Setleis syndrome

Eponyms:
- aplasia cutis congenita-bitemporal "forceps marks"
- ectodermal dysplasia facial
- temporal "forceps marks" scarring and unusual facies

Inheritance:
autosomal recessive

Semeiological Synthesis:
Cutaneous-orofacial disorder. Leonine appearance, pucker skin about the eyes, absent eyelashes, multiple rows on the upper lids, nose/chin rubbery feel, bilateral temporal marks forceps/like, other anomalies.

Group Sub group Signs:
DERMATOLOGICAL DISORDERS
- cutis, changes in appearance and/or features
  - isolated dermatological disorders
  - cutis, dysplastic, not including ectodermal dysplasia
  - cutis, aplasia, skin atrophy
  - cutis, scarring
  - ectodermal dysplasia
  - ectodermal dysplasia, syndactyly
  - hair, changes
  - hair, sparse not including alopecia totalis

GASTROINTESTINAL DISORDERS
- anorectal anomalies
  - anal/anorectal atresia, malformation, imperforate anus, stenosis

GENITAL DISORDERS
- breast, changes
  - nipples, supernumerary, polythelia
  - female genitalia, modified not including ambiguity
  - vagina, duplicated
  - uterus, anomalies
  - mullerian duct, agenesis/defects
  - uterus, anomalous including unicornis, bicornuate, biseptate, didelphys

OCULAR DISORDERS
- conjunctiva, changes
  - conjunctiva: infections, conjunctivitis
  - eyelids, anomalies
    - blepharophimosis
    - distichiasis, accessory rows of lashes
    - eyelashes, sparse, madarosis

OROCRANIOFACIAL ANOMALIES
- chin, changes
  - chin, fissured fovea
  - facies, modified appearance
  - dysmorphic face
  - facies, coarse
  - forehead-orbital region, changes
    - eyebrows, sparse laterally
    - frontal bossing
    - temporal dermal dysplasia
    - wrinkled eyelids,
      - lips, modified appearance
    - lower lip, thick
      - mouth, modified appearance
    - mouth, large, wide mouth, macrostomia
  - nose, modified appearance
    - nose, large, broad, bulbous

OTHERS
**inheritance**
- inheritance, autosomal recessive

**supergroups**
- cutaneous-facio-neuro-ocular disorders
- cutaneous-genito-neuro-skeletal disorders
- cutaneous-genito-neuro-urological disorders
- facio-gastrointestinal-neuro-ocular-skeletal disorders
- facio-genito-neuro-skeletal disorders
- facio-genito-skeleto-urinary
- gastrointestinal-oculo-skeletal disorders

**PRENATAL-NEONATAL MODIFIED DATA**
*foetal changes*
- foetal changes, recognized by ultrasound techniques

*prenatal diagnosis*
- prenatal diagnosis, echographic

**SKELETAL DISORDERS**
*stature, growth, modified habitus*
- growth delayed, failure to thrive, growth retardation

**UROLOGICAL DISORDERS**
*urinary tract-bladder, malformations*
- urinary tract malformations, unspecified type

<table>
<thead>
<tr>
<th>Super group:</th>
<th>cutaneous-facio-neuro-ocular disorders</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>cutaneous-genito-neuro-skeletal disorders</td>
</tr>
<tr>
<td></td>
<td>cutaneous-genito-neuro-urological disorders</td>
</tr>
<tr>
<td></td>
<td>facio-gastrointestinal-neuro-ocular-skeletal disorders</td>
</tr>
<tr>
<td></td>
<td>facio-genito-neuro-skeletal disorders</td>
</tr>
<tr>
<td></td>
<td>facio-genito-skeleto-urinary</td>
</tr>
<tr>
<td></td>
<td>gastrointestinal-oculo-skeletal disorders</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Super aggreg.</th>
<th>ECTODERMAL DYSPHASIA</th>
</tr>
</thead>
<tbody>
<tr>
<td>Aggregations:</td>
<td>ectodermal dysplasia, hydrotic</td>
</tr>
</tbody>
</table>

**FOETAL CHANGES**
foetal changes, recognized by ultrasound techniques

**OTHER**
dysmorphic face
isolated dermatological disorders
mullerian duct, agenesis/defects

<table>
<thead>
<tr>
<th>Differential diagnosis:</th>
<th>2008 aorticocardiac fistula</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>2040 aplasia cutis, congenita</td>
</tr>
<tr>
<td></td>
<td>9875 focal facial dermal dysplasia</td>
</tr>
</tbody>
</table>

**Aggregation(s) [in differential diagnosis]:**
- ectodermal dysplasia, hydrotic

**Bibliography**
OMIM ID: 227260