Genus database
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popliteal pterygium syndrome

**Eponyms:**
- cleft lip/palate paramedian-mucous cysts-popliteal pterygium
- facio-genito-popliteal syndrome
- lip-pits lower paramedian/popliteal pterygium
- PPS

**Inheritance:**
- autosomal dominant
- autosomal recessive
- genetic heterogeneity

**Semeiological Synthesis:**
Arthro-cutaneous-facio-ocular disorder. Pterigia from the heel to the ischial tuberosity, intercrural pterygium, cleft lip/palate, lower lip pits/fistula, ankyloblepharon, including ankyloblepharon filiforme adnatum (areas of fusion between the upper and lower lids by strands of fibrovascular tissue), genital anomalies, clitoromegaly, labia hypertrophy, flexion contractures. The severe, lethal form is autosomal recessive.

**Group**
**Sub group**
**Signs:**

**DERMATOLOGICAL DISORDERS**
- nail changes
  - nails and/or toenails, dysplastic, brittle nails, onychodystrophy
  - pterygium, webbing
  - popliteal webs
  - pterygium intercrural

**GENITAL DISORDERS**
- female genitalia, modified not including ambiguity
  - clitoris, hypertrophic, clitoromegaly
  - labia, hypertrophy
- genital dysfunctions
  - hypogenitalism, hypogonadism; small testes, microorchidism, hypoplastic scrotum
- genitalia, ambiguity
  - ambiguous genitalia, male
  - pseudohermaphroditisms, male
- male genitalia, modifications not including ambiguity
  - cryptorchidism
  - micropenis
  - scrotum, bifid
  - scrotum: absent, rudimentary

**JOINT DISORDERS**
- joint, mobility reduction
  - contractures, joint stiffness, not including: arthrogriposis, camptodactyly

**LABORATORY DATA**
- chromosomal assignment
  - chromosome 1q localization
- gene, structural-functional anomalies
  - gene analysis-DNA analysis
- PIT (LPS) (VWS) (IRF6) (PPS) van der Woude syndrome (lip pit syndrome), gene chr.1q32-q41

**MUSCULAR DISEASES**
- muscular defects, distrectual
  - hernia, inguinal

**NEUROLOGICAL DISORDERS**
- mental retardation
  - mental retardation
- spinal cord, changes
  - spina bifida occulta, including dysrafism

**OCULAR DISORDERS**
- eyelids, anomalies
- eyelids, fusion, adhesion, ankyloblepharon
  including ankyloblepharon filiforme adnatum

**OROCRANIOFACIAL ANOMALIES**

- *facies, modified appearance*
  - dysmorphic face
- *lip-palate, cleft*
  - lip and palate cleft, cheilognatopalatoschisis
    not including isolated palate cleft
- *lips, modified appearance*
  - lower lip, fistula, pits
- *oral mucous membranes, changes*
  - oral frenula, hypertrophy, multiple frenula
- *palatopharyngeal changes*
  - palate cleft, palatoschisis, including
    submucous, not including lip and palate cleft
  - uvula, bifida, cleft

**OTHERS**

- *inheritance*
  - inheritance, autosomal dominant
- *supergroups*
  - arthro-facio-skeletal disorders
  - arthro-neuro-skeletal disorders
  - cutaneous-facio-neuro-oculo disorders
  - cutaneous-genito-neuro-skeletal 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**

- *fibular, defects*
  - fibula, dislocated
- *fingers, defects*
  - ectrodactyly, adactyly, oligodactyly
  - fingers, modified form, deformity
  - fingers, destruction, mutilation fingers,
    constrictions fingers
- *hand-foot, changes*
  - club foot, talipes, pes equinovarus
  - foot calcaneovalgus, equinovalgus, foot
    abducted
- *hip, anomalies*
  - acetabulum, anomalous, including protrusion
- *patella, changes*
  - patella, absent, hypoplastic, small, stippling
- *skull shape, changes*
  - craniostenosis
- *spine, changes*
  - scoliosis, kyphoscoliosis
- *syndactyly*
  - syndactyly, in syndromic association
- *thumb-hallux, anomalies*
  - hallux, pyramid of skin overlying the nail
- *tibia, anomalies*
  - tibia, hypoplastic, short
- *vertebral changes*
  - vertebral anomalies, unspecified type

**Super group:**

- arthro-facio-skeletal disorders
- arthro-neuro-skeletal disorders
**Super aggreg. Aggregations:**

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

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

- **PSEUDO-HERMAPHRODITISM**
  - pseudohermaphroditisms, male

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**Bibliography**

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- Baraitser-Winter: Congenital Malformation Syndromes Mosby&Wolfe Ed. 1996, pag.75-76
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