floating-harbor syndrome

Eponyms: sporadic

Inheritance: sporadic

Semeiological Synthesis: Arthro-cardio-facio-neurological disorder. Proportionate short stature, delayed bone maturation, dysmorphic face, bulbous nose, significantly delayed speech, mild mental retardation, hirsutism, joint laxity, other clinical data.

Group Sub group Signs:

CARDIOVASCULAR DISORDERS
- cardiac congenital defects
  - pulmonary artery-valve, anomalies

DERMATOLOGICAL DISORDERS
- lanugo, modified appearance
  - hypertrichosis, hirsutism
  - pigmentation changes
  - cutis, hyperpigmentation

GASTROINTESTINAL DISORDERS
- intestinal dysfunctions
  - malabsorption, including diarrhea

GENITAL DISORDERS
- male genitalia, modifications not including ambiguity
  - hypospadias

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

NEUROLOGICAL DISORDERS
- performance changes, not including mental retardation
  - speech dyspraxia, including speech delayed

OROCRANIOFACIAL ANOMALIES
- external ear malformations
  - ear, low set
  - facies, modified appearance
    - dysmorphic face
    - eye, deep, sunken, enophthalmos
    - facies, small, triangular, narrow
    - proptosis, prominent eyes, exophthalmos, protuberant eyes, protruding eyes
  - lips, modified appearance
    - lips, mouth, downturned corners
  - mouth, modified appearance
    - mouth, large, wide mouth, macrostomia
  - neck, modified appearance
    - neck, short
  - nose, modified appearance
    - nose, large, broad, bulbous
    - nose, philtrum short
    - nose, prominent, long
    - nose, protruding columella
  - teeth, modified structures
    - teeth, supernumerary

OTHERS
- inheritance
  - inheritance, sporadic

supergroups
- arthro-facio-respiratory-skeletal disorders
- arthro-facio-skeletal disorders
- arthro-neuro-skeletal disorders
- cutaneous-genito-neuro-skeletal disorders
- cutaneous-genito-neuro-urological disorders
- facio-genito-neuro-skeletal disorders
- facio-genito-skeleto-urinary

PRENATAL-NEONATAL MODIFIED DATA
- foetal changes
  - birth length, decreased; low birth length
  - birth weight, low; foetal growth decreased, intrauterine growth retardation, intrauterine growth restriction, IUGR
  - foetal changes, recognized by ultrasound techniques
- prenatal diagnosis
  - prenatal diagnosis, echographic

RESPIRATORY DISORDERS
- larynx, changes
  - voice, high; pitched voice
  - voice, nasal

SKELETAL DISORDERS
- brachydactyl
  - brachydactyly, brachyphalangy, in syndromic association
- fingers, modified form, deformity
  - clinodactyl
- ossification, changes
  - bone age, delayed
- skull shape, changes
  - trigonocephaly, metopic ridging
- stature, growth, modified habitus
  - growth delayed, failure to thrive, growth retardation
  - stature, short, including micromelia, including short limbs

UROLOGICAL DISORDERS
- kidney, malformations
  - kidney, ectopic

Super group:
- arthro-facio-respiratory-skeletal disorders
- arthro-neuro-skeletal disorders
- cutaneous-genito-neuro-skeletal disorders
- cutaneous-genito-neuro-urological disorders
- facio-genito-neuro-skeletal disorders
- facio-genito-skeleto-urological disorders

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

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

Differential diagnosis:
- 4730  celiac disease
- 7830  Dubowitz syndrome
- 16820 MMM syndrome
- 23820 Russell-Silver syndrome
- 23230 Saul-Wilson syndrome
- 23760 SHORT syndrome

Bibliography
- OMIM ID: 136140
- Smith’s Recognizable Patterns of Human Malformation. 6th Edition pag.144-145