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abstract

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TRIM28 variants and Wilms' tumour predisposition

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Introduction: TRIM28 was recently identified as germline pathogenic variants in Wilms' tumour (WT) that occurred in 1% of isolated and 8% of familial WT cases. WT with TRIM28 variants are associated with epithelial histology and there is no data on the phenotypic characteristics of patients. We describe the reviewing genotype, phenotype, tumour histology, laterality, treatment, patient survival, and kidney outcome

Methodology: Retrospective analysis of patients with WT and TRIM28 pathogenic variant treated at a single centre between September 2019 to December 2024

Results: Germline TRIM28 mutations were identified in four patients (3 boys and 1 girl) with WT by Next-Generation Sequencing. In all cases patients presented with bilateral disease. Median age at diagnosis -22,5 months (range 6-41) and the median tumour volume prior to neoadjuvant chemotherapy was 140 ml (range 17,5 - 308) on the right and 126 ml (range 20,5 - 606) on the left. One patient presented with metastatic WT. All patients had facial dysmorphism and malformations of the genitourinary system: 2–cryptorchidism, 1–anterior hypospadias and the girl was diagnosed with an anomaly in the development of renal vessels.

Two patients presented with cutaneous hemangiomas. Histological characterization was reported for 8 tumours from 4 patients. Out of the 8 tumours, 6 (75%) were described as (monomorphic) epithelial type WTs, one (12,5%) as epithelial predominant with diffuse anaplasia, one (12,5%) as nephroblastomatosis. One patient with epithelial predominant with diffuse anaplasia died from disease progression, 3 are alive, however 2 patients have stage II - III chronic kidney disease.

Conclusion: Recognizing germline TRIM28 variants in patients with WT can enable counselling, genetic testing, and potential early detection of WT in other children in the family. No phenotypic features have been previously described for patients with TRIM28, but our cohort of patients exhibits characteristic developmental anomalies that are found in WT and other syndromes.