Schmitt-Gillenwater-Kelly syndrome

Eponyms:
- radial hypoplasia-triphalangeal thumbs-hypospadias-diastema

Inheritance:
supposed autosomal dominant

Semiological Synthesis:
Facio-genito-skeletal disorder. Tripalangeal nonopposable thumb, short forearms, radially deviated hands, mild hypospadias, upper central incisors diastema.

Group Sub group Signs:

GENITAL DISORDERS
- male genitalia, modifications not including ambiguity
- hypospadias

OROCRANIOFACIAL ANOMALIES
- teeth, modified structures
  - teeth, incisors: diastema, diastasis

OTHERS
- supergroups
  - genito-skeletal disorders

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

SKELETAL DISORDERS
- fingers, modified form, deformity
  - fingers, deviated, including: ulnar deviation
    - radioulnar changes
  - radius, short, hypoplastic radius
    - thumb-hallux, anomalies
      - triphalangism

Super group:
genito-skeletal disorders

Super aggreg. Aggregations:

FOETAL CHANGES
- foetal changes, recognized by ultrasound techniques

Differential diagnosis:
- 9310 Fanconi pancytopenia, type 1
- 11800 heart-hand II syndrome
- 11810 heart-hand III syndrome
- 22870 heart-hand syndrome IV
- 12280 Holt-Oram syndrome
- 22830 Roberts syndrome
- 28121 SC-phocomelia syndrome
- 24900 TAR syndrome

Bibliography
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