mesomelic dwarfism, Langer type

**Eponyms:**
- GCFX
- homozygous Leri-Weill
dyschondrosteosis
- Langer mesomelic dwarfism
- mesomelic dwarfism-hypoplastic ulna/fibula/mandible type
- PHOG
- SHOX
- SS

**Inheritance:**
- autosomal dominant
- genetic heterogeneity
- pseudoautosomal
- supposed X-linked recessive

**Semeiological Synthesis:**
Facio-skeletal disorder. Severe mesomelic dysplasia, normal intelligence, curved/short radius and tibia, hypoplastic ulna and fibula, restricted elbow movements, hands/feet displaced laterally, mandible hypoplasia.

**Group**
- LABORATORY DATA
  - *chromosomal assignment*
  - chromosome X localization
  - chromosome Xp localization
  - chromosome Yp localization
  - *gene, structural-functional anomalies*
    - ANT3, adenine nucleotide translocator-3 (liver), SLC25A6 (solute carrier Family 25, member A6, gene chr.Xp22.32
    - ANT3Y, adenine nucleotide translocator-3, gene chr.Yp
    - GCFX (SS) (PHOG) (SHOX) growth control factor X-linked (pseudoautosomal homeo box osteogenic gene), gene chr.Xpter-p22.32
    - IL3RAY interleukin-3 receptor, gene chr.Yp13.3
    - XGR regulator XG/MIC2, gene chr.Xp22.32 and Yp11
  - *plasma electrolytes-inorganic constituents, modified functions*
    - sodium, high levels; hyponatremia

**OCULAR DISORDERS**
- eyelids, anomalies
  - blepharophymosis

**OROCRANIOFACIAL ANOMALIES**
- mandibular changes
  - micrognathia, mandibular hypoplasia, small jaw, not including: severe micrognathia, agnathia

**OTHERS**
- *inheritance*
  - inheritance, genetic heterogeneity
- *supergroups*
  - facio-skeletal disorders

**PRENATAL-NEONATAL MODIFIED DATA**
- *foetal changes*
  - birth length, decreased; low birth length
  - birth weight, low; foetal growth decreased, intrauterine growth retardation, intrauterine growth restriction, IUGR
  - foetal changes, recognized by ultrasound techniques
- *prenatal diagnosis*
  - prenatal diagnosis, echographic
### SKELETAL DISORDERS

carpus - tarsus, changes  
- madelung deformities  
- wrist, deformities  
elbow, anomalies  
- elbow, ankylosis  
fibular, defects  
- fibula, absent  
- fibula, short, hypoplastic  
hand-foot, changes  
- hand, clubbing  
limb anomalies, limb defects  
- bone, bowing, limbs bowed, camptomelia  
- mesomelic dwarfism  
radial changes  
- radius, bowed  
- radius, short, hypoplastic radius  
- ulna, short, hypoplastic  
spine, changes  
- hyperlordosis  
stature, growth, modified habitus  
- growth delayed, failure to thrive, growth retardation  
- stature, short, including micromelia, including short limbs  
tibia, anomalies  
- tibia, hypoplastic, short

### Super group:  facio-skeletal disorders

### Super aggreg.  FOETAL CHANGES

### Aggregations:  osteochondrodystrophy, osteochondrodysplasias

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Aggregation(s) [in differential diagnosis]:  
- mesomelic dwarfism

### Bibliography

OMIM ID: 249700  
OMIM ID: 312865  
Smith's Recognizable Patterns of Human Malformation. 6th Edition pag. 512-513  
OMIM ID: 430000  
OMIM ID: 403000  
OMIM ID: 300151  
OMIM ID: 489500  
OMIM ID: 400020  