

# FACES:

The National Craniofacial Association 1-800-332-2373

## What is Cleidocranial Dysplasia?

Cleidocranial Dysplasia [ *cleido* , collar bone, + *cranial*, head, + *dysplasia*, abnormal forming ] also known as CCD, Cleidocranial Dysostosis and Marie-Sainton Disease, is a condition characterized by defective development of the cranial bones and by the complete or partial absence of the collarbones (clavicles). Characteristics include:

- Delayed closure (ossification) of the space between the bones of the skull (fontanelles)
- Premature closing of the coronal suture
- Protruding jaw (mandible) and protruding brow bone (frontal bossing)
- Wide nasal bridge due to increased space between the eyes (hypertelorism)
- High arched palate or possible cleft palate
- Short stature
- Scoliosis of the spine

## Why did this happen?

There is no link between anything the mother did or did not do while she was pregnant and the occurrence of Cleidocranial Dysplasia. It is transmitted as an autosomal dominant trait. CCD is caused by mutation in the RunX2 gene on Chromosome 6p21.

## Will this happen to children I have in the future?

If both parents are unaffected, it is called a spontaneous genetic mutation and the chances of having another child with it are very small. If one parent is affected, it is called an inherited genetic mutation and there is a 50% chance that a child will have it. Of all Cleidocranial Dysplasia cases, 1/3 are spontaneous and 2/3 are genetic.

## What kinds of problems could my child have?

In addition to the physical characteristics common to Cleidocranial Dysplasia, your child may have the following problems:

- Dental abnormalities - failure to lose the baby teeth (deciduous) at the expected time; slow eruption of secondary teeth; extra teeth; peg teeth; delayed or absent formation of teeth.
- Ability to touch shoulders together in front of the body.
- Wide pelvic bone
- Loose joints
- Hearing loss and/or frequent infections

## Will my child need surgery?

Due to the fact that the dental problems are the most significant complications, appropriate dental/orthodontic work is vital. Some of the suggested treatment options include the following:

- Apply dentures over the unerupted teeth.
- Teeth removal as they erupt, because very little bone structure would be left if the supernumerary, impacted, and unerupted teeth were all extracted at once.
- Some doctors suggest that the removal of primary or supernumerary teeth does not promote eruption of unerupted permanent teeth. In addition, permanent teeth may be difficult to extract due to malformed roots.

**New advances in procedures to correct Cleidocranial Dysplasia are constantly being developed. Be an advocate for your child!**

### **How do I get help for my child?**

Your child should be treated by a qualified craniofacial medical team at a craniofacial center. Currently, FACES has information on many of these teams. This is by no means a comprehensive list of all the craniofacial teams. Please contact FACES for details.

### **Am I alone?**

No! There are many families and organizations who will be glad to talk with you and help you with information and support. Don't forget books, videos, and websites. The listing below will get you started.

FACES: The National Craniofacial Association  
P.O. Box 11082  
Chattanooga, TN 37401  
(800) 332-2373  
E-mail: [faces@faces-cranio.org](mailto:faces@faces-cranio.org)  
Website: [www.faces-cranio.org](http://www.faces-cranio.org)

*We provide financial support for nonmedical expenses to patients traveling to a craniofacial center for treatment. Eligibility is based on financial and medical need. Resources include newsletters, information about craniofacial conditions, and networking opportunities.*

Office of Rare Diseases  
The Genetic and Rare Diseases Information Center  
P.O. Box 8126  
Gaithersburg, MD 20898-8126  
(888) 205-2311 (toll free)  
Fax: (202) 966-5689  
Email: [gardinfo@nih.gov](mailto:gardinfo@nih.gov)  
website: <http://rarediseases.info.nih.gov/>

*Excellent web site for finding information on a variety of craniofacial disorders, particularly very rare ones. Information specialists are available to answer your questions. The center does not give medical advice, provide treatment, or diagnose illness.*

National Health Law Program  
1101 14<sup>th</sup> Street NW, Suite # 405  
Washington, DC 20005  
(202) 289-7661  
website: [www.healthlaw.org](http://www.healthlaw.org)

*Provides extensive information on health care laws affecting families of children with special needs.*

### **Children with Facial Difference: A Parent's Guide.**

Written by Hope Charkins, MSW. Published by Amazon, 1996.

website: Website: [www.Amazon.com](http://www.Amazon.com)

*Excellent resource for parents to help them cope with medical, emotional, social, educational, legal, and financial challenges present by facial differences of their children.*

U. S. National Library of Medicine  
(888) 346-3656 (toll free)

[www.nlm.nih.gov/nlmhome.html](http://www.nlm.nih.gov/nlmhome.html)

*Has a descriptive article on Cleidocranial Dysplasia. Enter the words "cleidocranial dysplasia" in the search box.*

Let's Face It:

<http://media.dent.umich.edu>

*This site has some pictures illustrating some aspects of cleidocranial dysplasia.*