Cleidocranial dysplasia

Patient Information Series – What you should know, what you should ask.

What is cleidocranial dysplasia?

Cleidocranial dysplasia or dysostosis is a birth defect that primarily affects the development of the bones and teeth. The main features of the condition are the hypoplastic (very small) or absent clavicles and the delayed maturation of the skull (cranium). As a result, the shoulders are narrow and sloping, can be brought unusually close together in front of the body, and in some cases can be made to meet in the middle of the body. The cranial structural abnormalities are also characteristic of this condition and include delayed closing of skull sutures and larger than normal fontanelles.

How does cleidocranial dysplasia happen?

Cleidocranial dysplasia is mainly caused by mutations in the RUNX2 gene. The shortage of functional RUNX2 protein interferes with the normal development of cartilage, ossification of bones and teeth formation, resulting in the signs and symptoms of cleidocranial dysplasia.
In about 30% of cleidocranial dysplasia cases, no RUNX2 gene mutation is found. The cause of the condition in these cases remains unknown.

How is it diagnosed?

Diagnosis is usually suspected at the routine anomaly scan (around 20 weeks) although it can be suspected from 14 weeks. The most consistent feature is the abnormal clavicles, which are either or partially or totally absent. Other less specific findings include: frontal bossing abnormal skull shape or bone mineralization. In cases of family history prenatal testing is feasible

Should I have any more tests?

Evaluation should be carried out by an expert in Fetal Medicine. The diagnosis of cleidocranial dysplasia is established through its typical clinical and radiographic findings and by the identification of a heterozygous pathogenic variant in RUNX2. Therefore, genetic testing can be offered. Additional sonographic (by ultrasound) evaluation should include a detailed exam of all other organs.

Are there any associated anomalies?

Affected individuals are often shorter than other members of their family at the same age. They may also have short, tapered fingers and broad thumbs, flat feet, bowed legs or knock knees, short scapulae and scoliosis. They may have decreased bone density and develop osteoporosis. Women with cleidocranial dysplasia have an increased risk of requiring a cesarean section when delivering a baby, due to a narrow pelvis preventing passage of the infant's head. Dental abnormalities are very common. In addition to skeletal and dental abnormalities, cleidocranial dysplasia may manifest with hearing loss and susceptibility to sinus and ear infections. Infants with this condition may be mildly delayed in the development of motor skills, such as crawling and walking, but intelligence is unaffected.

What is the prognosis? What treatment will my child require?

Several studies have reported that life expectancy appears to be normal for people with cleidocranial dysplasia.
After birth, if the cranial defect is significant, the head needs protection from blunt trauma; helmets may be used for high-risk activities. Cosmetic surgery for the forehead and clavicles may be considered. If bone density is below normal, treatment with calcium and vitamin D supplementation should be considered. Preventive treatment for osteoporosis should be initiated at a young age. Dental procedures are common and speech therapy may be required during periods of dental treatment. Aggressive treatment of sinus and middle ear infections is mandatory. Finally, monitoring of affected women during their pregnancy will determine the mode of delivery.

Will it happen again?

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. Some affected individuals inherit the mutation from one affected parent. Often the parent is mildly affected, and in some cases had not previously been recognized as having the disorder. Other cases result from new mutations in the gene. These cases occur in people with no history of the disorder in their family. Thus, the recurrence risk depends on the carrier status of the parents. In case of an affected parent the recurrence risk is 50%. In de novo cases the recurrence risk is very low.

What other questions should I ask?

- Should I have any more tests?
- Are there any additional anomalies present?
- Have other skeletal anomalies been ruled out?

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