Wilms tumour I

Eponyms:
- nephroblastoma
- WT1

Inheritance:
- autosomal dominant
- chromosomic
- supposed contiguous genes

Semeiological Synthesis:
Urological disorder, isolated defect. Fixed abdominal mass in an upper quadrant, hematuria, hypertension, fever, abdominal pain. Occasionally congenital form. Incidence: 1:10,000. Risk for subsequent children in affected member: parent affected with bilateral tumour: 30%; parent affected with unilateral tumour: 10%; parent unaffected with two affected children: 30%; sib with bilateral tumour: 10%; sib with unilateral tumour and no chromosome defect: 1%. Wilms’ tumour may also occur in syndromal association.

Group Sub group Signs:

CARDIOVASCULAR DISORDERS
- peripheral vessels changes
  - hypertension

GENITAL DISORDERS
- female genitalia, modified not including ambiguity
  - clitoris, hypertrophic, clitoromegaly
- male genitalia, modifications not including ambiguity
  - cryptorchidism
  - hypospadias

LABORATORY DATA
- adrenal cortex hormones
  - adrenal insufficiency, adrenal hypoplasia
- chromosomal assignment
  - chromosome 11p localization
  - chromosome 17q localization
  - chromosome 19q localization
  - chromosome X localization
  - chromosome Xq localization
- chromosomal disorders
  - chromosomal numerical and/or structural anomalies
  - gene, structural-functional anomalies
    - FWT1, gene chr.17q12-21
    - FWT2, gene chr.19q
    - gene analysis-DNA analysis
    - GPC3 (SDYS) (SGB) (SGBS1) glypican 3, gene chr.Xq26
    - WT1 Wilms tumour-1, gene chr.11p13
    - WTS1 Wilms tumour suppressor locus, gene chr.7p15-p11.2

NEOPLASTIC DISEASES
- cancer, genetic features
  - tumour familial trait

OCULAR DISORDERS
- lens, defects
  - cataract, congenital

OTHERS
- inheritance
  - inheritance, autosomal dominant
  - inheritance, chromosomic
- supergroups
  - cardio-genito-skeleto-urological disorders

PRENATAL-NEONATAL MODIFIED DATA
- foetal changes
  - congenital tumour
  - foetal changes, recognized by laboratory data
Super group: cardio-genito-skeleto-urological disorders

Super aggreg. FOETAL CHANGES
Aggregations: foetal changes, recognized by laboratory data
                  foetal changes, recognized by ultrasound techniques
NEOPLASTIC DISORDERS
tumour, congenital
tumour, familial trait
OTHER
microdeletion syndrome

Differential diagnosis:
1720 aniridia-cerebellar ataxia-mental retardation syndrome
1740 aniridia-renal agenesis-mental retardation
7800 Drash syndrome
8181 mesoblastic nephroma, congenital
27918 oncocytoma
12840 renal cell carcinoma
28577 renal cell carcinoma papillary 2
26580 WAGR syndrome
26821 Wilms tumour II
26822 Wilms tumour III
28469 Wilms tumour IV
28516 Yates syndrome

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