### Knowles-Winter syndrome

**Eponyms:**
- dwarfism lethal short-limbed/platyspondylic type
- Luton lethal dysplasia
- thanatophoric variant type
- Torrance lethal dysplasia

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

**Semiological Synthesis:**
Skeletal disorder, isolated defect. Lethal short-limbed resembling thanatophoric dwarfism, straight long bones, widened cupped metaphyses, curved and short ribs, platyspondyly.

**Group**

<table>
<thead>
<tr>
<th>Sub group</th>
<th>Signs</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>OTHERS</strong></td>
<td>inheritance, genetic heterogeneity</td>
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</tbody>
</table>

**PRENATAL-NEONATAL MODIFIED DATA**

- **foetal changes**
  - birth length, decreased; low birth length
  - foetal changes, recognized by ultrasound techniques

- **prenatal diagnosis**
  - prenatal diagnosis, echographic

**SKELETAL DISORDERS**

- **limb anomalies, limb defects**
  - lethality, constant in osteochondrodysplasias
- **spine, changes**
  - platyspondily, including short spine
- **stature, growth, modified habitus**
  - stature, short, including micromelia, including short limbs
  - stature, very short

**Super group:**

**Super aggreg. Aggregations:**

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

- **LETHALITY**
  - lethality, constant in osteochondrodysplasias
  - osteochondrodysplasias and other severe skeletal defects

**Differential diagnosis:**

1 | achondrogenesis IA
2 | achondroplasia
3 | asphyxiating thoracic dystrophy
4 | camptomelic dysplasia, dominant type
5 | cloverleaf skull, Kozlowski type
6 | de la Chapelle syndrome
7 | dyssegmental dwarfism, Rolland type
8 | dyssegmental dwarfism. Silverman-Handmaker type
9 | fibrochondrogenesis
10 | Holmgren syndrome
11 | hydrops lethal-chondrodystrophy
12 | hydrops lethal-chondrodystrophy
13 | hypophosphatasia 1, congenital
14 | Jones-Jones-Miller syndrome
15 | metatropic dysplasia I
16 | micromelic dysplasia-cloverleaf skull
17 | osteogenesis imperfecta, lethal IIA
18 | Saldino-Noonan syndrome
19 | schleckenbecker skeletal dysplasia
20 | thanatoporic dwarfism
21 | thanatoporic dwarfism, Glasgow type
Genus database

25220  thanatophoric dwarfism, Horton type
25205  thanatophoric dwarfism-cloverleaf skull

Bibliography

OMIM ID: 151210