WCN24-AB-1228
Senior-Loken syndrome. Report of two cases.

Gómez Gabriela, Zarate Lucia, Ceccoli Ruben, Fabriani Damian, Camus Jimena, Garcia Daiana, Moya Vanesa, Ramirez Jesica.
Department of Nephrology, Central Hospital, Mendoza.

INTRODUCTION AND GOALS

Senior Loken Syndrome (SLS) is a rare genetic disorder having juvenile nephronophthisis and Leber congenital amaurosis or retinitis pigmentosa. Pathogenic variants in several genes as CEP290, IQCB1, NPHP1, NPHP4, SDCCAG8, TRAF3IP1, WDR19 and IFT40 are known to be associated with this condition. Nephronophthisis autosomal recessive cystic kidney disease leads to kidney failure in childhood/adolescence, the most common genetic cause of kidney failure in children. It can be combined with extrarenal manifestations, hepatic fibrosis, situs inversus or cardiac malformations. When combined with retinitis pigmentosa, the disorder is known as Senior-Loken síndrome.

GOALS: establish the importance, early detection and diagnosis of this syndrome, highlighting the need for a multidisciplinary evaluation and comprehensive management of these patients.

CASE PRESENTATION

We presente de case of 36-year-old woman wiyh history of pigmentous retinitis, myopia, strabismus, diagnosed at 5 years. In July 2020 she was admitted for optic neuritis and posterior subcapsular cataract in both eyes. She received treatment with methylprednisolone in this event. Referred to nephrology for CKD due to kidney symptoms and HTN of one year's duration Family history: 25-year-old brother with the same diagnosis since childhood. Both born to consanguineous parents. Studies performed: Normocytic normochromic anemia, uremia 1.46 gr/L, creatininemia 46 mg/L DCE 14 ml/min, non-nephrotic range proteinuria, negative immunological and infectious causes. Renal ultrasound 10/02/2020 both kidneys are of preserved shape and location with increased echogenicity and decreased size and thickness of the parenchyma. Treatment with hemodialysis was prescript. They were referred to genetic assesment. Multipanel germline next generation sequencing study modified by customer (BluePrint genetics) revealed two heterozygous variant of uncertain significance (VUS) c.70C>T p.(His24Tyr) and c.653A>T p.(Asp218Val) in IFT40 gene that confirm compound heterozygous status in both patients.

CONCLUSION: Spread knowledge of SENIOR LOKEN syndrome among health professionals and medical community in order to increase awareness and promote research in this area for the development of new therapeutic approaches.Co-segregations variants studies increased suspicion of pathogenicity in variant´s clasiffication.

MULTIPANEL GERMLINE NEXT GENERATION SEQUENCING STUDY

Gen:	Variation:
IFT40	c.70C>T p.(His24Tyr)
IFT40	c.653A>T p.(Asp218Val)