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The Lightning Strikes Twice: The Coexistence of Alport Syndrome and SEC61A1- Related Disorder

Olga Szczerbak
Agiellonian University,Poland

Alport syndrome (AS) is an inherited, progressive kidney disorder affecting the glomerular basal membrane, often associated with sensorineural hearing loss and ocular abnormalities.[1] Predominantly X-linked (XLAS), it is caused by COL4A5 mutations[2], typically presenting with hematuria at times with proteinuria by age 5.[3] Diseases linked to SEC61A1 mutations include severe congenital neutropenia (SCN), involving recurrent infections due to impaired myeloid maturation[4,5], and autosomal dominant tubulointerstitial kidney disease (ADTKD), characterized by progressive renal dysfunction.[6] A two-month-old boy was referred to the Pediatric Nephrology Department from a regional hospital with a persistent elevated creatinine levels, proteinuria, and abnormal renal ultrasound findings foregoing short-time history of eyelid swelling and transient oliguria. Physical examination revealed numerous dysmorphic features including dolichocephaly, broad nasal root, and polydactyly. He had no family history of congenital kidney diseases. Initial laboratory tests revealed interim neutropenia and glucosuria, persistent elevated serum creatinine (eGFR 40 ml/min), hypogammaglobulinemia, and elevated cystatin C. Renal ultrasound showed hyperechoic cortex and hypoechoic pyramids. Whole exome sequencing identified a pathogenic COL4A5 and a SEC61A1 variants linked to SCN and ADTKD. At six months, the boy was readmitted with hypertension, averaging 140/92mmHg and worsening cortical changes on renal ultrasound, beside normal renal arteries. Combined, antihypertensive therapy was successfully initiated, and the patient was discharged in stable condition. Follow-up urine tests showed neither proteinuria nor glucosuria. The coexistence of two pathogenic variants in COL4A5 and SEC61A1 is highly unusual, but probable. This case is remarkable since AS rarely causes deterioration of kidney function at such a young age and it's major hallmark – erythrocyturia was absent in this case. Similarly, renal manifestations of SEC61A1 mutations typically appear later in life. The rapid emergence of hypertension underscores the unusual severity of the disease's progression. This case highlights the importance of multidisciplinary clinical and genetic management in complex pediatric nephropathies..

Biography

Olga Szczerbak is a fourth-year medical student at Jagiellonian University in Kraków with a strong interest in nephrology. She leads the Renal Replacement Therapy Students' Scientific Group, where she oversees research projects and promotes academic growth. Additionally, she is an active member of the Pediatric Nephrology Students' Scientific Group, contributing to studies focused on kidney diseases in children. Her involvement in both groups demonstrates her dedication to scientific research and her aspiration to advance the field of nephrology through innovation and collaboration.

olga.szczerbak@student.uj.edu.pl

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