International Journal of Contemporary Pediatrics
Madhura S et al. Int J Contemp Pediatr. 2018 Mar;5(2):651-653
http://www.ijpediatrics.com
pISSN 2349-3283 | eISSN 2349-3291

DOI: http://dx.doi.org/10.18203/2349-3291.ijcp20180573

Case Report

Senior Loken syndrome with Atypical Retinitis Pigmentosa: a rare manifestation of rare disease

Madhura S., Sowrabha, Manjunath, Savitha M. R.*

Department of Pediatrics, Mysore Medical College, Mysore, Karnataka, India

Received: 21 December 2017
Accepted: 25 January 2018

***Correspondence:**
Dr. Savitha M. R.,
E-mail: drsavithamr@yahoo.com

ABSTRACT

Senior Loken syndrome is an autosomal recessive condition characterized by combination of nephronophthisis and retinal degeneration. The earliest presenting features include polyuria and polydipsia secondary to impaired urinary concentrating ability. Nephronophthisis progresses to end stage kidney disease (ESKD) during second decade. The treatment of choice for ESKD due to nephronophthisis is renal transplantation. Retinal lesions are variable ranging from severe infantile onset retinal dystrophy to more typical retinitis pigmentosa. There is a spectrum of other features associated with this condition including skeletal, dermatological and cerebellar anomalies. Till date very few cases have been reported due to lack of awareness of this rare condition. Here, we report a case of Senior loken syndrome with atypical retinitis pigmentosa in a 14-year-old boy.

Keywords: Atypical retinitis pigmentosa, Nephronopthisis, Senior loken syndrome

INTRODUCTION

Senior lokensyndrome (SLS) is a rare syndrome of retinopathy and nephronopthisis and was first described by Senior et al and Loken et al separately in 1961.[1,2] Nephronopthisis is the frequent cause of end stage kidney disease in first three decades of life.[3] Senior Loken syndrome accounts for 10-15% of all cases of nephronopthisis.[4] Senior loken syndrome is an autosomal recessive syndrome included under the category of ciliopathies, a class of genetic diseases that occur due to primary ciliary dysfunction.

The earliest presenting features include polyuria and polydipsia secondary to impaired urinary concentrating ability. Retinal lesions are variable ranging from severe infantile onset retinal dystrophy to more typical retinitis pigmentosa. There is a spectrum of other features associated with this condition including skeletal, dermatological and cerebellar anomalies. Here we report a case of Senior loken syndrome with atypical retinitis pigmentosa.

CASE REPORT

A 14-year-old male child presented to us with a history of facial puffiness for 6 days, swelling of lower limbs for 6 days, decreased urine output for 3 days. On detailed history, he complained of polyuria and nocturia in the past 1 month. Further questioning revealed history of progressive decreased vision in the dark since 1 month. Child was 3rd born to a second degree consanguineously married couple. There was no history of similar complaints in other family members. On examination, child's height was 141 cm (-3 and -2 SD) weight was 30 kg and BMI was 15.15 (-3 and -2 SD).

Further examination revealed bilateral puffiness of eyes and facial puffiness with bilateral pitting pedal edema till ankle joints. Skin was dry. Child was hypertensive.

Systemic examination was unremarkable. On fundoscopic examination child had features suggestive of atypical retinitis pigmentosa.

Laboratory investigation revealed renal dysfunction. Initial blood urea: 337 mg/dl, serum creatinine: 17.8 mg/dl and Urine routine showed 10-15 pus cells, 8-10 RBCs, 3+ albumin, and granular casts. CBC revealed child was anemic with haemoglobin of 8.5g/dl. ABG was suggestive of metabolic acidosis. USG abdomen revealed Grade III medical renal disease (MRD) with cystitis with no obvious cystic lesion.

Hence diagnosis of senior Loken syndrome was made considering the age of presentation, ophthalmoscopic findings and renal parameters. Child was subjected to haemodialysis and given supportive treatment.

DISCUSSION

Senior Loken syndrome is a rare syndrome of retinopathy and Nephronopthisis. It was first described by Senior et al and Loken et al separately in 1961.[1,2] Nephronopthisis is the most frequent cause of end stage kidney disease in first two decade of life.[3] Senior Loken syndrome accounts for 10-15% of all cases of Nephronopthisis.[4]

Renal involvement occurs in the form of nephronopthisis and is characterised by chronic tubulointerstitial nephritis that progresses to end stage kidney disease (ESKD) during 2nd decade of life. Mutation in the 9 genes (NPHP 1-9) are responsible for nephronopthisis.[5] There are three clinical variants of Nephronopthisis according to the age of onset of ESKD.[6] Infantile form is associated with blindness in infancy, hypertension and death from renal failure before the age of 10. It is caused by mutation in the NPHP 2 gene which is located on chromosome 9q31. It is associated with extrarenal features which include hypertension, situs inversus and ventricular septal defect.[4,6] Adolescent form is caused by mutation in NPHP3 gene located on chromosome 3q22 in which ESKD occur early in adulthood and histological form is similar to juvenile form.[4,6] Juvenile form is the most common type of nephronopthisis and it accounts for 5-10% of all cases of ESKD. It is caused by mutation in 8 different genes (NPHP 1, 2, 3, 4, 5, 6, 7, 8, 9).[3] NPHP 1 is the common gene which is responsible for juvenile nephronopthisis. It is characterised by late onset slow progression of renal disease and mild ocular manifestation.

Renal involvement is mild and asymptomatic. The earliest signs are decreased urinary concentration ability which leads to polyuria, polydipsia and renal loss of sodium by 4-6 year of age.[7] Proteinuria, hematuria are absent or minimal. BP remain normal until development of chronic kidney disease which invariably occur before 20 years of age.[8] USG findings may be normal, or they may show an increased echogenicity of renal parenchyma, poor corticomedullary differentiation, small kidney and medullary cysts. However, the lack of medullary cysts at presentation does not rule out diagnosis of juvenile nephronopthisis.[9]

Nephronopthisis should not be confused with autosomal dominant polycystic kidney disease (ADPKD) which is characterised by bilateral multiple renal cysts or Medullary cystic kidney disease (MCKD) which shares features of nephronopthisis. However, unlike nephronopthisis ADPKD and MCKD are inherited in an autosomal dominant pattern and progress to ESKD at later age.[10]

Retinal lesion in SLS are variable ranging from severe Leber's amaurosis to a more typical Retinitis pigmentosa.[11] Laber's amaurosis is a severe form of retinal dystrophy which leads to blindness in infancy, nstagmus and diffuse atypical retinal pigmentation and pallor of optic disc with early and complete extinction of Electroretinogram (ERG). Retinitis pigmentosa is characterised by bone spicule pigmentation of retina and presents initially with night blindness which gradually progress to day time blindness.

The diagnosis of SLS is based on typical clinical presentation of a clinical interstitial nephritis and retinopathy in 2nd decade of life. There are very few case reports of SLS due to lack of awareness of this rare condition.

Though the case we report here did not show any cystic changes in the kidney, the typical age of presentation, renal symptoms and ophthalmic findings are consistent with SLS which led to the diagnosis of SLS in the absence of genetic analysis.

CONCLUSION

SLS must be considered in any patient who present with renal failure in first decade of life with retinitis pigmentosa. Detailed ophthalmic evaluation must be done in all cases presenting with renal failure. Similarly, children presenting with retinitis pigmentosa early in the life must have regular assessment of renal parameters and renal ultrasound. All siblings of child with senior Loken syndrome must be screened as it is inherited autosomal recessively. There is no specific treatment for this condition. Renal transplantation is the preferred therapy. However regular monitoring of BP, restriction of high protein diet intake can delay the progression to ESKD and thus need for renal transplantation.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

REFERENCES

1. Senior B, Friedmann AI, Braudo JL. Juvenile familial nephropathy with tapetoretinal degeneration. A new oculorenal dystrophy. Am J Ophthalmol. 1961;52:625-33.

2. Loken AC, Hanssen O, Halvorsen S, Jolster NJ. Hereditary renal dysplasia and blindness. Acta Paediatr. 1961;50:177-84.
3. Hildebrandt F, Zhou W. Nephronophthisis associated ciliopathies. J Am Soc Nephrol. 2007;18:1855-71.
4. Salomon R, Saunier S, Niaudet P. Nephronophthisis. Pediatr Nephrol. 2009;24:2333-44.
5. Otto EA, Tory K, Attanasio M, Zhou W, Chaki M, Paruchuri Y, et al. Hypomorphic mutations in the meckelin (MKS3/TMEM67) cause nephronophthisis with liver fibrosis (NPHP11). J Med Genetics. 2009;46(10):663-70.
6. Saunier S, Salomon R, Antignac C. Nephronophthisis. Curr Opin Genet Dev. 2005;15(3):324-31.
7. Gusmano R, Ghiggeri GM, Caridi G. Nephronophthisis-medullary cystic disease: clinical and genetic aspects. J Nephrol. 1998;11(5):224-8.
8. Hildebrandt F, Strahm B, Nothwang HG, Gretz N, Schnieders B, Singh-Sawhney I, et al. Molecular genetic identification of families with juvenile nephronophthisis type 1: the rate of its progression to renal failure. Kidney Int. 1997;51(1):261-9.
9. Blowey DL, Querfeld U, Geary D, Warady BA, Alon U. Ultrasound findings in juvenile nephronophthisis. Pediatr Nephrol. 1996;10(1):22-4.
10. Simms RJ, Eley L, Sayer JA. Nephronophthisis. Eur J Hum Genet. 2009;17(4):406-16.
11. Satran D, Pierpont ME, Dobyns WB. Cerebello-oculo-renal syndromes including Arima, Senior-Loken and COACH syndromes: more than just variants of Joubert syndrome. Am J Med Genet A. 1999;86:459-69.

Cite this article as: Madhura S, Sowrabha, Manjunath, Savitha MR. Senior Loken syndrome with Atypical Retinitis Pigmentosa: a rare manifestation of rare disease. Int J Contemp Pediatr 2018;5:651-3.