SENIOR-LOKEN SYNDROME: ABOUT TWO CASES

A.IZEM, K. MABROUK, F.OUBAHAIBOU, N.MTIOUI, S. ELKHAYAT, M. ZAMD, G. MEDKOURI, M. BENGHANEM, B. RAMDANI

Department of Nephrology – Ibn Rochd University Hospital – Casablanca, Morocco

INTRODUCTION -AIMS

- The Senior- Loken syndrome is a rare autosomal recessive renal ocular disease characterized by the association of nephronophthisis, a chronic tubulointerstitial nephropathy, and retinal dystrophy and other systemic involvement.. The global prevalence is estimated at 1 / 1,000,000. Several cases have been reported in different countries in the world. Senior et al first described it in 1961 and Loken et al in the same year described the condition in two siblings.
- We report the first observation in our country.

CASE REPORTS

- There are two children of the same family from a consanguineous marriage, a boy and a girl, aged 16 years and 15 years.
- The two patients with Leber congenital amaurosis since the age of one year.
- The first patient 15 years old, followed for Leber congenital amaurosis from 1 year of age, ophthalmologic evaluation revealed a retinal dystrophy.
- This patient had a notion of polyuria-polydipsia syndrome one since childhood.
- At the age of 11, a report made on the occasion of a digestive symptoms objectifying advanced chronic renal failure, end-stage arrived at after a year.
- Abdominal ultrasonography: both kidneys small dedifferentiated. Moreover, the other organs are normal.
- The genetic study revealed ABCB1 gene mutation in the homozygous state.
- Currently the patient received a kidney transplant from a living donor related, with a good evolution .
- The investigations in older sister of this patient, aged 16, also having a blindness and chronic renal failure showed the same genetic mutation as part of this syndrome.

CONCLUSION

- The Senior-Loken syndrome is a genetically heterogeneous ciliopathy. It must be considered in patients who present with visual impairment and renal failure in the first two decades of life.
- Mutations in seven different genes have been reported Kidney and eye exams are recommended.
- Liver function tests to exclude hepatic fibrosis and neurological examination are also recommended in infants.
- Genetic diagnosis of Senior-Loken syndrome based on mutational analysis of genes involved.
- The early management of this syndrome can delay progression to end stage renal disease and reduce secondary complications.
- There is no specific treatment; however, renal transplantation is the preferred therapy because nephronophthisis will not recur after transplantation.

BIBLIOGRAPHIE

1-Giridhar S, Padmaraj R, Senguttuvan P. Twins with Senior-Loken syndrome. Indian J Pediatr 2006;73(11):1041-3

2. Muftah Othman, et al., Senior-Loken syndrome: Two cases from the State of Qatar .The Journal of Clinical and Diagnostic Research. 2012 October, Vol-6(8): 1411-1413

3-Sarangapani SC, hang L, Gregory-Evans K, Cataract surgery in Senior-Loken syndrome is beneficial despite severe retinopathy. Eye 2002;16(6):782-5

4-Senior B, Friedmann AI, Braudo JL. Juvenile familial nephropathy with tapetoretinal dege-neration: a new oculorenal dystrophy. Am J Ophthalmol 1961;52:625-33

DOI: 10.3252/pso.eu.52era.2015

ePosters supported by F. Hoffmann- La Roche Ltd.