trisomy 22 syndrome

Eponyms: chromosome 22 trisomy

Inheritance: chromosomal

Semeiological Synthesis:
Complex plurimalformations. Chromosomal disorder. Severe growth retardation and multiple structural abnormalities, oligohydramnios. Unfrequently survival beyond the first trimester.

Group Sub group Signs: DERMATOLOGICAL DISORDERS

cutis, changes in appearance and/or features
- nuchal lucency, increased foetal nuchal translucency

GENITAL DISORDERS
genitalia, ambiguity
- pseudohermaphroditisms, male
male genitalia, modifications not including ambiguity
- testis dysgenesis

NEUROLOGICAL DISORDERS
brain anomalies
- brain hypodeveloped, brain atrophy, brain degeneration, encephalopathy, including cerebral palsy
cerebellum, changes
- cerebellum agenesis, hypoplasia

OROCRANIOFACIAL ANOMALIES
external ear malformations
- anotia, microtia
facies, modified appearance
- dysmorphic face

PRENATAL-NEONATAL MODIFIED DATA
extrafoetal membrane, changes
- oligohydramnios
foetal changes
- birth weight, low; foetal growth decreased, intrauterine growth retardation, intrauterine growth restriction, IUGR
prenatal diagnosis
- prenatal diagnosis, echographic

SKELETAL DISORDERS
femur, anomalies
- femur, short, absent, anomalous

Super group: complex plurimalformations

Super aggreg. Aggregations: FOETAL CHANGES

foetal changes, recognized by laboratory data
foetal changes, recognized by ultrasound techniques
LETHALITY
lethality, in chromosomal disorders

PSEUDO-HERMAPHRODITISM
pseudohermaphroditisms, male

Differential diagnosis: 4085 Emanuel syndrome
Aggregation(s) [in differential diagnosis]:
- lethality, in chromosomal disorders

Bibliography
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