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Pediatrics

SENIOR-LOKEN SYNDROME: SIBLINGS WITH RARE DISEASE

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ABSTRACT Senior-Loken syndrome (SLS) is a rare disorder in which there is a combination of nephronophthisis and retinal dystrophy, which usually presents in the first two decades of life. The presenting features of the renal component are polyuria and polydipsia secondary to defective urinary concentrating ability. Nephronophthisis progresses to end-stage renal disease during the second decade. The retinal lesions are variable, ranging from severe infantile onset retinal dystrophy to more typical retinitis pigmentosa. The present case report is about two sisters who presented with decreased visual acuity and end stage renal disease. These cases are presented to highlight the importance of timely recognition of renal derangement in patients with retinal disease to intervene and delay end stage renal disease.

KEYWORDS : Nephronophthisis, renal failure, Tapeto-retinal degeneration, Retinitis pigmentosa

INTRODUCTION

Senior-Loken syndrome is a rare disorder in which there is a combination of nephronophthisis and retinal dystrophy, which usually presents in the first two decades of life. It was first reported by Senior et al, in 1961. [1], who described a family in which 6 of 13 children had nephronophthisis and Lebers tapeto-retinal degeneration. In the same year, Loken et al. [2] described the blindness and renal failure in two siblings. Senior-Loken syndrome is an autosomal recessive disorder that is also known as hereditary renal-retinal syndrome, juvenile nephronophthisis with Leber amaurosis, and renal dysplasia and retinal aplasia. This syndrome accounts for 10 to 15 percent of the cases of childhood genetic kidney diseases known as nephronophthisis. The early features of the renal component are polyuria and polydipsia secondary to defective urinary concentrating ability. With insidious onset of disease and most cases presented very late with renal failure. The retinal lesions are varied from infantile onset retinal dystrophy to retinitis pigmentosa. Considering the unique features and scarce data, this case will highlight the importance of timely recognition of renal derangement in patients with retinal disease to prevent end stage renal disease.

CASE REPORT

An eleven-year-old girl child presented with decreased vision since birth and decreased urine output since last 2 years. She was born through non-consanguineous marriage and had a history of previous blood transfusions for acute blood loss during menarche. General physical examination revealed that child was obese with a weight of 56 kg (> 3 SD), height of 128 cm (< 3 SD). She also had nystagmus in the eyes and visual acuity examination revealed perception of light only. Laboratory findings were suggestive of end stage renal disease (Hb 9g/dl, S. urea 56mg/ dl, Creatinine 7mg/dl, Serum sodium = 145meq/l, Serum potassium = 3.6 meq/l), on continuous peritoneal dialysis at home and erythropoietin injections. Arterial Blood Gas (ABG) showed metabolic acidosis. Blood sugar and liver function tests were normal. On imaging, chest X-ray suggested cardiomegaly, and X-ray of the hand showed short metacarpal bones. The echocardiographic findings were normal. Ophthalmology examination revealed nystagmus, retinal examination depicted pale optic disc and pigmentary changes in the retina. Renal ultrasound showed bilaterally small kidneys, grade III renal parenchymal changes and bilateral cortical cysts. Genetic studies were not done due to financial constrain. Renal transplant planned in next few months.

On detailed history, it was found out that elder sibling expired four years back with similar complaint of diminution of vision and progressive renal failure at the age of 9 years. She was on hemodialysis for 6 months during her last days. Considering the genetic cause of disease, younger sibling (current case) screened for renal parameters 4 years back, which were normal at that time.

DISCUSSION

Senior-Loken syndrome is a rare autosomal recessive disorder with an incidence of 1/100000. More cases are seen in children born out of consanguineous marriages with over 150 cases reported worldwide [3].

Renal involvement is in the form of nephronophthisis and its features started from polyuria that progresses to end-stage renal disease during the second decade. Mutations in at least 14 genes are responsible for nephronophthisis viz (NPHP1, NPHP2, NPHP3, NPHP4, NPHP5, NPHP6, NPHP7, NPHP8 and NPHP9, NPHP10, NPHP11, NPHP12, NPHP13 and NPHPL1) [4-6]. Nephronophthisis usually has insidious onset and progressed to end-stage renal failure before the age of 20 years [7]. However, renal failure is also reported in third and fourth decade [8]. Imaging shows renal cysts with increased echogenicity in the corticomedullary junction. Histopathological changes seen in disease are interstitial fibrosis, tubular atrophy with corticomedullary cyst development and disruption of tubular basement membrane [9].

The retinal involvement associated with SLS includes retinitis pigmentosa, sector retinitis, Leber Congenital amaurosis and tapeto-retinal degeneration [7]. Tapeto-retinal degeneration is the most common manifestation and characterized by a progressive degeneration of the choroid and the retina. Retinitis pigmentosa consist of bone spicule degeneration that begins from the periphery of the retina and progress to involve the entire retino-choroid [10].

CONCLUSION

In the past only 6 case reports have been published in India, which suggest rarity of the disease. Considering the unique features and scarce data, this case will highlight the importance of timely recognition of renal derangement in patients with retinal disease to prevent end stage renal disease.

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