

Dysostosis cleidocranialis

Dysostosis (dysplasia) **cleidocranialis** is an AD hereditary **disorder of desmogenous** (gum, calf, pelvis) and **enchondral** (arm, leg, spine) ossification. For example, actor Gaten Matarazzo (who played Dustin Henderson in Stranger Things) suffers from the disease.

Pathogenesis

- Mutation of the *RUNX2* gene (runt related transcription factor 2, gene naming recommended by HGNC; former gene name *CBFA1*) → abnormal bone tissue produced.^[1]

Clinical picture

It usually manifests itself in the first 2 years, **typical symptoms are**:

- broad head, relatively small face with hypertelorism, vertical shoulders, narrow chest, dentition disorder, persistent fontanelles,
- aplasia / hypoplasia of the clavicle,
- short stature
- hypermobility of the arm girdle,
- neuromuscular disorders from brachial plexus damage and agenesis of some chest muscles,
- scoliosis, hollow chest,
- brachycephaly,
- poorly developed dentition (delayed eruption and replacement of teeth).

X-ray image

- **Defective clavicles**(in the lateral and central part),
- **delayed closure of fontanelles** and cranial sutures (as opposed to craniostenosis),
- possibly deformities of the chest, pelvis, scoliosis, spina bifida occulta.

Therapy

- Reconstruction of the clavicles is not recommended, sometimes decompression of the brachial plexus,
- coxa vara: valgus intertrochanteric osteotomy,
- strengthening of the muscles of the shoulder girdle: exercise, bandage,
- treatment of scoliosis.

Links

Related articles

- Achondroplasia
- Thanatophoric dwarfism
- Diastrophic dysplasia (diastrophic dwarfism)
- Spondyloepiphyseal dysplasia
- Apert syndrome (acrocephalosyndactyly)
- Osteogenesis imperfecta (osteopsathyrosis, ossium fragility)
- Arthrogryposis multiplex congenita

Reference

1. MENDOZA-LONDONO, Roberto and Brendan LEE. *Cleidocranial Dysplasia* [online]. [feeling. 2017-05-29]. <<https://www.ncbi.nlm.nih.gov/books/NBK1513/>>.
2. WANG, Jiangping, Xinwen HUANG, and Can LAI, et al. Clinical spectrum of cleidocranial dysplasia in a family with twins. *Pediatr Int* [online] . 2013, vol. 55, no. 3, pp. 392-5, also available from <<https://www.ncbi.nlm.nih.gov/pubmed/23782374>>. ISSN 1442-200X.

Sources

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1. MENDOZA-LONDONO, Roberto – LEE, Brendan. *Cleidocranial Dysplasia* [online]. [cit. 2017-05-29]. <<https://www.ncbi.nlm.nih.gov/books/NBK1513/>>.

