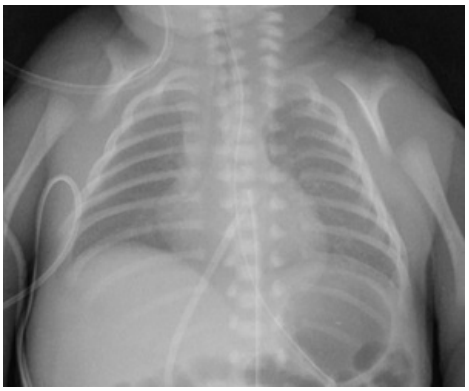
Cleidocranial dysplasia

Cleidocranial **dysplasia** (CCD) is a very rare autosomal dominantly inherited disease that affects the development of bones, skull and teeth. A typical sign is partial or full **aplasia of the clavicles**, **tooth anomalies**, **delayed closing of the fontanelles**, as well as **osteopenia , hearing disorders, hand bone abnormalities, recurrent otitis and sinusitis**. People with CCD may develop scoliosis, osteoporosis, and short stature.

The disorder is most often caused by the presence of mutations in the RUNX2 gene (*Runt-related transcription factor 2*), whose protein product is a key transcription factor affecting osteoblast differentiation and bone morphogenesis.

There is no causal treatment, the care is aimed at alleviating difficulties and depending on the severity of the symptoms, a craniofacial surgeon, dentist, orthodontist, endocrinologist, otorhinolaryngologist, speech therapist and orthopedist are involved.



Premature neonate with clavicle agenesis in cleidocranial dysplasia

Links

Related Articles

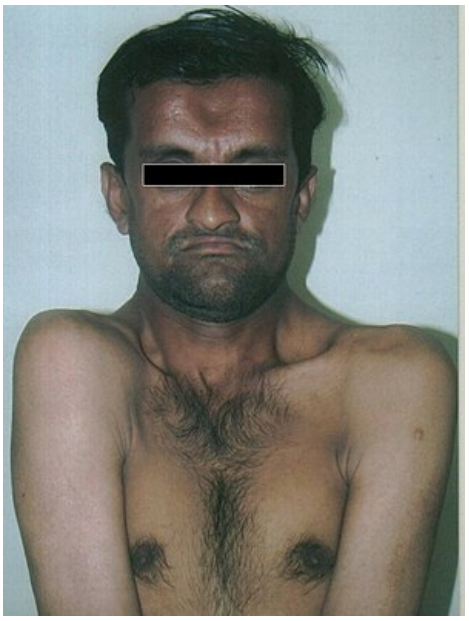
- Osteogenesis imperfecta

External links

References

Genetic and Rare Diseases Information Center. *Cleidocranial dysplasia [online]* [online]. The last revision 19-08-2020, [cit. 16-03-2023]. <https://rarediseases.info.nih.gov/diseases/6118/cleidocranial-dysplasia>.

National Organization for Rare Disorders. *Cleidocranial Dysplasia [online]* [online]. [cit. 16-03-2023]. <https://rarediseases.org/rare-diseases/cleidocranial-dysplasia/>.



Cleidocranial dysplasia - obvious abnormalities of the skull and increased mobility of the shoulder girdle.