KSN 2021
FULLY VIRTUAL MEETING
September 02 (Thu) - 05 (Sun)

Abstract Submission No. : 9179

PAX2-related nephropathy

Jiwon Lee
Division of Rare Disease Management, Korea Disease Control and Prevention Agency, Korea

Paired box 2 gene (PAX2) has a crucial role in kidney development and encodes for a transcription factor in the kidney and ureter, as well as in the eyes, ears, and central nervous system. Pathogenic variants in PAX2 causes renal and ophthalmological anomalies, such as renal coloboma syndrome (OMIM#120330) with or without vesicoureteral reflux (VUR). Renal coloboma syndrome is also referred to as papillorenal syndrome. However, PAX2-related disorders have clinical variability, showing non-renal and non-ophthalmological manifestation as well. This talk presents current understandings of PAX2-related disorders with the focus on nephropathy.