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WAGR SYNDROME

Background: Wilms tumor, aniridia, genitourinary anomalies, and range of developmental delays (WAGR, OMIM 194072) is a rare contiguous gene deletion syndrome, caused by deletions of the 11p13 region. Particularly, deletions of the *PAX6* and *WT1* genes lead to the clinical manifestations of WAGR.

Assay: A custom comparative genomic hybridization and single nucleotide polymorphism (CGH + SNP) array designed using Agilent technologies is used to detect copy number changes in the targeted region (Chr11p13) as small as 96kb. Only copy number variants within the 11p13 region will be analyzed. The array can detect copy neutral loss of heterozygosity. The analysis of the array hybridization data for targeted gene(s) is performed using Cytogenomics software (Agilent Technologies).

Utility: Confirmation of a clinical diagnosis, determine appropriate medical management, identification of at risk family members.

Sensitivity: 90.9%

References: Duffy et al. Front Pediatr. 2021 Dec;14;9:733018.

Name of Test	TAT	Cost	CPT codes
WAGR Syndrome: 11p13 high resolution copy number analysis only (aCGH)	3-4 weeks	$750	81479