Barber-Say syndrome

Eponyms:——Barber-Cesarino syndrome

Inheritance:——autosomal dominant

supposed genetic heterogeneity

Semiological Synthesis:Cutaneous-facio-ocular disorder. Bilateral lid absence, corneal and conjunctiva keratinization, macrostomia, hypertrichosis, wide nose, thin lips, redundant face-skin. Dysmorphic face, hustatism, macrostomia, ectropion, atrophic skin, other clinical data.

<table>
<thead>
<tr>
<th>Group</th>
<th>Sub group</th>
<th>Signs</th>
<th>AUDITORY DISORDERS</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td></td>
<td>deafness</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>deafness sensorineural, including unspecified type</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>DERMATOLOGICAL DISORDERS</th>
</tr>
</thead>
<tbody>
<tr>
<td>cutis, changes in appearance and/or features</td>
</tr>
<tr>
<td>cutis, redundant, loose, including cutis laxa</td>
</tr>
<tr>
<td>cutis, dysplastic, not including ectodermal dysplasia</td>
</tr>
<tr>
<td>cutis, aplasia, skin atrophy</td>
</tr>
<tr>
<td>hair, changes</td>
</tr>
<tr>
<td>hair, coarse</td>
</tr>
<tr>
<td>lanugo, modified appearance</td>
</tr>
<tr>
<td>hypertrichosis, hustatism</td>
</tr>
</tbody>
</table>

GENITAL DISORDERS

breast, changes

nipples: inverted, absent, hypoplastic
male genitalia, modifications not including ambiguity

- hypospadias
- scrotum, shawl, penoscrotal inversion

LABORATORY DATA

urine, constituent changes

- aminoaciduria

MUSCULAR DISEASES

systemic muscular defects

- hypotonia, muscular weakness, muscular hypoplasia, myastenia, not including atrophy

NEUROLOGICAL DISORDERS

mental retardation

OCULAR DISORDERS

corneal defects not including dystrophy

corneal clouding

eyelids, anomalies

ablepharon, cryptophthalmos, absent eyelids

ectropion, including lagophthalmos

eyelashes, sparse, madarosis

lacrimal gland-sac, defects

tears, absence, reduced lacrimation

OROCRANIOFACIAL ANOMALIES

external ear malformations

- ear lobe, flopping
- ear lobe, hypoplastic, small, poorly shaped, dysplastic pinnae
- ear lobe, malformed, unspecified type
- ear, external canal atresia, stenosis
- ear, low set

facies, modified appearance

dysmorphic face

facies, coarse
forehead-orbital region, changes
  - forehead, hairy, hypertrichosis
  - hypertelorism
lips, modified appearance
  - lower lip, thin
mouth, modified appearance
  - mouth, large, wide mouth, macrostomia
nose, modified appearance
  - nasal bridge, nasal root, broad, wide, squared
  - nasal bridge, nasal root, depressed, flat, saddle-nose
  - nose, flat
  - nose, large, broad, bulbous
palatopharyngeal changes
  - palate cleft, palatoschisis, including submucous, not including lip and palate cleft
teeth, modified structures
  - teeth at birth, including prematural eruption

OTHERS
inheritance
  - inheritance, autosomal dominant
supergroups
  - cutaneous-facio-neuro-ocular disorders
  - cutaneous-genito-neurop-skeletal disorders
  - cutaneous-ocular-oto-skeletal disorders
  - facio-genito-neuro-skeletal disorders
  - facio-genito-ocular-oto 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
stature, growth, modified habitus
  - stature, short, including micromelia, including short limbs

Super group: cutaneous-facio-neuro-ocular disorders
  cutaneous-genito-neuro-skeletal disorders
  cutaneous-ocular-oto-skeletal disorders
dysmorphic face-mental retardation: dermatological disorders
dysmorphic face-mental retardation: ocular disorders
  facio-genito-neuro-skeletal disorders
  facio-genito-ocular-oto disorders

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

Differential diagnosis: 50 ablepharon-macrostomia syndrome
  10070 Fraser syndrome

Bibliography
Synd.Ident.8(1),6-9,1982
OMIM ID: 209885
Annuario Orphanet-Italia delle Malattie Rare 2005, pag.117