bird headed dwarfism, Montreal type

Eponyms: — Montreal bird headed dwarfism

Inheritance: supposed autosomal recessive


Group Sub group Signs:

**DERMATOLOGICAL DISORDERS**
- cutis, changes in appearance and/or features
  - cutis, redundant, loose, including cutis laxa
  - cutis, wrinkled
  - hair, changes
  - hair, low set; low hairline
  - hair, sparse not including alopecia totalis
  - lanugo, modified appearance
  - hypotrichia, hypotrichosis
  - subcutaneous changes
  - subcutaneous, scarce, thin

**GENITAL DISORDERS**
- breast, changes
- nipples, widely spaced
- genital dysfunctions
  - hypogonitalism, hypogonadism; small testes, microorchidism, hypoplastic scrotum
  - male genitalia, modifications not including ambiguity
  - cryptorchidism
  - macropenis

**MUSCULAR DISEASES**
- muscular defects, disrectual
  - hernia, inguinal

**NEUROLOGICAL DISORDERS**
- brain anomalies
  - microcephaly, microcrania
  - mental retardation
  - mental retardation

**OCULAR DISORDERS**
- eye, motility defects
  - nystagmus
- eyelids, anomalies
  - eyelids, ptosis

**OROCRANIOFACIAL ANOMALIES**
- cutis, vascular changes
  - erythema
  - external ear malformations
    - ear lobe, malformed, unspecified type
    - ear, low set
- facies, modified appearance
  - dysmorphic face
  - facial dysmorphism due to cranial changes including microcephaly
  - facies, beard absence
  - facies, senile, folded progeroid facies, premature aging, pinched
- mandibular changes
  - micrognathia, mandibular hypoplasia, small jaw, not including: severe micrognathia,agnathia
  - nose, modified appearance
    - nose, hooked, parrot, beaked
teeth, modified structures
- teeth, carious

OTHERS
supergroups
- 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 ultrasound techniques

prenatal diagnosis
- prenatal diagnosis, echographic

SKELETAL DISORDERS
bones, lesions, structural changes
- osteoporosis

limb anomalies, limb defects
- asymmetric growth, hemihypertrophy, limbs asymmetry

ossification, changes
- osteopenia, bone hypodensity, demineralization of the bones, deficient ossification

stature, growth, modified habitus
- growth delayed, failure to thrive, growth retardation
- stature, short, including micromelia, including short limbs

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

Super aggreg. Aggregations: foetal changes, recognized by ultrasound techniques

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

Differential diagnosis:
2814 Bangstad syndrome
5930 Cockayne I syndrome
5931 Cockayne II syndrome
11570 Hallermann-Streiff syndrome
27704 Kozlowski-Turner syndrome
28794 Okamoto syndrome
19470 osteodysplastic primordial dwarfism I
19480 osteodysplastic primordial dwarfism II
19490 osteodysplastic primordial dwarfism III
21590 progeria
23610 Seckel syndrome 1-2
26720 Werner syndrome

Bibliography OMIM ID: 210700