WAGR syndrome

<table>
<thead>
<tr>
<th>Group</th>
<th>Sub group</th>
<th>Signs:</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td>GENITAL DISORDERS</td>
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<tr>
<td></td>
<td></td>
<td>genital dysfunctions</td>
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<tr>
<td></td>
<td></td>
<td>- hypogenitalism, hypogonadism; small testes, microorchidism, hypoplastic scrotum</td>
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<tr>
<td></td>
<td></td>
<td>genitalia, ambiguity</td>
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<tr>
<td></td>
<td></td>
<td>- ambiguous genitalia, male</td>
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<tr>
<td></td>
<td></td>
<td>male genitalia, modifications not including ambiguity</td>
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<tr>
<td></td>
<td></td>
<td>- cryptorchidism</td>
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<tr>
<td></td>
<td></td>
<td>- hypospadias</td>
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</tbody>
</table>

LABORATORY DATA
- chromosomal assignment
- chromosome 11p localization
- chromosomal disorders
- chromosomal microdeletions
- gene, structural-functional anomalies
- gene analysis-DNA analysis
- WT1 Wilms tumour-1, gene chr.11p13

MUSCULAR DISEASES
- muscular defects, distrectual
- hernia, inguinal

NEOPLASTIC DISEASES
- cancer, genetic features
- tumour familial trait

NEUROLOGICAL DISORDERS
- brain anomalies
- microcephaly, microcrania
- mental retardation
- mental retardation

OCULAR DISORDERS
- eye, motility defects
- nystagmus
- eyelids, anomalies
- eyelids, ptosis
- iris anomalies
- aniridia
- iris, coloboma
- lens, defects
- cataract, congenital
- cataract, no isolated defect
- cataract, unspecified type
- uvea, changes
- uvea, coloboma
- visus defects
- hyperopia, hypermetropy
- vitreous, defects including glaucoma
- glaucoma

Semeiological Synthesis: Facio-genito-neuro-ocular disorder. Wilms tumour, mental deficiency, hydrocephaly, ambiguous genitalia, dysmorphism, aniridia, congenital cataract/other ocular defects. Drash syndrome may be part of WAGR syndrome. 20% of patients develop renal failure.
OROCRANIOFACIAL ANOMALIES

* facies, modified appearance
  - dysmorphic face
  - facial dysmorphism due to cranial changes
    including microcephaly
* lips, modified appearance
  - lips, protruding, everted, full, thickened
* mandibular changes
  - micrognathia, mandibular hypoplasia, small jaw, not including: severe micrognathia, agnathia

OTHERS

* inheritance
  - inheritance, autosomal dominant
  - inheritance, chromosomic
* supergroups
  - facio-genito-neuro-skeletal disorders
  - facio-genito-skeleto-urinary

PRENATAL-NEONATAL MODIFIED DATA

* foetal changes
  - congenital tumour
  - foetal changes, recognized by laboratory data
  - foetal changes, recognized by ultrasound techniques
* prenatal diagnosis
  - prenatal diagnosis, echographic
  - prenatal diagnosis, molecular

SKELETAL DISORDERS

* fingers, modified form, deformity
  - clinodactyly
* limb anomalies, limb defects
  - isolated limb anomalies
* stature, growth, modified habitus
  - growth delayed, failure to thrive, growth retardation

UROLOGICAL DISORDERS

* kidney, malformations
  - kidney, tumour, renal tumour, including Wilms' tumour
* urinary tract-bladder, malformations
  - urinary tract malformations, unspecified type

Super group: dysmorphic face-mental retardation: ocular disorders
dysmorphic face-mental retardation: urogenital disorders
facio-genito-neuro-skeletal disorders
facio-genito-skeleto-urological disorders
facio-neuro-ocular disorders
facio-ocular disorders
genito-ocular disorders
neuro-urological disorders

Super aggreg. Aggregations: FOETAL CHANGES

* foetal changes, recognized by laboratory data
* foetal changes, recognized by ultrasound techniques

ISOLATED SKELETAL ANOMALIES NOT INCLUDING OSTEOCHONDRODYSPLASIAS

isolated limb anomalies

NEOPLASTIC DISORDERS

* tumour, congenital
* tumour, familial trait

OTHER

* cataract, not isolated defect
craniofacial dysmorphism due to cranial changes, including microcephaly
dysmorphic face
### Differential diagnosis:

<table>
<thead>
<tr>
<th>OMIM ID</th>
<th>Disorder Description</th>
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<tr>
<td>1720</td>
<td>aniridia-cerebellar ataxia-mental retardation syndrome</td>
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<tr>
<td>1740</td>
<td>aniridia-renal agenesis-mental retardation</td>
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<td>7800</td>
<td>Drash syndrome</td>
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<tr>
<td>26820</td>
<td>Wilms tumour I</td>
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<td>26821</td>
<td>Wilms tumour II</td>
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<td>Wilms tumour III</td>
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<td>28469</td>
<td>Wilms tumour IV</td>
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### Bibliography

- OMIM ID: 19407.0001
- OMIM ID: 194072
- OMIM ID: 137357
- OMIM ID: 607102

Smith’s Recognizable Patterns of Human Malformation. 6th Edition pag.52-53
Annuario Orphanet-Italia delle Malattie Rare 2005, pag. 817