








How You can Help

A contribution to CFC International will help our efforts with family outreach, research, treatment and education. All donations are tax deductible to the full extent permitted by law. Your help can make a difference in the life of a child born with Cardio-Facio-Cutaneous syndrome.



CFC International
Cardio-Facio-Cutaneous Syndrome

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Caring, Facilitating & Connecting

About Our Organization

CFC International is a 501(c)(3) non-profit organization founded in 1999 by parents of children with Cardio-Facio-Cutaneous syndrome. We are the leading international organization dedicated to providing support for persons and families dealing with CFC syndrome. We serve as a clearinghouse of information on all aspects of CFC syndrome. We are committed to supporting research on CFC syndrome. We seek treatments and keep reaching for a cure, offering hope to those individuals and their families affected by this condition. We are in frequent communication with researchers and update our members with any new developments through the private family listserv, CFC International Facebook site, and also the organization website. Please stay connected to us.

Each year we find strength and courage in each other, knowing that we are a growing genetic family whose connection lies at the very core of human existence. If you are interested in further information please contact us.

We are here to help!



CFC International
Cardio-Facio-Cutaneous Syndrome

*Improving the
quality of life through
family support, research,
and education.*



What is Cardio-Facio-Cutaneous (CFC) Syndrome?

Cardio-Facio-Cutaneous syndrome is a rare genetic condition that affects approximately 1 in 810,000 people. It causes a range of physical, cognitive, and developmental disabilities that can range from mild to severe. It affects both genders equally and is seen in all races/ethnic backgrounds.

Common features found in CFC syndrome

- A distinctive facial appearance
- Unusually sparse, brittle, curly hair (not seen in all cases)
- Skin abnormalities
- Heart defects that may be present at birth or develop later
- Growth delays
- Varying degrees of intellectual disability
- Vision abnormalities
- Seizures

Individuals with CFC typically have distinctive facial features with a larger head size (macrocephaly). In addition to having unusually dry, brittle, curly scalp hair, affected individuals also often lack eyebrows and eyelashes. Skin problems, such as dry, red, thickened scaly skin, are common in many individuals with CFC syndrome.

Heart problems such as ventricular septal defects (hole in the heart) or pulmonic stenosis (narrowing of one of the arteries in the heart) may be present at birth. Other heart problems, such as hypertrophic cardiomyopathy (thickened heart muscle) or arrhythmias (heart rhythm problems) may develop over time,

therefore, close monitoring by a cardiologist is important.

In addition, most individuals with CFC experience growth delays, and may need special assistance with feeding. Mild to severe intellectual disability, with delays in areas of speech, language, and motor skills are common to all children with CFC, however there is a huge range of ability and children will often continue to make developmental progress over time.

Vision abnormalities including strabismus, nystagmus, and optic nerve hypoplasia are present in most individuals and may result in decreased vision and acuity.

Various types of seizures can be seen in up to 50% of individuals with CFC syndrome and should be managed by a neurologist or specialist in seizures/epilepsy.

Because of the various medical and developmental issues that may occur in CFC syndrome, it is very important that all individuals diagnosed with or suspected of having CFC syndrome be referred to a geneticist. Multidisciplinary care involving specialists, doctors, and therapists is essential in providing the most up-to-date medical management and care for these children and adults. Healthcare guidelines for CFC syndrome can be found on our website www.cfcsyndrome.org.

What Causes CFC Syndrome?

CFC syndrome is caused by mutations (changes) in one of four different genes called BRAF, MEK1, MEK2 and KRAS. These genetic changes usually occur for the first time in the child, and are rarely inherited from a parent. Therefore, the chances of having another child with CFC syndrome would be very low. There is nothing that either parent did or did not do during the pregnancy to cause these changes in the genes; they just happen by chance.

Diagnosis

Diagnosis is based on clinical findings and genetic testing. Genetic testing is now available at a variety of laboratories throughout the world. To obtain a listing of the labs please check www.genetests.org or contact your local genetics specialist.



Family Services

Family Liaisons are available to provide support and guidance. CFC International is involved with networking individuals, families, doctors and therapists for the purpose of family support, education, medical treatments and knowledge.

Our organization also publishes a newsletter and a parent's guide. We host biennial international family conferences, a website, and a private family computer listserv. You can find us on Facebook at www.facebook.com/CFCSyndrome.

Research

CFC International has a vision and researchers have the determination to understand CFC and to find treatments to help people with the syndrome overcome the many challenges they face.

Through its **Small Grants Program**, CFC International has funded several research projects including studies to investigate growth in CFC syndrome and a consensus meeting to help publish management guidelines for this rare syndrome.

Additionally, CFC International maintains a worldwide registry of patients along with each person's genetic information.

