



Behavior in Cardiofaciocutaneous (CFC) Syndrome

What is CFC?

How does it affect a person?

CFC is a rare genetic syndrome that typically affects a person's heart (cardio-), facial features (facio-), and skin (cutaneous).

It is equally prevalent among males and females across all ethnicities, and is known to affect approximately 1 in every 810,000 individuals.



Common features of individuals with CFC (not seen in all cases):

- A distinctive facial appearance
- Unusually sparse, brittle, or curly hair
- Skin abnormalities
- Heart defects since birth or those that might develop later
- Growth delays
- Varying degrees of intellectual and/or learning disability
- Vision issues
- Seizures

What causes CFC?

CFC is caused by random changes in a person's genetic code, specifically in genes called *BRAF*, *MAP2K1*, *MAP2K2*, and *KRAS*. Genes can be thought of as instructions that tell a body how to function in a particular way. With random changes in these instructions, the body and brain develop in a way that is different from what is typical. It is important to note that these genetic changes in CFC usually occur for the first time in the child, and are not inherited from the parents. As a result of these random changes, children with CFC develop unique characteristics and behaviors.

Our Research Study:

With an aim to provide information and help families address the neurocognitive and behavioral effects of CFC, we investigated the behavioral functioning of 34 children diagnosed with CFC and assessed its impact on the caregiving experience through several questionnaires.

Our objectives were to:

- Obtain an improved characterization of behavioral features associated with CFC
- Identify key medical and developmental factors associated with behavioral outcomes
- Investigate how behavioral challenges impact caregivers

We have summarized our findings as answers to some frequently asked questions.



What are some typical behaviors seen in children with CFC?

A number of the most common behavioral symptoms of children with CFC were related to two areas: difficulties maintaining attention, and engaging in repetitive or unusual thinking and behavior patterns. More specifically, many children with CFC were described as:

- Restless or hyperactive
- Unable to concentrate for long
- Easily distracted
- Unable to complete tasks they started
- Unable to get their minds off certain thoughts
- Repeating acts over and over
- Having difficulty sleeping

Parents often reported that their child with CFC was highly dependent on caregivers and demanded a lot of attention.

Relative to the general population, children with CFC are more likely to experience emotional difficulties directed inwards (towards themselves), like feeling lonely or being socially withdrawn. These so-called “internalizing” behaviors were clinically significant in 29% of children with CFC. Children with CFC were also at greater risk for emotional challenges directed outwards (toward others), like engaging in physical aggression, acting defiant, or breaking rules. These “externalizing” behaviors were clinically significant in 21% of children with CFC, although this was not one of the most common problem areas.



Why do children with CFC exhibit these behaviors?

Our study examined how the children's sensory modulation (their ability to receive and sort out different kinds of sensory input like touch, smell, sight, or sound) and functional communication (their ability to communicate important messages including basic needs and desires) impacted their behavior. We found that ***behaviors exhibited by children with CFC were closely associated with how they respond to sensory input.***

Many children with CFC had high levels of "**sensory sensitivity**", and were easily overwhelmed by sights, sounds, or textures. These children exhibited a higher frequency of emotional dysregulation, such as feeling anxious, being withdrawn, acting irritable, or lacking energy or interest. Children with CFC who had "**sensory-seeking**" needs were more likely to act in an aggressive, disruptive or self-injurious manner.

We wondered whether difficulties in trying to communicate might be frustrating to children, resulting in more problematic behaviors. Our results showed that better communication skills were not always accompanied by better behavioral regulation.

The ability of children with CFC to communicate messages to others in an effective way did not have a significant association with their behavioral features.

Could greater medical and neurological severity related to CFC help explain some of its challenging behaviors?

We examined the presence or absence of seizure history, cardiac disease, birth complications, and gastrointestinal/feeding issues for each child and related it to behaviors. Birth-related complications were associated with a higher frequency of behavioral challenges, but none of the other medical issues predicted the extent to which a child exhibited behavioral problems.





What are some strategies that I can use to address my child's behavioral challenges?

1. Limit exposure to unfamiliar or excessive sensory input in the child's daily life, and/or introduce new sensory experiences to the child's life in a gradual and progressive manner.

2. For children who are resistant to change due to the overwhelming nature of unfamiliar stimuli, help them to anticipate what will come next and/or prepare them to expect the change. This can be done by preparing them

with calm, simple verbal directives or visual cues of what is coming. When a transition occurs or a new situation is introduced, the child may respond well to a visual activity schedule or a "social story" to help them fill in gaps in their understanding.

3. Children with milder cognitive deficits who tend to withdraw from or avoid social situations may benefit from having teachers/caregivers/therapists teach them "active" coping strategies to address stressful stimuli. Examples of "active coping strategies" include engaging in calming activities, problem solving, or seeking support from others.

4. For children with significantly delayed cognitive/communicative abilities, caregivers might want to expect a longer time for the children to be able to make connections through what is being told or taught; some children may have specific preferences such as visual stimuli or listening to "stories" (for those with better language comprehension) that clinicians and caregivers might want to use to help them learn.

5. For children with sensory-seeking needs, enabling activities that allow for greater movement or encouraging more purposeful, creative exploration may be beneficial.

6. In school settings, children who engage in challenging behaviors may benefit from a functional behavior assessment (FBA) to learn the purpose of a specific problem behavior (such as withdrawal, refusal to participate, disruptive behaviors or aggressive or self-injurious behaviors). This may prompt a logical plan to reduce the frequency or intensity of a challenging behavior or replace it with a more adaptive behavior.

7. If your child loves to receive attention, he or she may engage in behaviors to attract a response from you, even if it is a negative response. As long as the behavior is not unsafe, try ignoring it and giving no feedback. This may be enough to get rid of the behavior!

8. Reinforce positive coping and behaviors. If you identify things that are rewarding to your child (e.g., “high five,” special time with a parent, a preferred toy or activity), these can be used to encourage and shape positive behavior patterns.

Who can I go to for professional help?

Owing to the wide range of behavioral effects of CFC, ***coordinated services among a comprehensive education and treatment team will be necessary to help children and families manage the complex neurological and behavioral effects of CFC.*** This team will be key to identifying specific neurobehavioral strengths and weaknesses of an individual and for moving forward with informed treatment and educational planning.



Members of a comprehensive treatment team may include: physician specialists, neurologist, neuropsychologist, behavior specialist, social worker, rehabilitation therapists (e.g., speech/language pathologist, occupational therapist, physical therapist, feeding specialist), vision specialist, special education teachers, and other mental health professionals such as an Applied Behavioral Analysis specialist who could help develop a behavioral intervention plan.

Would medications be helpful for my child?

Should you consider using medication to treat behavioral issues, it is important to consult with a medical team that has full understanding of the overall health implications of the treatment for a medically and neurologically complex child. It is also important to be aware that some medications used to treat seizures, pain, or other co-occurring medical conditions (e.g., steroid medications) can significantly impact a child's emotional functioning and behavior.

In our study, 24% of parents reported that their child had taken medication to treat mood or behavioral issues, including stimulant or non-stimulant medications to manage attention or activity levels; antidepressants; or antipsychotics. While stimulant medication is known to be effective in treating ADHD symptoms of children with a related genetic syndrome called neurofibromatosis 1, no studies have yet examined the safety and efficacy of these medications specifically in CFC.



When and why do I find myself feeling stressed? What can I do to relieve it?

Parenting stress among caregivers of a child with CFC was found to be significantly higher for the caregivers whose children had more limited functional communication (i.e., had a hard time expressing their needs and wants) and those with greater overall problem behaviors. In fact, caregiver stress was more related to the extent of emotional and problematic behaviors a child had than to the severity of the child's disability or developmental delay. Further, difficulty coping with stress was more related to concerns about communicating with family/providers and less associated with the medical care aspects of parenting their child.

These findings suggest that interventions focused on improving the functional communication ability and overall behavior of children with CFC have the potential to positively affect the whole family and increase the well-being of parents.

Suggestions to help you cope and build resiliency:

1. Connect with support groups and other families that have children with special needs (specific resources below)
2. Seek out counselling and therapeutic services in the community if you find yourself experiencing frequent distress
3. Determine whether you are eligible for services of a Personal Care Aide/Assistant (PCA). Some families may be eligible for home -and community-based attendant services under Section 1915(k) of the Social Security act as part of a Medicaid state option in the United States.
4. Participate in parenting programs for kids with special needs

What resources are available to help my family and my child with CFC?

Support Groups & Social Media:

CFC International is a support group for lives touched by CFC.



<http://www.cfcsyndrome.org/>
<https://www.facebook.com/CFCSyndrome/>

RASopathies Network connects people interested in learning more



about rare genetic syndromes like CFC, Costello syndrome, Noonan syndrome, neurofibromatosis type 1 (RASopathies)
<https://rasopathiesnet.org/>

For Educational Advocacy:

If you have questions or concerns about how your child's educational programming (e.g., special education supports), eligibility, etc. relate to behavior, we recommend that you contact the PACER center: <http://www.pacer.org>

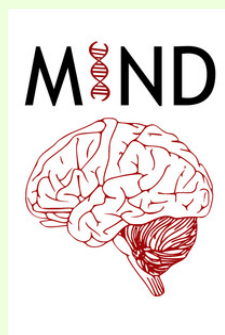
A large challenge for families is attaining some level of inclusion for their child in schools in the US. SWIFT is a good model for different ways to attain inclusion.
<http://www.swiftschools.org/>

Recommended Books:

1. Brown, I. (2011). The boy in the moon : A father's journey to understand his extraordinary son (1st U.S. ed.). New York: St. Martin's Press.
2. Biel, L., & Peske, N. K. (2009). Raising a sensory smart child : The definitive handbook for helping your child with sensory processing issues (Updated & rev. ed.). New York, N.Y.: Penguin Books.
3. Voss, A. (2011). Understanding your child's sensory signals : A practical daily use handbook for parents and teachers. Charleston, SC: publisher not identified.
4. Kranowitz, C. S. (2006). The out-of-sync child has fun : Activities for kids with sensory processing disorder (Rev. ed.). New York, N.Y.: Perigee Book.
5. A link to an article listing “top 10” books for parenting children with disabilities:
<https://www.brainchildmag.com/2015/01/top-ten-books-for-parenting-children-with-disabilities/>

Our Research Team:

The Minnesota Investigators in Neurodevelopmental Disabilities (MIND) Group



We are a collaborative group of researchers at the University of Minnesota who are working with children and families to gain scientific insights that will improve the lives of children with neurodevelopmental differences.

<https://sites.google.com/a/umn.edu/mind/>