hypospadias-mental retardation syndrome

Eponyms: — Goldblatt-Wallis-Viljoen syndrome

Inheritance: genetic heterogeneity
supposed autosomal recessive
supposed X-linked recessive

Semeiological Synthesis: Cutaneous-genital-facial disorder. Mental retardation, dysmorphic face, hypospadias, other clinical data.

Group Sub group Signs:

DERMATOLOGICAL DISORDERS
nail changes
- nails and/or toenails, hyperconvex, nails
clawlike, watshglass

GENITAL DISORDERS
male genitalia, modifications not including ambiguity
- hypospadias

JOINT DISORDERS
joint, laxity, dislocations
- joint, laxity, hyperlaxity, hypermobility

LABORATORY DATA
chromosomal assignment
- chromosome X localization

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

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

OROCRANIOFACIAL ANOMALIES
external ear malformations
- ear lobe, folded, crumpled, overfolded, fleshy
- ear, post angulation, posteriorly rotated
facies, modified appearance
- dysmorphic face
- facial dysmorphism due to cranial changes including microcephaly
forehead-orbital region, changes
- synophrys, eyebrows meeting across the midline
maxilla-cheek changes
- midface hypoplasia, malar hypoplasia, hypoplastic zygomatica
nose, modified appearance
- nose, hooked' parrot, beaked

OTHERS
inheritance
- inheritance, genetic heterogeneity
supergroups
- arthro-facio-skeletal disorders
- arthro-neuro-skeletal disorders
- cutaneous-genito-neuro-skeletal disorders
- facio-genito-neuro-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
- clinodactyly
skull shape, changes
- trigonocephaly, metopic ridging

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

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

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

Differential diagnosis:
10260 G syndrome
11720 Harrod-Keele-Howard syndrome
27458 Marden-Walker like syndrome
27694 Mizoguchi-Nishimura-Honda syndrome
16930 Morillo-Cucci syndrome
28553 Opitz syndrome, type I
27612 Toriello-Kienbaum syndrome
25886 trigono-brachycephaly, Teebi type

Bibliography OMIM ID: 241760