Costello Syndrome

- Very rare genetic disorder
- Results from a pathogenic variant (mutation) in the HRAS gene
- Feeding issues and failure to thrive (FTT)
- Heart differences
- Neurological concerns
- Developmental delay/Intellectual disability
- Orthopedic concerns
- Short stature
- Vision concerns
- Increased chance of cancer
- Skin issues
- Characteristic facial features: full cheeks, full lips, large mouth

HELP US RAISE FUNDS FOR THE
COSTELLO SYNDROME FAMILY NETWORK

For more details visit:

costellosyndromeusa.org

Costello Syndrome
FAMILY NETWORK

CSFN (Costello Syndrome Family Network)
A 501(c)3 non-profit organization
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facebook.com/CostelloSyndromeFamilyNetwork
Charity EIN: 02-0622876

ICSSG (International Costello Syndrome Support Group)
UK Registered Charity Number 1085605
http://costellokids.com

A pamphlet for parents by parents

REVISED OCTOBER 2024

COSTELLO SYNDROME

raising awareness & hope

Costello Syndrome
FAMILY NETWORK

Since 2005, when the gene responsible for Costello syndrome was identified, we have been learning more and more about how each variant (mutation) in the gene affects the body. The HRAS gene is the only gene associated with Costello syndrome. However, there are several different variants within the gene that alter the way each individual is affected. The most common gene variant is p.G12S. About 4/5 of all individuals who have a diagnosis of Costello syndrome have the p.G12S variant. It appears that some variants like p.G13C, though much rarer, may cause a milder phenotype.

The great news is that almost all individuals with Costello syndrome have a great sense of humor. Their charm and sheer resilience are incredible. This helps parents and caregivers get through the rough times. Our children are truly amazing!

Gripp et al (2019) Costello syndrome: Clinical phenotype, genotype, and management guidelines. Am J Med Genet A, 179(9): 1725-1744. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC8238015/NBK1507/

Common concerns in individuals with Costello Syndrome

The first hurdle parents normally encounter is ***failure to thrive (FTT),*** when growth and weight gain is less than expected. This can happen soon after birth or several months later, and is usually due to swallowing difficulties and reflux. Many require a gastronomy tube (G-tube) and extra calories. Fortunately, most children who get a G-tube outgrow the need for it between the ages of 2 and 14.

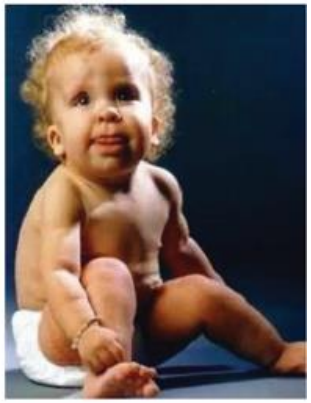
Many children with Costello syndrome are born with, or go on to develop, a ***heart issue.*** These issues include arrhythmias, pulmonary valve stenosis, and hypertrophic cardiomyopathy.

Neurological concerns may impact individuals with Costello syndrome. Chiari I malformation (a malformation of the lower part of the brainstem) and/or tethered cord (when the spinal cord is "tied" to the bottom of the spinal column) are two diagnoses that some with Costello syndrome may encounter.

Developmentally, children with Costello syndrome learn more slowly than other children the same age. Most children have some speech delay. The onset of speech frequently coincides with the ability to feed orally. People with Costello syndrome have a good memory for stories and events. They will need a specialized education program in school to help with academics.

Children with Costello syndrome have ***orthopedic concerns.*** Most children learn to walk. For some this is complicated by tight Achilles tendons and loose joints.

Hypotonia (low muscle tone) and contractures (shortened and stiff joint) challenge many with Costello syndrome. Physical therapy may be helpful. Many teens and young adults with Costello syndrome have been diagnosed with scoliosis.



Endocrine issues such as precocious or delayed puberty can be seen. A number of individuals have a documented growth hormone deficiency for which they receive growth hormone therapy.

Vision concerns are also common. Many individuals with Costello syndrome have delayed visual maturation, ptosis (droopy upper eyelids), nystagmus, strabismus and may be prescribed glasses.

The ***risk of cancer*** is increased in individuals with Costello syndrome. The most common type is embryonal rhabdomyosarcoma, a childhood cancer. Bladder carcinoma is another type of cancer found in people with Costello syndrome. Regular ultrasounds are recommended to catch the cancer early for better outcomes. Fortunately, this cancer responds well to treatment. A third type of cancer that can be seen in those with Costello syndrome is neuroblastoma.

Dermatologic, or skin, issues affect almost all individuals with Costello syndrome. Most prevalent are deep creases on the palms and soles, hyperpigmentation, and loose skin. Hyperkeratosis, or thickening of the outer layer of the skin, causes calluses on the hands and feet. It may appear in pre-teens through adulthood. Also, many with Costello syndrome will have papillomas, a type of skin tag, which may appear on the face, underarms, on the nose, etc.

In Loving Memory of:
Dr. Ginny Proud who dedicated much of her professional life to Costello Syndrome.