chromosome 4p+ syndrome

Eponyms:

Inheritance: chromosomal


Group Sub group Signs:

DERMATOLOGICAL DISORDERS
- nail changes
  - nails and/or toenails, dysplastic, brittle nails, onychodystrophy

GENITAL DISORDERS
- breast, changes
  - nipples, widely spaced
- male genitalia, modifications not including ambiguity
  - cryptorchidism
  - hypospadias

JOINT DISORDERS
- joint, mobility reduction
  - camptodactyly

LABORATORY DATA
- chromosomal disorders
  - chromosomal numerical and/or structural anomalies

MUSCULAR DISEASES
- systemic muscular defects
  - hypotonia, muscular weakness, muscular hypoplasia, myastenia, not including atrophy
  - muscular hypertonia, contractures

NEUROLOGICAL DISORDERS
- brain anomalies
  - corpus callosum, agenesis
  - microcephaly, microcrania
- mental retardation
  - mental retardation
  - neurological dysfunctions
    - seizures, convulsions, epilepsy

OCULAR DISORDERS
- corneal defects not including dystrophy
  - microphthalmos (anteposterior globe diameter less than 20 mm, in adult), anophthalmos
  - uvea, changes
    - uvea, coloboma

OROCRANIOFACIAL ANOMALIES
- external ear malformations
  - ear lobe, malformed, unspecified type
- facies, modified appearance
  - facies, round; moon face, doll-like
- forehead-orbital region, changes
  - forehead, flat
  - supraorbital ridges, prominent
  - synophrys, eyebrows meeting across the midline
- lip-palate, cleft
  - lip and palate cleft, chelognatopalatoschisis
    - not including isolated palate cleft
- mandibular changes
- micrognathia, mandibular hypoplasia, small jaw, not including: severe micrognathia, agnathia

**neck, modified appearance**
- neck, short

**nose, modified appearance**
- nasal bridge, nasal root, depressed, flat, saddle-nose
- nose, large, broad, bulbous

**OTHERS**
**inheritance**
- inheritance, chromosomic

**supergroups**
- arthro-facio-skeletal disorders
- arthro-neuro-skeletal disorders
- cutaneous-facio-neuro-oculo disorders
- cutaneous-genito-neuro-skeletal disorders
- facio-genito-neuro-skeletal disorders

**PRENATAL-NEONATAL MODIFIED DATA**
**foetal changes**
- birth weight, low; foetal growth decreased, intrauterine growth retardation, intrauterine growth restriction, IUGR
- foetal changes, recognized by laboratory data
- foetal changes, recognized by ultrasound techniques

**prenatal diagnosis**
- prenatal diagnosis, cytogenetic
- prenatal diagnosis, echographic

**SKELETAL DISORDERS**
**fingers, modified form, deformity**
- clinodactyly

**hand-foot, changes**
- club foot, talipes, pes equinovarus

**ribs, anomalies**
- rbs, supernumerary

**spine, changes**
- scoliosis, kyphoscoliosis

**stature, growth, modified habitus**
- growth delayed, failure to thrive, growth retardation

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**Super group:** arthro-facio-skeletal disorders
arthro-neuro-skeletal disorders
complex plurimalformations
cutaneous-facio-neuro-ocular disorders
cutaneous-genito-neuro-skeletal disorders
dysmorphic face-mental retardation
facio-genito-neuro-skeletal disorders

**Super agreg. Aggregations:**
**FOETAL CHANGES**
foetal changes, recognized by laboratory data
foetal changes, recognized by ultrasound techniques

**Differential diagnosis:**
9943 chromosome 10q+ syndrome
9944 chromosome 11q-
9945 chromosome 13q- syndrome
9946 chromosome 15q+
9947 chromosome 18p-
9948 chromosome 18q-
9937 chromosome 3p- syndrome
9940 chromosome 4q- syndrome
9941 chromosome 9p- syndrome
9942 chromosome 9p+ syndrome

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chromosome 4p+ syndrome
28873  Pallister-Killian syndrome

Bibliography  Smith's Recognizable Patterns of Human Malformation. 5th Edition pag.40-41

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