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SCHIMKE immuno - osseous dysplasia: the first clinical case report in Kazakhstan.

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Case Study : Background: SMARCAL1 gene polymorphism is reported to present with defects of multiple systems of organs, including cellular immunity defects, growth delays, spondyloepiphyseal dysplasia, focal segmental glomerulosclerosis and others, collectively known as Schimke syndrome. Schimke immune – osseous dysplasia (SIOD) is an ultra extremely rare multisystemic, monogenetic and autosomal recessive. Less than 100 cases have been reported and its prevalence is 1 in 1–3 million new births, so Orphanet considers it an ultra-rare disease. Early-onset affected patients show severe symptoms and die at around 10 years of age due to strokes, severe opportunistic infections, bone marrow failure, kidney failure, cardiovascular issues, and other complications. NPHP4 gene polymorphism is a known cause of isolated nephronophthisis. Case description: Patient Zh. is a 8 years old male presenting with the signs of corticosteroid resistance nephrotic syndrome. The patient had ischemic stroke in the basin of the middle, anterior cerebral artery on the left, right-sided hemoplegia, motor aphasia, hypogonadism and growth delay, symptomatic arterial hypertension, secondary cardiopathy, severe IgG hypogammaglobulinemia. During hospitalization the patient was found to have SMARCAL1 and NPHP4 genetic polymorphisms. Both polymorphisms carry bad prognosis for children. Discussion: We present the first Kazakhstani case of SIOD reported in the literature. The case of a child with a combination of both polymorphisms will be reported. Conclusion: Schimke immune – osseous dysplasia (SIOD) is extremely rare, but taking into account the involvement of various organs and systems in the pathological process, it is necessary to involve Practical implications will be discussed and recommendations will be given. a multidisciplinary team of specialists for timely diagnosis. The presence of nephrotic syndrome, especially morphologically FSGS, with pronounced growth retardation, cerebrovascular involvement in a child at the onset of the disease should always alert doctors in terms of diagnosis of Schimke syndrome.