WAGR (Wilms' tumor/aniridia/genitourinary anomalies/mental retardation syndrome)

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Identity

Inheritance: Generally sporadic, a few inherited cases sometimes with milder phenotype were reported; occurrence: rare

Clinics

Phenotype and clinics
- High Wilms' tumor (WT) risk (can also manifest bilaterally),
- Aniridia (AN),
- Genitourinary anomalies (GU) (hypospadias and kryptorchism in males),
- Mental retardation,
- (Growth retardation).
Various combinations of these features can be present, partly depending on deletion extent.

Neoplastic risk
High.

Cytogenetics

Inborn conditions
del(11)(p13), contiguous gene syndrome with WT/GU and AN loci separated by about 700 kb; deletions may be cytogenetically invisible.

Cytogenetics of cancer
Deletions of the second chromosome 11 copy are rare; Wilms' tumors of WAGR patients frequently show subtle mutations of the remaining WT1 allele.

Genes involved and proteins

WT1 (Wilms' tumor suppressor gene)
Location
11p13
DNA/RNA
Description: 10 exons
Transcription: 3 kb mRNA; four alternative splice forms.

PAX6 (paired-homeodomain protein)
Location
11p13
DNA/RNA
Description: http://www.hgu.mrc.ac.uk/Softdata/PAX6/About/pax6c.dna.htm
Protein
Description: Paired-homeobox transcription factor; see http://www.hgu.mrc.ac.uk/Softdata/PAX6/About/about.htm.
Expression: Mainly eye, CNS and nasal development.
Localisation: Nuclear.
Function: Transcriptional regulator.
Homology: Pax gene family.

Mutations
Germinal: Mostly nonsense mutations; see (The Human PAX6 Mutation Database).
Somatic: Not known.

References

This article should be referenced as such: