Diastrophic dwarfism

Diastrophic dysplasia	
Clinical picture	Skeletal dysplasia with shortening of the trunk and limbs, final adult height 80–140 cm.
Cause	Mutations in the *SLC26A2 gene*
Diagnostics	Molecular genetic analysis of the *SLC26A2* gene (in the Czech Republic it is carried out by the ÚBLG FN Motol)
Incidence in the world	1–1.3:100,000
Classification and references	
ICD-10	Q77.5
OMIM	222600
orphaned	628

Lautrec-dwarfism

Diastrophic dysplasia (diastrophic dwarfism) is a generalized AR hereditary **disorder of enchondral ossification**, there is a reduced sulfation of the proteoglycans of the cartilaginous matrix - it inhibits the effect of FGF on cells. Atypical chodrocytes are formed and the cartilage is not mechanically resistant).

Clinical picture

Characterized by **dwarfism with short limbs and trunk**, height at maturity 80–140 cm. There is deformation of the limbs and spine (pedes equinovari, scoliosis, thoracolumbar and cervical kyphosis), involvement of the auricles (flower-shaped deformity) and trachea. Diastrophic dwarfism is characterized by a so-called hitchhiker's **thumb** (short triangular first metacarpal and radial subluxation of the metacarpophalangeal joint). We often see bilateral teratological hip dislocation (or dislocation of the patella, head of the radius). Joints are excessively loose or (more often) stiff (multiple contractures at birth). Another sign is a "cherubic" face - fullness around the mouth, nostrils and middle nosewide).

Therapy

- symptomatic
- correction of deformities very demanding

Links

related articles

- Achondroplasia
- Thanatophoric dwarfism
- Larsen syndrome
- Congenital limb defects
- Developmental hip dysplasia
- Pes equinovarus congenitus

References

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