nephropathy-XY gonadal dysgenesis, Frasier type

Eponyms:  
- Frasier XY gonadal dysgenesis-nephropathy syndrome
- SRY

Inheritance:  
- sporadic

Semeiological Synthesis:  
Genito-urologic disorder. Male pseudohermaphroditism, XY gonadal dysgenesis, renal failure, occurrence of gonadoblastoma but not Wilms' tumour.

Group Sub group Signs:  
GENITAL DISORDERS  
genital dysfunctions  
- amenorrhea, primary  
genitalia, ambiguity  
- ambiguous genitalia, male  
- pseudohermaphroditisms, male  
ovarium, changes  
- ovarian tumour, including gonadoblastoma  
- ovarium cysts

LABORATORY DATA  
chromosomal assignment  
- chromosome 11p localization  
gene, structural-functional anomalies  
- gene analysis-DNA analysis  
- SRY (TDF) testis determining factor region Y, ribosomal protein S4, Y-linked (RPS4Y), gene chr.Yp11.3  
- WT1 Wilms tumour-1, gene chr.11p13  
urine, constituent changes  
- hematuria  
- proteinuria, albuminuria

NEOPLASTIC DISEASES  
cancer, genetic features  
- tumour susceptibility

OTHERS  
inheritance  
- inheritance, sporadic

PRENATAL-NEONATAL MODIFIED DATA  
foetal changes  
- foetal changes, recognized by ultrasound techniques  
prenatal diagnosis  
- prenatal diagnosis, echographic

UROLOGICAL DISORDERS  
kidney, malformations  
- kidney, small, underdeveloped, including renal agenesis  
renal dysfunctions  
- renal failure, including: nephritis, pyelonephritis, glomerulonephritis

Super group:

Super aggreg. Aggregations:  
FOETAL CHANGES  
foetal changes, recognized by ultrasound techniques

NEOPLASTIC DISORDERS  
tumour, susceptibility

PSEUDO-HERMAPHRODITISM  
pseudohermaphroditisms, male

Differential diagnosis:  
7800 Drash syndrome  
11210 gonadal dysgenesis, XY
28118  gonadoblastoma

**Bibliography**
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- Annuario Orphanet-Italia delle Malattie Rare 2005, pag.384