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Senior Loken Syndrome*

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The association of nephronopthisis and tapetoretinal degeneration was first described by Senior[1] and Loken[2] in 1961, and only 36 cases have been published up till 1976.[3] The severe juvenile type produces blindness in infancy and death due to renal failure before the age of ten. This disease has an autosomal recessive inheritance pattern. Here we would like to present a case of Senior Loken syndrome in which the family history was not investigated.

Case Report

S. N., a 11 year old girl, was admitted to Hacettepe Children's Hospital in June 1976 with the complaints of amblyopia and polyuria which were noticed soon after birth, and contractions of her extremities observed over the last 15 days.

Her parents were first cousins and unaffected. Her family history revealed that the first two children of the family had died when they were 2-3 months old with complaints of convulsinos, whereas her other three siblings were said to be in good health. There was no previous family history of renal disease, hypertension or ocular changes.

The Positive Physical Findings Were Limited to: Total blindness and nystagmus, diffuse atypical retinal pigmentation with pallor of the optic disc and narrowing of the retinal arteries were noted on fundal examination. Her blood pressure was 150/90 mmHg, height 134 cm, weight 34 kg.

Laboratory Findings: Renal function was impaired, her daily urine volume was 2.5 l with maximal urinary osmolality of 164 mosm/kg after water restriction, BUN 76 mg/dl, serum creatinine 3.2 mg/dl, endoge-

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nous creatinine clearance 22 mg/min/m^2. Hyperchloremic acidosis was observed (CO_2 content 10.8 mEq/l, chloride 111 mEq/l). Hypocalcemia (7.2 mg/dl) and leukocyturia were present. Bacteriuria and mild proteinuria were also observed. 100.000 colonies/ml Klebsiella were seen in her urine culture.

Renal biopsy showed tubulo-interstitial lesions compatible with nephronophthisis. (Figure 1).

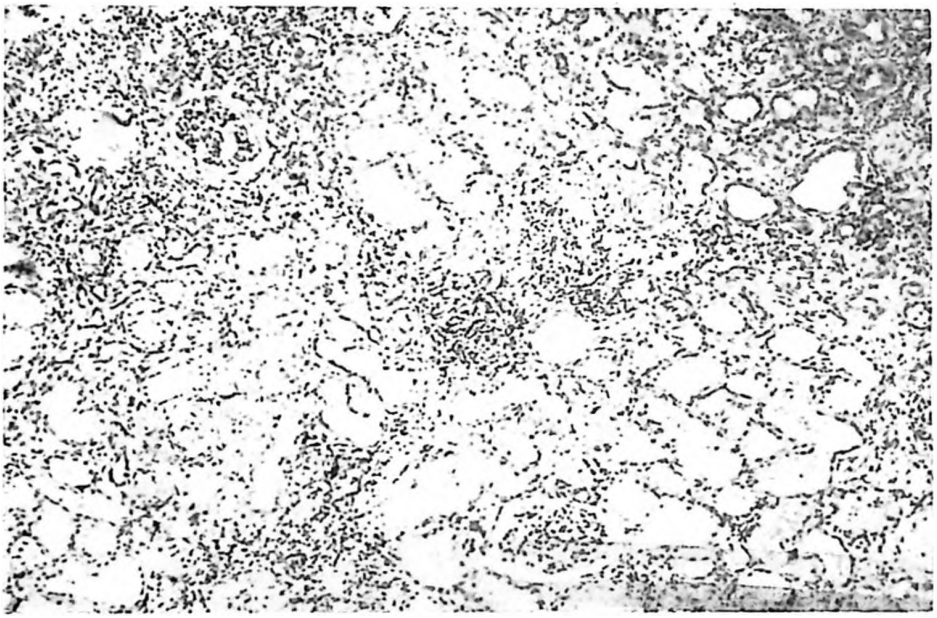

Figure 1

Discussion

Ocular lesions in association with renal disease have been reported with increasing frequency since the original report by Sohar.[4] However, the most common renal abnormalities with ocular lesions are known to be hereditary hematuric nephritis or Alports syndrome. In this syndrome the most frequently reported ocular lesions have been spherophakia, anterior lenticonus and cataracts.[4, 5, 6] This condition is characterized by remission and exacerbation in both symptoms and urinary findings.

Association of nephronophthisis and tapetoretinal degeneration is another recognized hereditary disorder.[1, 2, 7] The term tapetoretinal degeneration covers a group of disorders characterized by progressive degeneration of the choroid and retina but varying in nature and severity. The case presented here had amaurosis congenital.[3] This severe form

leads to blindness in infancy. There is nystagmus and diffuse atypical fundal pigmentation with pallor of the optic disc and narrowing of the retinal arteries.

In the renal component of this syndrome polyuria and polydipsia are frequently the only symptoms and impaired urinary concentration ability is the earliest sign. Anemia, hypocalcemia, hyperphosphatemia, and hyperchloremic acidosis are found proportional to the degree of renal failure. Although the urinary sediment is usually normal, leukocyturia was noted in our case.

The renal disease is progressive, but some patients may reach the third decade.[8, 9] Our case progressed to terminal renal failure few months after she was diagnosed.

The pathogenesis of Senior Loken syndrome remains obscure. Some authors regard it as a genetically conditioned malformation affecting two different ectodermal tissues by a single gene with pleotropic action and variable expressions, producing nephronophthisis or tapetoretinal degeneration alone or in combination.[3] Recessive inheritance is generally accepted for this syndrome and the parental consanguinity of our patient supports this. In our case the clinical symptoms, course and laboratory data were compatible with the fully developed picture of this syndrome.

Summary

A 11 year old girl with Senior Loken Syndrome is presented. The case shows the association of nephronophthisis and tapetoretinal degeneration. Her parents were first cousins and unaffected. Also, there was no known history of renal disease.

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