Brachmann-De Lange-like syndrome

Eponyms:

Inheritance: supposed autosomal dominant

Semeiological Synthesis:

Group Sub group Signs:

DERMATOLOGICAL DISORDERS
- dermatoglyphics, changes
  - dermatoglyphics, fingertip atypical
  lanugo, modified appearance
  - hypertrichosis, hirsutism

NEUROLOGICAL DISORDERS
- brain anomalies
  - choroid plexus changes, including cysts
  - microcephaly, microcrania
  mental retardation
  - mental retardation

OROCRANIOFACIAL ANOMALIES
- facies, modified appearance
  - dysmorphic face
  - facial dysmorphism due to cranial changes including microcephaly
  forehead-orbital region, changes
  - forehead, hairy, hypertrichosis

OTHERS
- supergroups
  - facio-neuro-skeletal disorders

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

SKELETAL DISORDERS
- metacarpus-metatarsus changes
  - metacarpus metatarsus, unspecified anomalies
  stature, growth, modified habitus
  - growth delayed, failure to thrive, growth retardation

Super group: dysmorphic face-mental retardation
facio-neuro-skeletal disorders

Super aggreg. Aggregations:

FOETAL CHANGES
- foetal changes, recognized by ultrasound techniques

OTHER
- craniofacial dysmorphism due to cranial changes, including microcephaly dysmorphic face

Differential diagnosis:
6410 Cornelia de Lange syndrome

Bibliography
OMIM ID: 112370