**Eponyms:**
- ALPL
- HOPS
- hypophosphatasia, lethal form

**Inheritance:** autosomal recessive

**Semiological Synthesis:**
Endocrino-metabolic-skeletal disorder. Bone formation defects, almost complete lack of mineralization, intrauterin fractures, increased urinary phosphatides.

**Group**

**Sub group**

**Signs:**

**LABORATORY DATA**
- chromosomal assignment
- chromosome 1p localization
- foetal-amniotic biochemical data
- phosphatase alkaline, low levels (foetal)
- gene, structural-functional anomalies
- ALPL (HOPS) alkaline phosphatase
- liver/bone/kidney, tissue non-specific alkaline phosphatase (TNSALP), gene chr.1p36.1-p34
- gene analysis-DNA analysis

**myelo-erythropoietic disorders**
- anaemia, no haemolytic
- plasma electrolytes-inorganic constituents, modified functions
- calcium, high levels (hypercalcemia)
- plasma, enzymes, modified functions
- phosphatase alkaline, low levels
- tissue, biochemical changes
- collagen biosynthesis/structure disorders
- collagen disorders, not including Ehlers Danlos and osteogenesis imperfecta
- urine, constituent changes
- lipiduria
- phosphoethanolamine, high levels

**MUSCULAR DISEASES**
- systemic muscular defects
- hypotonia, muscular weakness, muscular hypoplasia, myastenia, not including atrophy

**NEUROLOGICAL DISORDERS**
- behaviour, changes
- aggressivity, irritability, hyperactivity
- brain anomalies
- microcephaly, microcrania
- neurological dysfunctions
- seizures, convulsions, epilepsy

**OCULAR DISORDERS**
- sclera, changes
- sclerae blue

**OROCRANIOFACIAL ANOMALIES**
- facies, modified appearance
- dysmorphic face
- facial dysmorphism due to cranial changes including microcephaly
- teeth, modified structures
- teeth, early loss
- teeth, enamel defects not including amelogenesis imperfecta

**OTHERS**
- inheritance
- inheritance, autosomal recessive

**PRENATAL-NEONATAL MODIFIED DATA**
foetal changes
- birth length, decreased; low birth length
- foetal changes, recognized by laboratory data
- foetal changes, recognized by ultrasound techniques
- foetal fractures, fractures in utero

prenatal diagnosis
- prenatal diagnosis, echographic
- prenatal diagnosis, metabolic
- prenatal diagnosis, molecular

RESPIRATORY DISORDERS
respiratory distress
- respiratory distress, including asthma

SKELETAL DISORDERS
bones, lesions, structural changes
- bone, fractures not including: fractures in utero
- osteomalacia
fontanelles-sutures, changes
- fontanelles, cranial sutures, closure delayed
limb anomalies, limb defects
- bone, bowing, limbs bowed, camptomelia
- lethality, constant in osteochondrodysplasia
- limbs deformity
metaphyseal changes
- metaphyseal changes not including: dumbbell-like erlenmeyer-like shaped
ossification, changes
- bone, mineralization disorders not including osteogenesis imperfecta
- osteopenia, bone hypodensity, demineralization of the bones, deficient ossification
periosteal changes
- periosteal cloaking, rickets changes
ribs, anomalies
- ribs, short, hypoplastic
skull shape, changes
- craniosynostosis
- skull base, short
skull, lesions, mineralization changes
- skull, soft globular hypomineralization, including: exencephaly, acrania
spine, changes
- platyspondily, including short spine
stature, growth, modified habitus
- growth delayed, failure to thrive, growth retardation
- stature, short, including micromelia, including short limbs
- stature, very short

UROLOGICAL DISORDERS
kidney, malformations
- nephrocalcinification (calcinosi)
OSTEOCHONDRDRODYSTROPHY, OSTEOCHONDRDRODYSPLASIAS
bone, mineralization disorders not including osteogenesis imperfecta

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

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<th>Differential diagnosis:</th>
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<td>Caffey disease</td>
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<tr>
<td>dyssegmental dwarfism, Rolland type</td>
<td>7970</td>
<td>Smith's Recognizable Patterns of Human Malformation. 6th Edition pag. 442-443</td>
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<td>osteogenesis imperfecta, lethal IIA</td>
<td>19590</td>
<td>Smith's Recognizable Patterns of Human Malformation. 6th Edition pag. 442-443</td>
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<td>osteoglophonic dwarfism</td>
<td>19620</td>
<td>Prenat.Diagn. 27(3),222-227,2007</td>
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<td>prenatal bowing</td>
<td>21558</td>
<td>Annuario Orfanet-Italia delle Malattie Rare 2005, pag. 457</td>
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<td>thanatophoric dwarfism</td>
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