oculo-palato-cerebral syndrome

Eponyms:
- Frydman syndrome
- OPC dwarfism
- persisten hyperplastic primary vitreous
- PHPV

Inheritance: supposed autosomal recessive

Semeiological Synthesis:
Arthro-neuro-oculo-orofacial disorder. Primary vitreous persistence, cataract, other ocular defects, mental retardation, microcephaly, cleft palate, spastic quadriplegia, short stature, joint hypermobility. Occasionally associated PHPV.

Group Sub group Signs:

AUDITORY DISORDERS
- deafness
  - deafness sensorineural, including unspecified type
  - deafness, in syndromic association

GENITAL DISORDERS
- genital dysfunctions
  - hypogenitalism, hypogonadism; small testes, microorchidism, hypoplastic scrotum
  - male genitilia, modifications not including ambiguity
  - cryptorchidism

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

MUSCULAR DISEASES
- muscular defects, distrectual
  - hernia, umbilical, navel

NEUROLOGICAL DISORDERS
- brain anomalies
  - microcephaly, microcrania
  - mental retardation
  - mental retardation
  - neurological dysfunctions
  - spasm, spastic paralysis, spastic paraplegia

OCULAR DISORDERS
- corneal defects not including dystrophy
  - corneal clouding
    - microphthalmos (anteposterior globe diameter less than 20 mm, in adult), anophthalmos
  - lens, defects
    - cataract, no isolated defect
    - cataract, unspecified type
  - vitreous, defects including glaucoma
    - glaucoma
    - vitreous, primary persistence

OROCRANIOFACIAL ANOMALIES
- facies, modified appearance
  - dysmorphic face
  - facial dysmorphism due to cranial changes including microcephaly
  - palatopharyngeal changes
    - palate cleft, palatoschisis, including submucous, not including lip and palate cleft

OTHERS
- supergroups
  - arthro-facio-neuro-oculo-osto-skeletal disorders
### Genus database

Copyright V. Ventruto / A. Di Luccio

Super group:  
- arthro-neuro-oculo-oto-skeletal disorders  
- arthro-neuro-skeletal disorders  
- arthro-oculo-oto disorders  
- facio-genito-neuro-skeletal disorders  
- facio-genito-oculo-oto disorders  
- facio-neuro-oculo-oto disorders  
- facio-neuro-oculo-skeletal disorders  

PRENATAL-NEONATAL MODIFIED DATA  

<table>
<thead>
<tr>
<th>foetal changes</th>
<th>foetal changes, recognized by ultrasound techniques</th>
</tr>
</thead>
<tbody>
<tr>
<td>prenatal diagnosis</td>
<td>prenatal diagnosis, echographic</td>
</tr>
</tbody>
</table>

SKELETAL DISORDERS  

<table>
<thead>
<tr>
<th>skull shape, changes</th>
<th>craniostenosis</th>
</tr>
</thead>
<tbody>
<tr>
<td>stature, growth, modified habitus</td>
<td>growth delayed, failure to thrive, growth retardation</td>
</tr>
<tr>
<td>stature, short, including micromelia, including short limbs</td>
<td></td>
</tr>
</tbody>
</table>

Super group:  
- arthro-facio-neuro-oculo-oto-skeletal disorders  
- arthro-neuro-skeletal disorders  
- arthro-oculo-oto disorders  
- dysmorphic face-mental retardation: deafness  
- facio-genito-neuro-skeletal disorders  
- facio-genito-oculo-oto disorders  
- facio-neuro-oculo-skeletal disorders  

Super aggreg.  
DEAFNESS  
    deafness, in syndromic association  

FOETAL CHANGES  
    foetal changes, recognized by ultrasound techniques  

OTHER  
    cataract, not isolated defect  
    craniofacial dysmorphism due to cranial changes, including microcephaly  
    dysmorphic face  

Differential diagnosis:  

<table>
<thead>
<tr>
<th>OMIM ID</th>
<th>Condition</th>
</tr>
</thead>
<tbody>
<tr>
<td>27531</td>
<td>microphthalmia-clefting-mental retardation syndrome</td>
</tr>
<tr>
<td>18460</td>
<td>Norrie disease</td>
</tr>
<tr>
<td>26505</td>
<td>persistent hyperplastic primary vitreous</td>
</tr>
<tr>
<td>9717</td>
<td>spastic paraplegia, hereditary-9</td>
</tr>
</tbody>
</table>

Bibliography  
OMIM ID: 257910