Pashayan syndrome

Eponyms:

Inheritance: sporadic

Semeiological Synthesis: Facio-neuro-oculo-skeletal disorder. Dysmorphic face, ocular anomalies, mental retardation, brachydactyly, other skeletal changes, obesity, other clinical findings.

<table>
<thead>
<tr>
<th>Group</th>
<th>Sub group</th>
<th>Signs</th>
</tr>
</thead>
<tbody>
<tr>
<td>DERMATOLOGICAL DISORDERS</td>
<td></td>
<td>hair, changes</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- hair, low set; low hairline</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- hair, sparse not including alopecia totalis</td>
</tr>
<tr>
<td>ENDOCRINOLOGICAL, METABOLIC DISORDERS</td>
<td></td>
<td>obesity</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- obesity, total</td>
</tr>
<tr>
<td>GENITAL DISORDERS</td>
<td></td>
<td>breast, changes</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- nipples, widely spaced</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- nipples: inverted, absent, hypoplastic</td>
</tr>
<tr>
<td>NEUROLOGICAL DISORDERS</td>
<td></td>
<td>mental retardation</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- mental retardation</td>
</tr>
<tr>
<td>OCULAR DISORDERS</td>
<td></td>
<td>eye, motility defects</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- nystagmus</td>
</tr>
<tr>
<td></td>
<td></td>
<td>eyelids, anomalies</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- blepharophymosis</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- eyelashes, sparse, madarosis</td>
</tr>
<tr>
<td></td>
<td></td>
<td>vitreous, defects including glaucoma</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- glaucoma</td>
</tr>
<tr>
<td>OROCRANIOFACIAL ANOMALIES</td>
<td></td>
<td>chin, changes</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- chin, fissured fovea</td>
</tr>
<tr>
<td></td>
<td></td>
<td>external ear malformations</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- ear lobe, hypoplastic, small, poorly shaped, dysplastic pinnae</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- ear lobe, malformed, unspecified type</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- ear, external canal atresia, stenosis</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- ear, low set</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- ear, post angulation, posteriorly rotated</td>
</tr>
<tr>
<td></td>
<td></td>
<td>facies, modified appearance</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- dysmorphic face</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- eye, deep, sunken, enophthalmos</td>
</tr>
<tr>
<td></td>
<td></td>
<td>forehead-orbital region, changes</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- epicanthal</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- eyelids, fissures, upslanting, mongolid slant</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- hypertelorism</td>
</tr>
<tr>
<td></td>
<td></td>
<td>lips, modified appearance</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- lips, mouth, downturned corners</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- lower lip, thin</td>
</tr>
<tr>
<td></td>
<td></td>
<td>mandibular changes</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- micrognathia, mandibular hypoplasia, small jaw, not including: severe micrognathia, agnathia</td>
</tr>
<tr>
<td></td>
<td></td>
<td>neck, modified appearance</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- neck, short</td>
</tr>
<tr>
<td></td>
<td></td>
<td>pterygium colli, webbed neck, including: thickened nuchal fold, nuchal thickening</td>
</tr>
<tr>
<td></td>
<td></td>
<td>nose, modified appearance</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- nasal bridge, nasal root, depressed, flat, saddle-nose</td>
</tr>
</tbody>
</table>
nose, philtrum flat, smooth, poorly developed
- nose, pointed, anteverted nostrils
- nose, small, nose tiny, nose short, not including very small
  teeth, modified structures
- microdontia, hypodontia; teeth, hypoplasia, peg shaped, conical shaped

OTHERS
inheritance
- inheritance, sporadic

supergroups
- cutaneous-facio-neuro-ocular disorders
- cutaneous-genito-neuro-skeletal disorders
- endocrino-neuro 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
brachydactyly
- brachydactyly, brachyphalangy, in syndromic association

fingers, modified form, deformity
- clinodactyly

ribs, anomalies
- ribs, short, hypoplastic

shoulders, changes
- scapulae, winged, hypoplastic
- Sprengel anomaly

skull shape, changes
- platybasia, occiput flat

Super group: cutaneous-facio-neuro-ocular disorders
cutaneous-genito-neuro-skeletal disorders
dysmorphic face-mental retardation: ocular disorders
dysmorphic face-mental retardation: skeletal disorders
endocrino-neuro disorders
facio-genito-neuro-skeletal disorders
facio-oculo-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:
13700 Kabuki make-up syndrome

Bibliography Synd.Ident.1(2),12-13,1973