A Parent’s Guide
to the
Cardio-Facio-Cutaneous Syndrome

Caring, Facilitating & Connecting
Forging a path to improve lives through family support, research and education.

Luntsford Family, Montana

Nathaniel and Mom

Kaci
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Introduction

Most likely you have never met someone who has the Cardio-Facio-Cutaneous (CFC) syndrome. You are told that this is the reason your child has medical problems and/or physical differences. On the one hand, you may feel relief that your search for a diagnosis is over. On the other hand, you may feel overwhelmed by this news. It can be difficult to understand the information you are being given about the CFC syndrome. This booklet is yours to be read at your own pace. As your child grows and develops, you’ll probably refer to it often. In time, you will have the information you need to be the best possible parent to your child. Your child is fortunate that you care enough to have brought him or her to the doctor. Your interest in helping your child is obvious by your initiative to learn more and read this booklet.

Establishing a diagnosis can help parents, teachers, and doctors provide the best possible care for your child. Knowing a diagnosis can help you to anticipate possible future medical difficulties and developmental hurdles. In addition to a pediatrician, your child may need to see medical specialists such as: geneticists, cardiologists, neurologists, ophthalmologists, GI doctors, and Ear Nose Throat specialists. Therapies may include speech, physical, and occupational. Many children have a different lifestyle due to frequent visits to doctors and therapists. Early Intervention services can help children learn new skills. Many parents have found that most new skills are obtained through constant reinforcement. Some parents report very few developmental delays while others express concerns over constant feeding problems and language impairments. Receptive language skills are consistently higher than expressive skills. Although the children share a common thread of Cardio-Facio-Cutaneous syndrome, they all vary in their development and medical conditions.

In the following pages you will find information that will answer many of your questions concerning CFC syndrome. CFC International hopes this booklet helps you adjust to the diagnosis and dispel some of your fears.
What is CFC syndrome?

What is a syndrome? The word syndrome is used to describe a cluster of manifestations seen together and thought to have one underlying cause. Down syndrome is an example in which Dr. Down noticed that many people with mental retardation had a similar appearance. Syndromes are caused either by mutations in our genes, environmental factors, or a combination of the two.

What is Cardio-Facio-Cutaneous syndrome? This is a description of the physical findings in children with this syndrome. “Cardio” refers to the heart, “facio” refers to the face, and “cutaneous” refers to the skin.

Why hasn’t anyone heard of it? Dr. Jim Reynolds and his colleagues first described the CFC syndrome in 1986. It is a very rare condition with over 100 cases reported in the medical literature. It is not known how frequently a child is born with the CFC syndrome, but it is estimated that there are 200-300 individuals worldwide.

CFC and Noonan syndrome. In years past, the medical journals often referred to CFC as a more severe form of Noonan syndrome. However, now researchers have discovered that the genes that cause Noonan syndrome are different than the genes that cause CFC syndrome, although the two conditions still share many similarities with one another. Regardless, your child will benefit from the anticipatory guidance that is possible from the diagnosis, and you may benefit by meeting other parents of children with the CFC syndrome through the international support group.

Wallace Family, Texas
Findings in CFC syndrome
(Adapted from Online Mendelian Inheritance in Man)

The head:
- Macrocephaly (relatively large head circumference)
- High forehead
- Bitemporal constriction (narrowed temples)
- Hypoplastic supraorbital ridges (some brow ridges which are underdeveloped)
- Sparse brittle hair

The face:
- Nystagmus (unsteadiness of the eyes)
- Downward slanting eyelid openings
- Sparse eyebrows
- Depressed nasal bridge
- Posterior angulated ears (ears tilted backward)
- Prominent ear helices (the outer rim of the ears appears large and fleshy)
- Ptosis (appears to have “droopy” eyes)

The chest:
- Pectus carinatum/excavatum (protrusion or indentation of the breastbone)

The hands:
- Predominant finger tip pads
- Thin, fast-growing, opal-colored nails

The skin:
- Generalized over-pigmentation
- Generalized ichthyosis-like dermatosis (scaly skin)
- Patchy hyperkeratosis (patches of thickened skin)
- Keratosis plantaris (thick skin on the sole of the foot)
- Keratosis pilaris (hair follicle prominence of the eyebrows)

The heart:
- Pulmonic stenosis (narrowing of the valve in the pulmonary arteries)
- Atrial septal defect (abnormal opening between the left and right upper chambers of the heart—the tubes leading to the lungs)
- Hypertrophic cardiomyopathy (enlarged heart, thick heart muscle)
What Causes CFC Syndrome?

CFC syndrome is caused by a mutation (change) in one of our genes. Genes are the bits of hereditary instructions that determine how we look and how our bodies develop and work. Everyone has about 25,000 pairs of genes. Our genes are located on structures called chromosomes that are found in every cell of our body. Chromosomes are inherited from our parents through the egg and sperm (one copy from our mother and one from our father). We look a little like both sides of our family because we inherit a copy of every gene from each parent. During conception, the egg and sperm fuse to form one cell with 23 pairs (total = 46) of chromosomes. The fertilized egg then duplicates over and over to form a baby.

Among the 25,000 genes in our bodies, there are at least four known genes that cause the CFC syndrome. Individuals with CFC syndrome have one normal gene and one altered gene. The four genes found to be associated with CFC syndrome are called BRAF, MEK1, MEK2, and KRAS. Most individuals with CFC syndrome (75-80%) have a mutation in BRAF, with 10-15% having a mutation in MEK1 or MEK2 and <5% with a mutation in KRAS. Mutations in KRAS have also been identified in a few individuals with Noonan syndrome, therefore this may make it a little more difficult to interpret the results if an individual has a KRAS mutation. At this time we are still learning about the specific mutations in these genes and how they affect our children; however it is often difficult to predict the extent and severity of the condition in any given child.
Is Prenatal Testing Available?

If you are considering having more children, you may be concerned that you will have another child with the CFC syndrome. You should remember that the risk for unaffected parents of a CFC child to have another child similarly affected is very low. Or, perhaps your sister or brother is planning a family and is concerned. The risk your brother or sister may have an affected child is no greater than in the general population. Prenatal diagnosis is possible if the mutation in the individual with CFC syndrome is known. There are two different procedures that can be performed at different gestational ages. Chorionic Villus sampling (CVS) is a procedure that is performed between 10-14 weeks gestation and involves sampling some of the cells from the placenta and performing genetic testing on those cells. The risk of miscarriage from this procedure is thought to be <1%. The second procedure, called an amniocentesis, is performed between 16-20 weeks gestation and involves sampling cells found in the amniotic fluid. The risk of miscarriage related to amniocentesis is very low. Before making any decisions regarding prenatal diagnosis it is important to discuss the risks, benefits, and limitations with your healthcare provider or genetics professional. Some reassurance may be gained by a detailed ultrasound examination. This is a non-invasive procedure using sound waves to produce an image of the baby. Findings such as excess amniotic fluid or a heart defect would be a clue to your doctors that this baby may have the CFC syndrome. However, it is very important to remember that some babies without the CFC syndrome also have these “nonspecific” findings. As a result, the CFC syndrome may be suspected, but not diagnosed, during pregnancy. Conversely, some mildly affected CFC babies may not show conspicuous changes on prenatal ultrasonography.

How Is Someone Diagnosed With CFC?

In the past, the diagnosis of CFC was dependent on the observations of a clinician. This means that a doctor recognizes several signs of the CFC syndrome occurring together in an infant, child, or adult. Although a diagnosis may be possible in a newborn infant, most often the diagnostic changes do not become fully evident until early childhood. With the discovery of genes associated with CFC syndrome, the clinical diagnosis is now able to be confirmed in those suspected of having CFC. Genetic testing of all four genes is clinically available in laboratories around the world.
Is There a Cure For CFC Syndrome?

There is no way to cure the CFC syndrome. The genetic change responsible for CFC is in every cell of the body. At this time, there is no way to access every cell in the body to fix the gene mutation.

However, it is possible to treat many of the medical problems associated with CFC syndrome. Treatment should be based on the child’s needs, rather than the diagnosis. For example, a child with a heart defect should be seen by a pediatric cardiologist and treated by that doctor as he or she would treat any child with the same heart defect.

Will Growth Hormone Therapy Improve My Child’s Height?

People with the CFC syndrome are generally shorter than average because they are genetically programmed to be small.

A small number of these children have been found to be growth hormone deficient. Their parents decided to try growth hormone therapy. Their endocrinologist (a doctor who specializes in the function of hormones) suggested the children might benefit from treatment. At the present time, there is no data to document their success or to determine if a larger majority of the CFC children might also be growth hormone deficient. At best, these children may
achieve a more normal adult height. If not, these children will have a growth spurt on treatment, but their final adult height probably will be the same as untreated individuals with the CFC syndrome. As this population of children ages more will be learned.

If you are interested in using growth hormone therapy, your child’s pediatrician can refer you to a pediatric endocrinologist to evaluate whether your child has any evidence of growth hormone deficiency. If so, treatment is indicated. If not, opinions vary on whether to treat or not. Growth hormone is, in any event, not a standard treatment of the CFC syndrome.

A delay in puberty may also be seen with CFC syndrome. Adolescents often go through puberty later than their peers. Once a person with CFC syndrome has matured into an adult, they may be able to have children. Data have not been obtained on a second generation of CFC syndrome individuals at this date.

Will My Child Attend Regular School?

Individuals with CFC syndrome may have normal, near-normal, or sub-normal cognitive abilities (IQ) and attend a regular school. Those with learning disabilities or cognitive disabilities may require extra help in some subjects. Sometimes children attend a resource room or have a classroom aide assigned to them in a regular classroom. Educational teams often recommend therapies to help the children learn new skills. The therapy recommendations often depend on the amount of delay that the child is displaying.

Development

Doctors and teachers have timeline references for when a child should reach developmental milestones such as walking or talking. Frequently, children with CFC syndrome attain these milestones a bit later than usual. This is referred to as developmental delay. Sometimes children continue to be delayed and will be cognitively disabled. Other times, the children catch up and perform equally with their peers.
Learning Disabilities and Cognitive Disability

Children with CFC syndrome have varying degrees of cognitive disability. Accessing the actual intelligence level can often be difficult since visual and language impairments can interfere with obtaining accurate results.

Delays in verbal expression is the most common learning disability in the CFC syndrome. Many parents have reported that their child can understand verbal directions much better than they can express themselves. This suggests that receptive language skills (the comprehension of language) are noted to be better than expressive language and speech production skills (the ability to communicate clearly). It seems that many of these children are learning and storing language skills but are unable to indicate this knowledge until their expressive abilities improve. It is recommended that you stimulate your child’s receptive and expressive language skills as much as possible. A speech language pathologist can give you specific suggestions to facilitate this development.

In addition, many children have delays in oral motor skills, meaning they have difficulty with range, coordination, and sequencing of the movements within the muscles and structures used for speech. These difficulties can interfere with feeding and swallowing and with intelligible speech production.

Sign language, communication boards and augmentative communication devices are often used with the younger children to reduce frustration. The children often can understand the language but without being able to express their own needs, they become upset. A speech pathologist with a background in communication devices can assist families with this transition phase.

Children with learning disabilities and cognitive disability (often diagnosed in infancy as developmental delay), usually benefit from early intervention programs. These programs are designed to provide services to children at risk for developmental delays. Often, early intervention programs involve the help of professionals such as physical therapists, occupational therapists and speech language pathologists. By beginning these programs in infancy, children are able to reach their developmental milestones sooner and able to reach their ultimate potential.
Hearing, the Palate and Speech

Mild hearing loss has been documented in individuals who have CFC syndrome. The hearing loss may be attributed to recurrent ear infections as well as heavy wax build up in the noted small (stenotic) ear canals. Recurrent ear infections and earwax build up may result in speech delay. Infants and children should have thorough hearing evaluations. Medical management can sometimes minimize the speech delays that can result from the wax build up and the recurrent ear infections. Many children visit their ENT quite often to have the earwax removed, their hearing monitored and medical management of the ear infections. Often the identification of a hearing problem can help explain some speech delay.

The palate (roof of the mouth) is often abnormally formed. The shape of the palate may be high and narrow or even short or it may have a defect called sub-mucous cleft. This is where the tissues or bones in the midline of the palate have not closed, although the mucous membrane covering it is intact. These are sometimes difficult to detect and can contribute to speech and feeding difficulties. If your child is having difficulty with oral motor skills, feeding or swallowing you should find a speech language pathologist with particular expertise in these areas. Not all speech language pathologists have this background with oral motor therapy. To find qualified therapists in your area, you can contact the American Speech Language and Hearing Association.
Central Nervous System

At birth children with CFC syndrome often have large heads. CT scans or brain MRIs may show a lack of substance (particularly the cortex, which is known as the surface of the brain) and the cavities inside the brain are larger than usual. Thinning of the corpus callosum is also seen in CFC syndrome. All of these findings are non-specific findings.

The major conclusion from the neuroradiological findings and images is that there is virtually no correlation between structure and function of the individual. The tests do not predict whether the child is going to have seizures or not. They do not predict the cognitive outcome and they do not predict the motor outcome.

Nearly 45% of children experience seizures. Most seizures begin in infancy or early childhood; however they may develop later in childhood as well. They are often controlled with medications. Working with a pediatric neurologist is important.

Hypotonia (reduced muscle tone) is a common trait of CFC syndrome. If the hypotonia is accompanied by normal deep tendon reflexes this is a good prognostic sign. The control of muscle tone is age dependent. Children who start off hypotonic and who have decent deep tendon reflexes almost invariably improve. Because they can’t do the motor things they’re expected to do at given ages, the child may mistakenly be identified as mentally retarded. In fact, motor development is not strongly correlated with cognitive development at all!

The Eye/Vision

The eyes are an outgrowth of the brain. There are quite a few indications from the literature and from families reporting that the eye nerve is underdeveloped.
Nearly all individuals with CFC syndrome have some associated eye abnormalities. The most common findings are:

- Nystagmus (involuntary eye movements)
- Hypertelorism (widely spaced eyes)
- Ptosis (droopy eyelids)
- Strabismus (muscle imbalance)
- Optic Atrophy (under development of the optic nerve)
- Epicanthal folds (folded skin in corner of eye)
- Downslanting eyes
- Amblyopia (reduced vision)

Many of these contribute to the characteristic facial appearance seen in CFC syndrome. A majority of the individuals with CFC syndrome will need prescription glasses. Although ptosis and strabismus are not usually severe, in many cases surgery may be required.

Because of the high prevalence of eye problems, it is suggested that children with CFC syndrome be seen by a pediatric ophthalmologist. It is especially important to schedule a detailed eye exam in early childhood to assess the level of eye involvement in your child. Some of the CFC children have qualified for services from the teachers of the visually impaired. Check with your ophthalmologist to obtain his/her opinion on this service.

**Heart Defects**

A large majority of the babies born with CFC syndrome have a heart defect. These can be characterized as congenital (present at birth) or may develop later. Although many different types of heart defects have been observed, the most common are pulmonic stenosis, hypertrophic cardiomyopathy, and septal defect (ASD).

The heart is a muscle that pumps deoxygenated blood to the lungs and oxygenated blood out to the body. Blood that has given up its oxygen to the brain, organs, and muscles returns to the heart via the veins. The blood enters the heart through the right atrium and then it flows to the right ventricle and is pumped out through the pulmonary artery to the lungs. Once oxygenated, the blood returns to the heart via the pulmonary veins and enters the left atrium. Blood then flows from the left atrium to the left ventricle which pumps the oxygenated blood out to the body through the arteries.
Within the heart there are several valves that help to direct the flow of blood. A valve opens to let blood pass through and then closes to prevent the blood from flowing backwards. The pulmonary valve is located between the right ventricle and the pulmonary artery. Pulmonic valve stenosis is common in CFC syndrome. The valve is often small and underdeveloped (dysplastic), making it harder for blood to flow from the right ventricle to the lungs. Often, this is not severe and children do not require surgery.

However, in a small majority of cases, surgery is needed to help restore proper blood flow. Surgeons can repair the valves, replace them with artificial valves, or increase their size by using a “balloon”. In milder cases, balloon valvuloplasty is the preferred intervention for pulmonic stenosis. A balloon tipped catheter is inserted into the valve. The balloon is inflated and the valve is widened, allowing more blood to pass through the valve.

The second most common heart defect associated with CFC syndrome is hypertrophic cardiomyopathy. In this condition the heart muscle is thickened and its function is impaired. Thickening of the heart walls leads to a decrease in the amount of blood able to be pumped out to the body. The degree of severity of hypertrophic cardiomyopathy is variable. However, most people with CFC syndrome remain asymptomatic for many years.

Several other structural heart differences have been found in CFC Syndrome. These are less common than pulmonic stenosis and hypertrophic cardiomyopathy.
Skin Manifestations

A variety of skin manifestations are present in the CFC syndrome. Not one feature is present in all cases, and there are a few individuals who have no skin abnormalities. A set of the most frequent manifestations observed in individuals who have CFC syndrome can be found below.

The hair is usually sparse, curly, and thin, has a lower posterior border, is fair, dry, brittle, and grows slowly. Eyelashes and eyebrows are sparse, sometimes even absent. Slow growth has also been reported in nails, which are normal in most cases, but nail dystrophy has been reported in 15% of CFC individuals.

Dry skin, keratosis pilaris (hair follicle prominence) and hyperkeratosis (thickened skin) are the most frequent skin features in CFC syndrome. The dryness of the skin is sometimes a predisposing fact for allergic dermatitis (eczema), which can also suffer bacterial infection, so the use of powerful moisturizers is highly recommended. Keratosis pilaris varies in intensity from case to case. Some individuals have such a pronounced production of keratin in the hair follicle that the follicle itself becomes shut down, with no hair production. Keratosis pilaris can happen anywhere in the body (except over palms and soles, where there are no hair follicles), but is more frequently seen on the face (cheeks and eyebrows), arms and legs. Hyperkeratosis is also noted in different patterns. Most individuals have thickened skin on elbows and knees, others have thickened skin over the entire body, and very few have just palms and soles thickened.

Generalized hyperpigmentation is described in some individuals, who have a darker skin color when compared to the rest of the family. But this is just a sign, leading to no complication or consequence. Hyperpigmented spots and café-au-lait spots (brown spots) are also reported, again with no complications related.

Blood vessel skin lesions such as hemangiomas and cutis marmorata (purple marks in a network pattern, over legs and arms) are also present in some individuals. There are no reports of hemangiomas needing treatment; they are more of the flat kind, leaving just a red to purple spot on the skin. Cutis Marmorata gets worse when it is cold, so it is recommended to avoid exposure of the extremities, especially in winter. Again, no complications related to vascular skin manifestations are reported.
Feeding and Nutrition

Severe feeding difficulties are common in the infant with CFC syndrome which may present as “failure to thrive”. This may be related to poor sucking and/or swallowing coordination or difficulty with weight gain. Infants, and occasionally older children, may require some assistance with the use of an NG (nasogastric) tube or G-tube (gastrostomy). Older children with CFC syndrome may also experience oral aversion to certain textures. This can often make feeding a difficult challenge for parents. Despite adequate nutrition and caloric intake, most individuals with CFC will be lower on the growth curves compared to siblings.
Management
(taken from Gene Reviews)

The following evaluations are recommended at the time of diagnosis and as needed during subsequent doctors visits:

1. Genetics consultation and genetic testing
2. Complete physical exam including growth parameters
3. Cardiac evaluation including echocardiogram and electrocardiogram (EKG)
4. Neurologic evaluation
5. MRI of brain to detect any structural changes
6. EEG (electroencephalogram) if seizure suspected
7. Abdominal ultrasound to evaluate for renal anomalies
8. Psychomotor development evaluation
9. Endocrine evaluation if growth delay is suspected
10. Ophthalmologic evaluation
11. Audiologic examination
12. Nutrition and feeding evaluation
13. Dermatologic evaluation
14. Referral to Early Intervention Services
Summary of Recommendations and Management in CFC syndrome

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Summary of Recommendations and Management in CFC syndrome (Cont.)

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Conclusion

Hopefully, this booklet has answered many of your questions about Cardio-Facio-Cutaneous syndrome. It is important for you to remember that you cannot control the genes you pass on to your children. Thus, there is nothing you can do prior or during a pregnancy that can cause or prevent the occurrence of CFC syndrome.

The diagnosis of a genetic syndrome is often difficult to accept. There is no right or wrong reaction. Feelings of sadness, anger, confusion, guilt, helplessness and fear may surface at once or in stages. Parents, siblings and/or extended family members may react toward the CFC child with rejection, embarrassment, over indulgence or in other ways.

Many parents find comfort and hope in talking to other families touched by CFC syndrome. The international support group is available to provide you with more resources and information regarding CFC syndrome.

This brochure is intended to provide basic information about Cardio-Facio-Cutaneous syndrome. It is not intended to, nor does it constitute medical or other advice. Readers are warned not to take any action with regard to medical treatment or otherwise based on the information in this brochure without first consulting a physician. CFC International, Inc. does not promote or recommend any treatment, therapy, institution or health care plan. The information contained in this brochure is intended to be for your general education and information only and not for use in pursuing treatment or course of action. Ultimately, the course of action in treating a given individual must be individualized after a thorough discussion with the individual’s physician(s).
References


Online Mendelian Inheritance in Man (OMIM) is a database of human genes and genetic disorders written and maintained by medical professionals at Johns Hopkins and elsewhere, and developed for the Internet by the National Center for Biotechnology Information (NCBI). The information on this site is geared toward medical professionals.


Acknowledgements

This booklet was modeled after “Understanding Noonan Syndrome: A Parent’s Guide” by Maura Kenton and Eric A. Wulfsberg. We wish to thank The Noonan Syndrome Support Group for sharing their valuable resources with us!

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Published 2002
Revised 2005
Revised 2007
Conger Family,
New York

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Vestal, NY 13850
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