

Thanatophoric dwarfism

- **Thanatophoric dwarfism** (thanatophoric dysplasia) is the most common **lethal form of dwarfism**, it is caused by various mutations in the *FGFR3* gene .
- disorder of enchondral ossification, often associated with malformations of the cardiovascular system and central nervous system
- children have a large head, sometimes with craniofacial disproportion (the so-called *cloverleaf skull syndrome* - it has the shape of a trefoil), vertebral platyspondyl
- short limbs and ribs with marked narrowing and shortening of the chest (resulting in ARDS)
- femurs tend to be shaped like a telephone handset
- differential diagnosis: achondroplasia



radiogram of a baby born with thanatophoric dwarfism

Links

Related articles

- Achondroplasia
- Diastrophic dwarfism

References

- DUNGL, P., et al. *Ortopedie*. 1. edition. Praha : Grada Publishing, 2005. ISBN 80-247-0550-8.