Rokitansky-Kuster syndrome

Eponyms:
- Mayer syndrome
- MRK anomaly
- R-K-H syndrome
- Rokitansky syndrome
- vaginal atresia/absence

Inheritance: supposed autosomal recessive

Semiological Synthesis: Genito-oto disorder. Vagina absence, rudimentary bipartitutus uterus; normal tubes, ovaries and secondary sexual characteristics. CFTR mutation are also associated with congenital absence of uterus and vagina.

Group Sub group Signs:

AUDITORY DISORDERS
deafness
- deafness conductive type
- deafness sensorineural, including unspecified type
- deafness, in syndromic association

GENITAL DISORDERS
female genitalia, modified not including ambiguity
- vagina, atresia, absence
genital dysfunctions
- amenorrhea, primary
uterus, anomalies
- mullerian duct, agenesis/defects
- uterus, absent
- uterus, anomalous including unicorns, bicornuate, biseptate, didelphys
- uterus, small, rudimentary

LABORATORY DATA
chromosomal assignment
- chromosome 1p localization
gene, structural-functional anomalies
- gene analysis-DNA analysis
- WNT4, gene chr.1p35

OROCRANIOFACIAL ANOMALIES
facies, modified appearance
- facies, asymmetric, unilateral atrophy, hemifacial atrophy

OTHERS
supergroups
- facio-genito-skeleto-urinary
- oto-skeleto-urological disorders

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

SKELETAL DISORDERS
radioulnar changes
- radius, short, hypoplastic radius
ribs, anomalies
- ribs, fusion
- ribs, short, hypoplastic
vertebral changes
- vertebral anomalies, unspecified type

UROLOGICAL DISORDERS
kidney, malformations
- kidney, malformation, including horseshoe
kidney
### Genus database

**Super group:**  
- urinary tract-bladder, malformations  
  - urinary tract malformations, unspecified type

#### Super agg.  
**DEAFNESS**  
Aggregations:  
- deafness, in syndromic association

#### FOETAL CHANGES  
- foetal changes, recognized by ultrasound techniques

#### OTHER  
- mullerian duct, agenesis/defects

#### Differential diagnosis:

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<th>Code</th>
<th>Condition</th>
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<td>12590</td>
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#### OMIM ID: 277000

Bibliography

- New Eng. J. Med. 351, 792-798, 2004