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First clinical case of Wilms tumor 1 gene in a patient with Denys-Drash syndrome, followed by kidney transplantation, Central Asia, Kazakhstan.

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Case Study : Introduction: Denys-Drash syndrome is a rare genetic disorder characterized by early-onset nephrotic syndrome that rapidly progresses to renal failure within the first few years of life, pseudohermaphroditism, and a high risk of developing Wilm's tumor. Variants of the Wilms tumor 1 gene have been shown to be pathogenic for DDS. The WT1 gene encodes proteins that regulate progenitor cells and their differentiation, particularly those of the gonads, uterus, and kidneys. Case presentation: The onset of infantile nephrotic syndrome debuted at 9 months: high activity of nephrotic syndrome. At the age of 1.5 years,a diagnostic nephrobiopsy was performed: the specimen is uninformative. At the age of 2 years, during a control ultrasound of the kidneys: a space-occupying lesion in the left kidney. In order to clarify the diagnosis, a CT scan: a showed a space-occupying lesion in the left kidney, oval in shape, with a clear even contour, measuring 6.2 x 4.9 x 4.8 cm. The following surgery was performed: left tumor nephrourethroectomy. The morphological picture and immunophenotype correspond to Wilms' tumor, mixed variant with a predominance of the blastomatous variant. Resection margin and ureter wall, venous vessel without tumor growth. Hyperplasia of lymph nodes. Stage III. The child has completed the course of treatment: 1 course of NPCT, 12 courses of APCT. Cytogenetic study result: karyotype 46,XY.4 years a surgery was performed to implant a peritoneal catheter, eliminating cryptorchidism on both sides. 5 years have passed since the kidney transplant from a living donor (donor father). According to the results of the examination, the transplanted kidney function is satisfactory. Conclusions: Early recognition of DDS in patients with SRNS is critical due to its low prevalence, the specific treatment approach required with early detection of Wilms tumor. Little data is available regarding long-term outcomes.