocol allowed alleviation of fluid restriction and created room for optimisation of nutritional intake. It was also not necessary to maintain venous access constantly. She was discharged after 4 weeks of hospital stay. It was possible to continue the management on an outpatient basis without compromising her health. During clinic visits, she showed satisfactory weight gain as well as neurodevelopmental milestones.

DISCUSSION

Infants with CNS typically present with heavy proteinuria, hypoalbuminaemia and oedema. The degree of proteinuria, however, varies amongst individuals and the clinical signs may not be apparent during the first week of life. The true magnitude of proteinuria may be detectable only after partial correction of hypoalbuminaemia by albumin infusion.[2] Common causes of CNS are mutations in genes encoding structural or regulatory proteins of the kidney filtration barrier located in the glomerular capillary wall.[3]

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