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# UNUSUAL SYSTEMIC ASSOCIATION OF RETINITIS PIGMENTOSA SENIOR LOKEN SYNDROME- A RARE CASE REPORT

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**ABSTRACT** A 22 year male patient presented to our institute with complaints of facial puffiness followed by pedal edema,polyuria .Patient was referred to ophthalmology OP for loss of vision since childhood similar complaints are present in his sister.on examination visual acuity is absence of perception of light and pupils not reacting to light in both eyes. Horizontal nystagmuspresent.fundus picture showed advanced retinitis pigmentosa. USG and CT revealed echogenicity and multiple cortical cysts.

**CONCLUSION:** Thus association of nephronophthisis and retinitis pigmentosa with positive family history correlates to rare presentation known as senior loken syndrome.

**KEYWORDS :** polyuria, nystagmus, retinitis pigmentosa, family history, cortical cysts

## INTRODUCTION:

Senior- Loken syndrome refers to a disorder in which there is combination of nephronophthisis and retinal dystrophy. It was first described in 1961 by Senior-et-al, who described a family in which 6 of 13 children had nephronophthisis and tapeto-retinal degeneration[1]. In the same year Loken et al [2], described the same condition in two siblings both siblings had blindness and severe renal failure and a kidney biopsy showed renal tubular atrophy and dilatation.Thisgroup of illness affecting approximately 1 in 50000 births.

## CASE REPORT:

A 22 year male patient presented to our institute with complaints of facial puffiness followed by pedal edema. Complaints of polyuria and dysuria are present. Patient also complained of loss of vision since childhood. On examination nystagmus was present for which he was referred to ophthalmology. On ophthalmological examination visual acuity is absence of perception of light in both eyes. Horizontal nystagmus is present. Anterior segment is normal in both eyes and pupils are not reacting to the light . Dilated fundus examination showed pale optic disc and arteriolar attenuation with background retina showing bony spicules which suggests the picture of typical retinitis pigmentosa .USG and CT KUB revealed echogenicity and multiple renal cortical cysts.patient was born out of second degree consanguinous marriage. He was the third child out of 4 children where similar complaints and findings are present in his elder sister.Other general examination of the patient is within normal.

Blood investigations include serum creatinine-7.5mg/dl blood urea-189 mg/dl , Hb-7.7%,ESR 110, platelet count 92000 cell/dl. LFT and serum electrolytes are normal. Urine examination shows albumin 3+, pus cells 3-5, EP cells 3-4.

Ultranosonography and CT KUB was done which revealed multiple corticalcysts.patient was on maintainance hemodialysis.

**Figure 1,2-fundus photographs showing bony spicules in right eye and left eye**

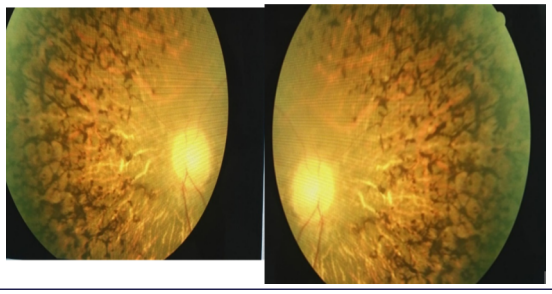
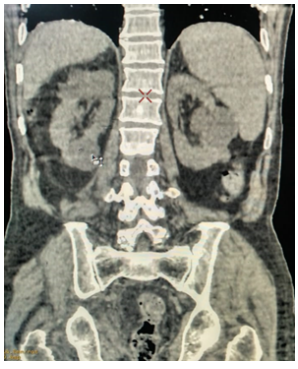


**Figure 3-CT KUB showing Renal Cortical cysts**



## DISCUSSION:

This is a patient presented with pedal edema , facial puffiness, polyuria and typical retinitis pigmentosa with nystagmus and renal cortical cysts suggests the possible diagnosis of Senior-Loken syndrome.

It is a rare autosomal disorder that results in nephronophthisis and retinal dystrophies.[3]

Renal involvement is in the form of nephronophthisis and is characterized by chronic tubulointerstitial nephritis that progresses to end stage renal disease during second decade.[4].The initial symptoms are polyuria,polydipsia and enuresis due to a concentrating defect , which is consistent with the symptomotology of our patient.

The ocular involvement takes several forms : it could be early and severe congenital amaurosis of Leber type or late-onset pigmentary retinal degeneration. Tapeto –retinal degeneration varies in its nature and severity. It is characterized by a progressive degeneration of the choroid and the retina.[5]

Other symptoms have been noted in particular families such as diabetes insipidus, neurosensory hearing loss, muscular incoordination caused by disease of the cerebellum in the brain, abnormal formation of fibrous tissue in the liver and skeletal abnormalities.

It is currently known to be caused by mutation atleast 10 genes that is NPHP1,NPHP2,NPHP3,NPHP3,NPHP4,ICQB1/NPHP5,CEP290/NPHP6,SDCCAG8/NPHP10,WDR19/NPHP13,CEP164, and TRAF31P1

. The proteins created by these genes play a role in cell structures called

cilia,which are microscopic hair like structures that stick out from surface of the cell and responsible for transmitting information between cells .Cilia are essential for sensory input, since retinal photoreceptors have a cilium connecting outer and inner segments of the cell and these cilia are also important for the structure and functionof the cells in the kidneys.

. Mutations in one of the gene lead to problems with the functioning of cilia resulting in disruption of important signaling within the cells.hence the damaged cilia are responsible for r Senior –Lokensyndrome considered as a ciliopathy which is a heterogeneous genetic disorder ,affecting many parts including kindney, eye, liver and brain.

There is no proven treatments for nephrnoophthisis . Management primarily depends on delaying the progression of renal failure and the need for dialysis. Newer agents such as vasopressin V2 receptor antagonists,which alter the cytogenesis and progression of the disease, may be available for future use[8]

As our patient is in the end stage renal disease he was on hemodialysis and no ocular intervention can be done as the patient could not perceive the light

Diagnosis of this syndrome is through genetic testing , clinicalexamination, kidney and ophthalmological evaluations as well as hepatic and neurological examinations are recommended.
Conclusion: Patients with Retinitis pigmentosa should be evaluated systemically to rule out the nephronophthisis & serves to take thorough measures to prevent progression of systemic disease and helps for genetic counseling regarding ocular condition

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