

In the meantime, treat each of your child's problems based on the speciality of the problem. Don't forget that your child shares your genes - and has needs and responses like any other child. Not everything can be "blamed" on the syndrome ☺

While they have incredibly challenging issues, our children are wonderful sons and daughters.

Photos (credit the parents of): Kelsi Moore, Michael Weber, Shelby Adams, Helaina Stone, Kelsi Moore's hand and foot, Jayne Keizer, Jalisa Sullivan at birth, 3 and 8, and Quin Johnson (in memoriam).

* Van Eeghen et al, *American Journal of Medical Genetics*, 82:187-193 (1999). This article has an excellent chart of involvements.

Please Contact Us!

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(International Costello Syndrome Support Group)

UK Registered Charity Number 1085605

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COSTELLO SYNDROME



A BROCHURE BY PARENTS FOR PARENTS

Welcome to our group!

We hope to provide you with support and understanding, and share what we know as we learn more about this very rare syndrome. If you are interested in looking at more photos of our children, they are available at the website described on the last page.

“**Costello syndrome** is a distinct multiple congenital malformation syndrome characterized by postnatal growth retardation, distinctive face, lax skin, and developmental delay. The cause remains uncertain; although the facial traits bear a remarkable resemblance to a storage disorder, there is no progressive coarsening with age. Either an autosomal dominant mutation in a single gene or a microdeletion seems the most probable cause.”*



We all live in this limbo.

Many of us first received our children's diagnosis as “probable,” having some but not all the signs, with poor chances of it becoming definite. This is because there is no way, other than by clinical observation to make the diagnosis.

You may have heard from your child's geneticist that your child might have a combination of disorders, possibly some of which are not yet defined. Some of you have had many biopsies done to help try to define your child's disorder, and they all come back “Within Normal Limits.”

the next doctor, you have all the information at hand. It makes a **big** difference! *Even if you don't understand them*, being able to provide reports to each specialist helps that specialist immensely.

Remember that YOU are with your child for life, and the specialists are not. Trust your instincts, and do what you can to educate yourself so that you are taken seriously when you meet with the specialists your child will need to visit. If you have access to the internet, join our listserv! (see last page)



IDEAS: There is a thought from the UK that Costello syndrome has to do with the delayed switching of a genetic developmental code. This makes medical sense with respect to the feeding, cancer, and heart problems. Incredible research work is being done internationally on our kids' syndrome. Members of our Medical Advisory Board are actively researching and recruiting researchers. Your participation would be greatly appreciated!

HANG IN THERE!

Our children are “globally developmentally delayed,” but they generally eventually reach all the childhood milestones. Thank goodness for their “**warm, social personalities**” -- when it kicks in (usually after the gastrointestinal issues start getting resolved). It seems that wherever they go, they endear people to them. And their sense of humor – mature beyond their developmental age – helps us help them through the tough times. Have you noticed?

Because of the rarity of this syndrome, your child needs to be seen by the best in each field – if possible. Oddly, it’s important to hear “I don’t know” from the specialists, because it means they’re being honest with you. You are building and maintaining a team to support you. You need to be able to trust them with what they know – and don’t.

A typical child with Costello syndrome may see many doctors, including:

- ⊖ a geneticist,
- ⊖ a gastroenterologist,
- ⊖ a cardiologist,
- ⊖ an orthopedist,
- ⊖ a neuro-ophthalmologist,
- ⊖ a pulmonologist,
- ⊖ an ENT (ear, nose and throat) specialist,
- ⊖ a pediatric dentist specializing in craniofacial deformities,
- ⊖ a neurologist,
- ⊖ a developmental pediatrician,
- ⊖ in addition to the primary pediatrician.

Children – people with Costello syndrome are amazingly tough survivors.

YOU ARE THE EXPERT ON YOUR CHILD

Be prepared to be the one who knows the most about your child, and the integrator of your child’s medical and educational needs. If you’re up to it, ask for copies of all your child’s medical reports so that when you visit

Another thing we notice about Costello syndrome beyond the two most common identifying features (feeding issues, or **Failure to Thrive** and global developmental delay) is its **breadth**.



With what we guess to be about 150 cases identified WORLDWIDE, our children **are** the information from which the scientists and doctors are making theories! If we round up to 200, then your child is 1 in 30 million.

The Importance of Parent-to-Parent Communication

In addition to doctors and researchers observing our children for patterns, we parents have been important participants in developing a more detailed picture of what Costello syndrome is. Our direct communication with each other speeds up the identification of a pattern, and starts an immediate database for a doctor interested in following the research.

Among the shared observations many parents notice but is still a mystery is that our kids are “hot-blooded.” Whether it’s the metabolism or the heart issue (both can cause a person to be hot all the time) or something else, or all of the above, our kids appear to **sweat a lot** - even to be a bit “ripe-smelling.” Some children experience **unexplainable fevers**.

Many parents notice their child is very **sensitive to sunlight** and **touch**, particularly the hands and feet.

Most parents talk about difficulty **sleeping** through the night. Many grow out of it, but many don't.

THE GASTROINTESTINAL SYSTEM

This system is just about always involved, and some kind of intervention is required. **This is, by far, the most chronically difficult issue for our families.** Families outside the USA and Canada tell of spending time in the hospital every couple of months or so, with NG (nasogastric) tube feedings and iv.'s for dehydration. Others feed their children via NG tube at home.

In the US particularly, most kids have **g-tubes** (gastrostomy tubes), which go directly through the stomach wall for feeding either by "bolus" (pouring in the formula or meal in one sitting) or timed drip-feeding (requiring a pump, often scheduled for feeding overnight)). Some children have a surgical procedure, a Nissen's fundoplication, performed (where a surgeon puts an extra fold in the esophagus just above the stomach to help reduce reflux). But for some children, the "fundo" is not appropriate.

Our children tend to need more calories too.



We've noticed that a good number of our kids **metabolize pain medications** very quickly. Be sure that you tell your child's doctors so that pain management can be more effective. If your child needs surgery, be sure to enlist the anesthesiologist's help. One parent was alerted by a

very observant anesthesiologist, who not only gave a copy of his report to her, proving how her child had needed more anesthesia, but he also instructed her to be vigilant with each future anesthesiologist her child needed.

SEIZURES?

There appear to be increasing numbers of newly diagnosed children with seizures -- but we don't have enough data to link it to Costello syndrome. This may be something that's showing up in addition to the Costello syndrome issue. One child had seizures that went away when surgery was done on her Chiari malformation (a deformity in the brain) -- which, prior to the surgery, wasn't clearly the source of the seizure. If more parents report on this issue, we will have better information from which to prove or disprove a link!



THE SKIN

Your child's doctor will probably bring up **papillomata** – wartlike skin growths, because they are in the literature. These may start to show on your child starting at age 2, or they may never show up. Doctors do recommend that any skin eruption be tested to distinguish between papillomata, which can be benign tumors, and wart-like 'lesions.' (see Van Eeghen article quoted at the beginning of this booklet and cited at the end.)

Some children suffer from **Acanthosis Nigricans**, the darkening of the skin. Dermatologists' treatments for this appear to work fine, for the most part. **Itchy skin**, from mild to extreme, is also a common problem.

A FEW OTHER THINGS

Many parents also talk about trouble with stuffy and/or runny noses. Combinations of over-the-counter medications mostly help. Those who have had **tonsillectomy** and/or **adenoidectomy surgery** do better. Several children who didn't sleep well did, after this surgery – but not all.

Some kids also have **tracheal** and/or **laryngeal malacia** (floppiness with each breath), for which a small number of kids needed a tracheostomy. The good news is that they appear to grow out of it. One child who had a trach (**tracheotomy**) grew out of the need for it in a couple of years.

The good news is that **the feeding issues usually resolve themselves** somewhere between the ages of 2 - 8 years old. There are a few cases where the child will clearly be a g-tube-fed person for life. There are also a few cases where the child never had an eating problem!

Many children have difficulty with **constipation**. All do better with more fluids, but it's hard to get a child –any child- to drink enough! If your child is having trouble with constipation even with what the doctor can prescribe, you may find some useful suggestions on our website's Frequently Asked Questions (FAQ) section.

ORTHOPEDICS

Hips and **ankles** are involved so that sitting and walking will be delayed. Several children have wheelchairs to help them get around school, which over time have been replaced with walkers. (They have little protection reflex if they fall.) While his protection reflex isn't the greatest, one boy loves to play basketball and ski -- unassisted!

Many children wear ankle-foot orthotics (AFO's or DAFO's), and have their Achilles tendons "released." Hip tenotomies (tendon-releasing surgery) have also been performed on several children, with generally good success. A few have had hip reconstruction surgery.

Most kids have trouble straightening out their **elbows**. In Australia, the description of the arm position is called "kangaroo arms."



Most kids' **hands** (and feet) have lots of loose skin (cutis laxa), are *very* flexible, and tend to turn away from the midline (ulnar deviation – towards the ulna bone in the forearm). The flexibility makes it hard for them to build fine-motor strength in their hands.

The **ribs** are unusually shaped, which may explain why many of the children don't go through a crawling phase, even with therapeutic intervention. Several children have **scoliosis** (the spine curves sideways) and have braces for their backs. Some have had surgery. There are a few children whose bones are so soft that they pose difficulties. One child's doctors had trouble putting in rods to correct the scoliosis because the bones were so soft.

Kyphosis (the spine curves outward) is not unusual either, but the thought among orthopedists these days is to leave it alone, as the cure is worse than the problem.



THERAPIES

Our kids respond very well to **Occupational** and **Physical Therapy** (OT and PT); and **early intervention** (something that in the US is provided by the public school system and state programs).

Our children's receptive language skills appear to be much better than their expressive skills. **Speech** is delayed and limited. It's probably connected to the poor oral-motor coordination overall, small mouths and slightly larger-than-normal tongues (macroglossia). **Speech therapy** (ST) is strongly recommended, as the children do appear to respond well. Many children learn sign language and may have communication boards to help them until (or if) their speech develops well enough.

VISION

A higher-than-average number of kids are **myopic** (near-sighted), often with **lateral-beating nystagmus** (the eyes jiggle – horizontally), and some have **strabismus** (cross-eyes). Except for some kids having delayed vision (several parents talked about being fooled by their child's effective use of hearing to mask their visual delay!), most visual issues appear to be correctible with glasses.

THE HEART

Just about all our kids have heart issues, but this can be anything from mild arrhythmia to such severe HCM (Hypertrophic Cardiomyopathy) that the child dies from it (not common). Because of this, it is recommended that your child get an **echocardiogram** done to rule out any problems. An article by Angela Lin and associates, *Further delineation of cardiac abnormalities in Costello syndrome*, published in the American Journal of Medical Genetics in 2002, will give your doctor medical justification for this test.

CANCER

There have been around 18 published cases of cancer or about 18% of the documented kids. Karen Gripp and associates' 2002 article, *Five additional Costello syndrome patients with rhabdomyosarcoma: proposal for a tumor screening protocol*, in the American Journal of Medical Genetics is a good resource. We urge you **NOT** to be worried about cancer before testing, because the odds are still smaller than one in five (1:5).

Some screening tests, like for neuroblastoma (using a urine sample) actually have more false positives, so they could make us worry needlessly. The most common cancer is embryonal rhabdomyosarcoma (RMS), which has no screening test. Lisa Schoyer (a mom) is very interested in collecting information about this, so if your child does develop embryonal RMS, please contact her. She would appreciate all the data that can be gathered, in order to be as accurate as possible. (See back panel.)