

Cockayne Syndrome: A Manual for Parents and Caregivers

National Initiative for Cockayne Syndrome
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National Initiative for Cockayne Syndrome

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Dedication

Cockayne Syndrome: A Manual for Parents and Caregivers is dedicated to all of the clinicians who see, treat, and are inspired by our Cockayne syndrome (CS) children, and to the parents and caregivers who provide respite and loving care for them.

To all of the parents around the world who have walked this path daily, shedding light and working so feverishly to get information as well as support and advocate for CS research: we thank you. For the families who have children living with CS: we walk beside you. For the families who received a recent diagnosis: we know the grief you experience, but we are grateful that you have found this manual. We understand the path you walk, and you are never alone.

The dream to compile this comprehensive manual was inspired by many CS parents and children/angels around the world. The hope is to have a go-to resource providing guidance for clinicians and the medical team, and support for parents and caregivers. Many of our CS angels sacrificed for lessons our future CS kids will benefit from. Their lives were never in vain.

Sincere thanks to many CS parents around the world who came together to provide feedback on what would be necessary to include in this manual. We would like to specifically acknowledge Amy Marini, Haylee Carroll, Nikki Cohen, Missy Miller, Christina Polchin, and Maria Pellicane—all of whom are CS Moms with the power to get anything done that they put their minds to.

And to our Clinical Advisory Board members: without you and your amazing contributions, this manual would never have taken the form that it is before us. We are forever grateful for all of your hard work. We know that this manual is not the end of your CS work—that children not even diagnosed yet will benefit from all of your research and expertise for years to come.

Additionally, we would like to acknowledge Thistle Editorial LLC, who provided assistance with content development.

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Introduction

A Note From the Authors

We developed this manual with two parents of children with Cockayne syndrome, or CS. Other families from all over the world have also shared information. We hope you find this manual helpful. We know the path you walk, and we want you to know that you are never alone.

Why We Wrote This Manual

This manual is to help you and your child's medical team better understand Cockayne syndrome. It explains some of the most common health concerns for children with this disorder. However, it does not replace the advice of your child's doctors. If your child needs medical care, you may want to use this manual to learn about what your doctors are doing and make suggestions.

Every child is different, including children with Cockayne syndrome. Your child may experience things this manual does not cover. We are constantly learning new information, and the manual will change as we learn more. In the meantime, we hope the information here helps you and your child live better with Cockayne syndrome.

What Is Cockayne Syndrome?

Cockayne syndrome, or CS, is a rare genetic (juh-NET-ick) condition that runs in families. A gene is a small piece of information that tells the body how to grow and develop. People with

Tips From a CS Parent

It's important to find a pediatrician you trust.

Your pediatrician will work with you and other clinicians to make sure your child gets specialist care.

Cockayne syndrome have changes within specific genes, and their bodies grow and develop differently from most people's.

About 8 in 10 children with Cockayne syndrome have a mutation on a gene called *ERCC8*. About 2 in 10 have a mutation on the gene *ERCC6*. Some other mutations are related to CS, but these are more rare.

Children with CS have much smaller heads than normal, a condition called microcephaly (my-crow-SEFF-uh-lee). They also do not grow and gain weight as quickly as other children. They are shorter than other children and often cannot do the same activities at the same age. Some children with CS have specific face and body changes.

Some doctors divide Cockayne syndrome into several different types. These are categorized according to when CS develops, how a child is affected, and how severe the condition is. However, the differences between types are not always clear. Some doctors and scientists believe there is a range of Cockayne syndrome

symptoms instead of separate types.

Children with CS have many different needs. They usually see several doctors and other medical specialists for care. The table on the right shows a list of different specialists that you and your child might see.

Treatments Your Child Might Need

Children with Cockayne syndrome usually need different types of treatment, also called therapy, from their medical teams. Here are some types of therapy that your child might need, depending on their symptoms:

- Physical therapy
- Occupational therapy
- Speech therapy
- Hearing therapy, including a teacher for the deaf or hard of hearing
- Vision therapy
- Water (pool) therapy
- Equine (horse) therapy

Name of medical specialty	This specialist cares for your child's...
Audiology	Hearing
Cardiology	Heart
Dentistry	Teeth and mouth
Dermatology	Skin
Otolaryngology	Ears, nose, and throat
Endocrinology	Glands and hormones
Gastroenterology	Stomach, digestion, and bowel movements
Genetics	Inherited conditions, including learning about the genes that cause CS
Hepatology	Liver
Neurology	Nervous system, including the brain and spinal cord
Nephrology	Kidneys
Nutrition	Diet
Ophthalmology	Eyes and vision
Physiatry	Muscles and bones, including how your child walks and does activities
Pulmonology	Lungs and breathing
Urology	Urine system and genitals (sex organs)

Audiology

Caring for Your Child's Hearing

How CS Affects Hearing

Hearing loss is a common problem for children with Cockayne syndrome.¹ Your child might have hearing loss when they are born or develop it later. Some children with CS lose a small amount of their hearing, some lose a moderate amount, and some have serious hearing problems.^{2,3} The hearing loss usually affects both ears and gets worse as your child gets older.²

You and your child will see an audiologist if your child has hearing problems.

Some children with Cockayne syndrome have difficulty responding quickly. This can make it seem like they did not hear you, but they actually just take longer to get moving after they hear you. Talk with your audiologist if your child responds slowly. They might have a hearing problem, or there might be another cause for the slow response.

Tests for Hearing Problems

Your child may need diagnostic tests to learn how well the ears are working and how well they hear. These tests might include some or all of the following:

- **Hearing booth test:** This test is done in a soundproof booth. The audiologist will play various sounds to learn if your child has hearing loss in either ear. Your child might need to do something to show they hear the sound, like

Meet Your Specialist Audiologist

An **audiologist** is a specialist who studies and treats hearing problems.

Audiology is the branch of science and medicine that deals with the sense of hearing.

raising their hand, or the testers might have them respond a different way.

- **Nerve response test:** This is also called an **OAE test** (an otoacoustic test). The audiologist tests the nerves inside your child's ear to see how well they respond.
- **Brain response test:** This is also called an **ABR** or **BAER test** (an auditory brainstem response or brainstem auditory-evoked response). The audiologist tests how the hearing part of your child's brain responds to sound. Your child might need medication for this test to make them relaxed and sleepy. This is because they need to be very still during the test.
- **Air pressure test:** This is also called **tympanometry (tim-pan-OM-uh-tree)**. The audiologist measures how your child's eardrum and middle ear area respond to air pressure.

Treating Hearing Problems

If the hearing loss is mild or moderate and your child can still hear, they might benefit from hearing aids. If your child's hearing gets worse, the audiologist may talk with you about an implant to help your child hear. This is placed inside the ear with surgery, typically by an otolaryngologist. It is called a **cochlear implant (COCK-lee-ur implant)**.⁴

How Often Should My Child See an Audiologist?

Your child should see the audiologist at least once a year.⁵ However, they may need to see the audiologist as often as every six months or more, based on your doctor's recommendations. Children who have hearing problems often have eye problems, too, so it is a good idea to see an eye doctor (ophthalmologist) as well.⁵

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Medical Terms You Might Need

Audiologist (odd-ee-OLL-uh-jist):

A medical professional with a doctoral degree in audiology who tests how well someone can hear. They can recommend hearing aids and other things to help with hearing loss. They often work with ear, nose, and throat (ENT) specialists or otolaryngologists.

Cochlear implant (COCK-lee-ur implant):

A small device a doctor can put inside a child's ear to help them hear.

Otolaryngologist (oh-toe-larr-in-GOLL-uh-jist):

A doctor who specializes in treating ear, nose, and throat conditions. Some of them also specialize in treating children. This type of doctor is called a pediatric otolaryngologist. These doctors are also known as ear, nose, and throat (ENT) specialists.

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Cardiology

Caring for Your Child's Heart

How CS Affects the Heart

Children with Cockayne syndrome develop heart and blood vessel problems that usually happen in older adults. These include hardening of the arteries, heart murmurs, blood vessel disease, and high blood pressure.¹⁻³ Blood vessel disease can sometimes cause a stroke.²

High blood pressure can also be caused by a kidney problem. You may want to read the [Kidneys](#) section of this manual. Ask your child's doctor to check your child's blood pressure at each appointment so you know what it should be.

Your child might have cold hands and feet. This happens when CS makes the small blood vessels in these areas even smaller. You can rub your child's hands and feet, give them a warm bath, cover them with a blanket, and make sure they have warm socks and shoes.

Tests for Heart Problems

Your child will have tests to learn how well their heart is working. These may include:³

- **Echocardiogram (eck-oh-CAR-dee-oh-gram):** This is also called an **echo**. This test uses sound waves to create a picture of the heart. It allows your doctor to see the structure of your child's heart and how well blood is flowing inside it.

Meet Your Specialist Cardiologist

A **cardiologist** is a specialist who studies and treats heart problems.

Cardiology is the branch of science and medicine that deals with heart problems.

- **Electrocardiogram (ee-leck-tro-CAR-dee-oh-gram):** This is also called an **EKG** or **ECG**. This test measures electrical activity in your child's heart. All hearts have electrical activity. This test tells the doctor if your child's heart is working normally.

Treating Heart Problems

Your child's doctor can check their blood pressure. They can also help you decide when to see a cardiologist to treat specific heart problems, if needed.³

How Often Should My Child See a Cardiologist?

Your child's doctor can tell you how often your child should see a cardiologist. You may see them every year or more often, or only if your child develops heart problems.

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Medical Terms You Might Need

Atherosclerosis (ath-uh-row-scluh-ROW-sis): The medical term for hardening of the arteries. Arteries are the blood vessels that carry blood from the heart to the rest of the body. Old age and diseases such as CS can make them stiff and hard, and this can lead to problems.

Echocardiogram (eck-oh-CAR-dee-oh-gram): Also called an echo. This test uses sound waves to create a picture of the heart.

Electrocardiogram (ee-leck-tro-CAR-dee-oh-gram): Also called an EKG or ECG. This test measures electrical activity in your child's heart.



Dental and Oral Medicine

Caring for Your Child's Mouth and Teeth

How CS Affects Teeth

Your child may be more likely to have problems with their teeth than other children if they have CS.^{1,2} These problems can include:³

- **More cavities than usual.** Brush your child's teeth twice a day to help prevent cavities.³ If your child will not open their mouth, you can clean their teeth with a washcloth or NUK brush.
- **Thin enamel.** The medical term for this is enamel hypoplasia (uh-NAM-ul hi-po-PLAY-shuh). The enamel is the hard, white outside layer of your teeth. Thin enamel can cause:³
 - » Small dents in the teeth
 - » Yellow, brown or rough-looking teeth instead of smooth white teeth
 - » Teeth that are an unusual shape or size

Your child should see a dentist as soon as they get their first tooth. They may need X-rays and treatment if there are any problems.

Your child's teeth might be sensitive to hot or cold foods and drinks. The roof of your child's mouth might also be higher than usual.²

Changes as your child grows

Your child with CS may be slower to get their baby teeth than other children. They might lose these teeth earlier or around the usual time. They might be

Meet Your Specialist Dentist

A **dentist** cares for the teeth and mouth.

Dentistry is the branch of science and medicine that deals with studying, preventing, and treating problems of the mouth and teeth.

more likely to lose them earlier if they often need a breathing tube.

Be sure to check your child's mouth regularly to see if they have lost any teeth. They might swallow a tooth and could choke on it.

Your child's adult teeth will probably come in at the usual time. Taking good care of them is very important to avoid cavities and other problems, and you might need to help your child do this. If your child does not want you to care for their teeth, you might need help from another adult. You may also need to be very patient with your child.

Your child's mouth will probably be too crowded with their adult teeth. These teeth are larger than baby teeth, and your child has a small mouth. Your child's dentist may want to take some of the adult teeth out to give the others more room.

Tests for dental problems

Your child's dentist will probably want to do **X-rays**, including when the first baby tooth comes in. X-rays show the

structure of your child's bones and teeth. This can be especially important when your child has CS. It can help your dentist watch out for future problems and make a plan for your child's treatment.

Treating dental problems

Your child's dentist will take care of problems when they happen. Getting regular checkups helps the dentist find problems early. This can help your child avoid pain and discomfort.



Medical Terms You Might Need

Enamel hypoplasia (uh-NAM-ul hi-po-PLAY-shuh): Thin tooth enamel. The enamel is the hard white material that covers each tooth.

How often should my child see a dentist?

Every six months, or more often if the dentist recommends it.

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Dermatology

Caring for Your Child's Skin

How CS Affects the Skin

Cockayne syndrome will probably make your child's skin very sensitive to light. The medical term is **photosensitivity (foe-toe-sen-suh-TIV-uh-tee)**. Almost all children with CS have this problem.^{1,2} You are likely to notice it when your child is a baby. They may get red blotches on both cheeks and across their nose in a pattern that looks like a butterfly.³ We've included here some other problems to watch for.

Alert

CS makes your child very sensitive to sun! Their skin is so sensitive that they can even get a sunburn through a car window.⁴ Use a sunscreen with SPF of at least 30 every day, even if it is not sunny. Dress your child in clothes with long sleeves. Have them wear hats and long pants, and keep them in the shade. Put on more sunscreen every two hours if your child is outside.

Other skin and hair changes can include:

- Dry skin²
- Thin, dry hair⁴
- Nail problems. For example, your child's nails might have an unusual shape⁴
- Cold, swollen hands or feet. The skin may look blue or purple.

Meet Your Specialist Dermatologist

A **dermatologist** is a specialist who cares for the skin and treats skin problems.

Dermatology is the branch of science and medicine that deals with studying, preventing, and treating skin problems.

Alert

Some children with CS cannot sweat when they get hot. They can get heat exhaustion or heat stroke. Keep your child cool by:

- Staying in the shade
- Putting a canopy on their wheelchair or stroller
- Using a spray bottle to spray them lightly with water if they get hot
- Using a damp towel to help them cool down
- Avoiding hot baths, showers, and hot tubs

Get them to a cool place if they seem hot. **Call 911 if they stop responding to you or their temperature is over 102 degrees Fahrenheit.**

Tests for Skin Problems

Your child's doctor may use a special light to check how their skin responds.⁴ Cockayne syndrome causes a very specific result from this test.¹ This skin test is actually one of the main ways to learn if your child has CS.

Treating Skin Problems

Talk with your child's dermatologist about the best treatment for skin problems caused by CS. Preventing sunburn is one of the most important ways to prevent skin problems.

How Often Should My Child See a Dermatologist?

Your child should see a dermatologist every year, or more often if the doctor recommends it.

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Medical Terms You Might Need

Photosensitivity (foe-toe-sen-suh-TIV-uh-tee): Being very sensitive to bright light. Children with CS often have this condition, including children with CS and xeroderma pigmentosum (XP). They can get a sunburn easily, even through a car window.

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Emergency Medicine

What to Know to Keep Your Child Safe

When Your Child With CS Has an Emergency

Your child may go to the emergency room for CS problems or another condition, such as an accident. You should know that children with CS need special emergency room care. Please share the information below with your child's ER doctor and team. It could save your child's life.

Meet Your Specialist Emergency Medicine Specialist

An **emergency medicine specialist**, or **ER doctor**, is trained to care for people with unexpected illness and injury.

Emergency medicine is the branch of science and medicine that involves unexpected illness and injury.

! Alerts

For the emergency department or urgent care center:

- **No Flagyl® (metronidazole).** This common antibiotic can make your child's liver stop working. This is very dangerous. Children with CS should never take it.¹
 - **Your child will have abnormal liver test results.** Some results may be two, three, or even four times higher than expected. This is normal with CS.²
 - **Your child needs less fluid in IVs.** Your child's kidneys cannot handle large amounts of fluid at once. They should get half the normal amount of IV fluids for their weight. This is because their kidneys do not work as well as normal kidneys.³ If your child gets too much fluid, they might need a substance called **albumin (al-BEW-min)** to correct this.
 - **Medicine dosing should go by weight, not age.** Children with CS are smaller than normal for their age. Give all medicines in the correct dose for the child's weight, not their age. Doctors should do this even if your child is older than 18 years. They may not need the adult dose because of their small body size, and it could be too much medicine for their system to handle.
-

- **Opioids and sedatives could harm your child.** A child with CS might respond more quickly or sleep more deeply than most people. This could be dangerous.
- **Many children with CS have small windpipes (airways).** This can make it more difficult to put in a breathing tube. If you see a new doctor or nurse, or go to the emergency room, let them know your child may have a small airway.
- **Be sure to know your child's regular body temperature.** When they get sick, children with CS can have a lower temperature, instead of a fever.

Older children with CS are more likely to have problems with blood sugar, including diabetes. If your child with CS is older than 16, they may need their blood sugar and insulin levels checked.⁴

Here are some things to watch for if your child has a feeding tube. Having the tube can hide some symptoms of being sick or affect them when they are sick, including:

- If the feeding tube is always somewhat uncomfortable, it can be difficult to tell if your child has new stomach pain from being sick.
- Your child might need their levels of **triglycerides (try-GLISS-err-ides)** checked. People with CS are at risk for high blood

Medical Terms You Might Need

Flagyl or metronidazole (met-row-NYE-da-zoll): A medication that can be dangerous for children with CS. Let the doctor, nurse or other health care provider know that your child has CS and should not take this medication.

Fluid overload: A condition that can happen to children with CS if they receive a normal IV dose of fluid. Their kidneys cannot handle a normal amount of fluid. Make sure the doctor, nurse or other health care provider knows your child has CS.

Triglycerides (try-GLISS-err-ides): Fat in the bloodstream. It is normal to have this, but it can be dangerous if the level goes too high.

sugar and high triglycerides, whether they have a feeding tube or not. But having a feeding tube is one thing that can affect blood sugar and triglycerides. Your child might need insulin to bring down their blood sugar. And if their triglycerides get too high, they are at risk for a painful condition called **pancreatitis (pan-cree-uh-TYE-tis)**. The health care team can do a blood test to check blood sugar and triglycerides.⁵⁻⁶

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Endocrinology

About Your Child's Glands and Hormones

How CS Affects the Glands and Hormone System

Cockayne syndrome may cause several problems with the endocrine system. This is a system of glands throughout the body. The substances that the glands make, called hormones, tell the body how to grow and develop. Problems with these glands can cause problems with growth, development, and general health. For example, a problem with the hormone called insulin leads to diabetes.

Problems for Children With CS

Your child may not have endocrine system problems.¹ If they do, the problems may include:²⁻³

- Sex organs that are smaller than normal
- No or later than normal sexual development (puberty)
- Testicles that do not “drop” in boys
- Irregular menstrual periods in girls
- Low thyroid hormone levels⁵
- Diabetes
- Weak bones that can break easily

One study showed that 13% of children over 16 with CS also had diabetes.⁵ This was slightly more than 1 in 10 children.

Children with CS may also have **insulin resistance**.⁶ This means the hormone insulin does not work well to

Meet Your Specialist Endocrinologist

An **endocrinologist** is a doctor who specializes in treating the glands that make your body's hormones.

Endocrinology is the branch of medicine and science that deals with these glands. These are just some of the glands in your body. These glands belong to a system called the endocrine system.

help their bodies process sugar. Their blood sugar level may stay too high, especially if they have a feeding tube.⁴⁻⁵ Constant high blood sugar damages the body's organs, such as the heart and eyes.

Tests for Endocrine Problems

- Blood and urine tests to check your child's hormone levels
- Tests to check the level of sugar in your child's blood
- Other tests related to diabetes and insulin resistance
- Bone density testing with a type of X-ray called **bone densitometry (den-sit-AH-met-ree)**

Treating Endocrine Problems

Your child's doctor can recommend specific treatment for individual problems. Your child should see a specialist for children with diabetes (pediatric endocrinologist) if they have diabetes. This specialist can also help with any

growth and development problems your child has.

If endocrine problems affect your child's bones, they may need to take vitamin D and calcium. Ask your child's doctor about this when you learn your child has CS. It can help prevent weak bones in the future.

How Often Should My Child See an Endocrinologist?

Your child's doctor will talk with you about how often to make appointments. If your child does not have growth problems or diabetes, they still need their blood sugar checked at least once a year.⁵ Their regular doctor may do this as part of a checkup.

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Medical Terms You Might Need

Bone densitometry (den-sit-AH-met-ree): A type of X-ray that measures how healthy the bones are. If your child is losing bone density, their bones could become weak and brittle. They are more likely to break a bone.

Endocrine (EN-do-crin) system: The system of glands that make hormones to control the body. Hormones are a type of chemical. They send signals to the brain and other parts of the body.

Puberty (PEW-bur-tee): The time when a child's body turns into an adult body.

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Ear, Nose, and Throat (Otolaryngology)

About Your Child's Ears, Nose, and Throat

How CS Affects the Ears, Nose, and Throat

Many children with Cockayne syndrome have hearing problems.¹ They may see an otolaryngologist (ear, nose, and throat specialist) if you or their pediatrician notice they do not hear well. Read the [Audiology](#) chapter in this manual to learn more about hearing problems with CS.

Ear infections are common in children, including children with CS. They happen when the area behind your child's eardrum gets infected. This can be very painful. Your child may have a fever, and you may see liquid draining from the ear.

Tests for Ear, Nose, and Throat Problems

Your child should see a specialist called an **audiologist (odd-ee-ALL-uh-jist)** if they have hearing problems. This is a health care professional who can test your child's hearing and find problems. They share the results with you and your child's doctor. Then, the doctor will talk with you to decide if your child needs hearing aids or other treatment.

Your child might also have an **MRI**. This test uses magnets to create pictures of the inside of the body. It can show the inside of the ear. Your ear, nose, and throat doctor may recommend an MRI to learn if the structure of your child's ear is causing hearing

Meet Your Specialist Ear, nose, and throat specialist (otolaryngologist)

An **ear, nose, and throat specialist** is referred to as an **otolaryngologist**, and sometimes called an ENT. This is a doctor who cares for diseases and conditions of the ears, nose, and throat, sometimes including swallowing and voice problems as well as the teeth and mouth. Otolaryngologists can perform surgical procedures, such as placing cochlear implants.

Otolaryngology is the branch of science and medicine that deals with studying, preventing, and treating diseases and problems of the ears, nose, and throat.

problems. It can also help prepare for your child to get an implant that helps them hear (cochlear implant). You can read more about this implant below.

Treating Ear, Nose, and Throat Problems

Ear infections need treatment to avoid serious problems, including a broken (perforated) ear drum and hearing loss. If your child has ear infections often, your doctor might talk with you about putting tubes in your child's ears. This can help prevent more infections.

Children with CS may need hearing aids. If hearing aids do not help,

an implant called a cochlear implant (COCK-lee-ur implant) may help them hear.² Check the [Audiology](#) section of this manual for more information.

How Often Should My Child See an Ear, Nose, and Throat Specialist?

Your child should see an otolaryngologist every year to have their ears and hearing checked.³ The doctor will probably also check their nose and throat, and check for swallowing problems. Your child may need to see the otolaryngologist more often if your child has hearing aids. These can cause wax to build up in the ear. Your child can have earwax removed at their appointment.

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Medical Terms You Might Need

Audiologist (odd-ee-OLL-uh-jist): A medical professional with a doctoral degree in audiology who tests how well someone can hear. They can recommend hearing aids and other things to help with hearing loss. They work with ear, nose, and throat doctors.

Cochlear implant (COCK-lee-ur implant): A small device a doctor can put inside a child's ear to help them hear.

Otolaryngologist (oh-toe-larr-in-GOLL-uh-jist): A doctor who specializes in treating ear, nose, and throat conditions. Some of them also specialize in treating children. This type of doctor is called a pediatric otolaryngologist. These doctors are also known as ear, nose, and throat (ENT) specialists.

Digestive System and Liver

Food, Nutrition, and Metabolism With CS

How CS Affects the Digestive System and Liver

Cockayne syndrome can cause digestive system and liver problems.¹⁻³ For example, your child's digestive system might not move food and liquids through their body normally. It can be slower than normal or not move much at all. The medical term is **gut dysmotility (diss-mow-TILL-uh-tee)**. Your child might also have problems swallowing food or liquids.

Additional problems with the digestive system and liver can include:

- Fast metabolism¹
- Low weight or losing weight³
- Not getting enough nutrition from foods and drinks³
- Not eating enough
- Enlarged liver³ or spleen
- Constipation and diarrhea²
- Itching—this may be related to liver problems or something else

Alert

Your child should not receive the medication called Flagyl (metronidazole).⁶ It can cause serious problems, including liver failure.

Meet Your Specialists Gastroenterologist Hepatologist

A **gastroenterologist** is a doctor who specializes in treating the digestive system. Some of them also treat the liver. A

hepatologist is a doctor who specializes in treating the liver.

Gastroenterology is the branch of medicine and science that deals with the organs and diseases of the digestive system.

Hepatology does the same for the liver.

Tests for Digestive and Liver Problems

Your child might have tests to check their acid reflux and find the right treatment. These may include:

- A video examination with a tiny camera. The medical term for this is **endoscopy (en-DAW-suh-pee)**.
- X-rays with a substance called **barium (BEAR-ee-um)**. This helps your child's doctor see the digestive system and watch how well your child swallows.
- A test to learn if food or liquid is getting into the lungs
- A test to look for acid in the throat and stomach

Your child also needs tests to check their blood sugar and how well their liver is working. A doctor who specializes in

the liver should check the results of liver tests carefully. Children with Cockayne syndrome have unusual results from these tests. Talk with a liver specialist (hepatologist) first if you are told your child needs a liver biopsy. A liver biopsy is not typically recommended for children with CS.

How Often Should My Child See a Specialist?

That depends on your child's condition and any problems they have. Every child with Cockayne syndrome is different. If you are concerned about something, talk with your doctor or someone else on your child's health care team. You can also talk with a CS support group for suggestions and encouragement in getting care for your child.

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Medical Terms You Might Need

Acid reflux: Also called gastrointestinal (gas-trow-in-TEST-tuh-null) reflux disease, GERD (gurd), reflux, and heartburn. This condition happens when acid from the stomach comes back up into the throat. It causes pain and discomfort. You might have had it yourself, and it is common in children with CS.

Barium (BEAR-ee-um): A thick liquid your child might drink before having X-rays. Barium can help your child's doctor see more in the X-ray image.

Endoscopy (en-DAW-scuH-pee): A test that uses a thin tube with a camera and light to examine the throat and upper part of the digestive system, such as the stomach.

Gut dysmotility (diss-mow-TILL-uh-tee): A condition where your child's digestive system does not move food and liquids through the body normally.

Swallow study: A test that shows what happens in the throat and esophagus (ee-SOF-uh-gus) when your child swallows.

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Genetics

How CS Happens

How Genes Cause CS

People with Cockayne syndrome have changes, or **mutations (mew-TAY-shuns)**, on specific genes. A gene gives your body instructions on how to grow and develop, and a mutation is like a mistake in the gene. Genes with mistakes can produce different results than normal, and this is true with Cockayne syndrome.

About 8 of 10 children with Cockayne syndrome have a mutation on a gene called *ERCC8*. About 2 in 10 have a mutation on the gene *ERCC6*.¹⁻² Some other mutations are related to CS, but those are the main ones.³⁻⁴

A child develops Cockayne syndrome if they get a copy of a CS mutation gene from each parent. If they get it from just one parent, they will not develop CS. Both parents have to pass the mutation to their child. The scientific term for this type of inheritance is **autosomal recessive**.⁵

Tests for CS Genes

There are several different tests to look for gene mutations that cause CS. They include:⁶

- A test that looks for changes (mutations) in several different genes. The medical term for this is a **multi-gene panel**.
- A test to check how well your genetic information fixes mistakes when they happen. With CS, the genes do not fix mistakes.

Meet Your Specialist

Geneticist

Genetic counselor

A **geneticist** is a doctor who specializes in how genes affect your health. A **genetic counselor** is a health care professional who is trained to help families understand their genes and the conditions those genes cause.

Genetics is the branch of medicine and science that deals with genes.

If CS Runs in Your Family

A genetic counselor can talk with you about the CS mutations in your family. This may include talking about:⁶

- Which mutation is present
- How it was passed to your child
- If other children have a risk of developing CS
- Whether you want to have more children in the future

A genetic counselor can also talk with you about the results of tests for CS when you are pregnant. You may want to get tested if CS runs in your family.

How Often Should My Child or I See a Geneticist or Genetic Counselor?

You and your child will probably see a geneticist when you learn your child has Cockayne syndrome. You might see them a few more times as you learn about CS and what it will mean

for your child. The geneticist might be a part of your child's care team in the future, or they might not.

Your genetic counselor will probably become an important part of your child's health care team. They can give you information and talk with you about each family member's individual risk of passing on CS.

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Medical Terms You Might Need

Gene: A small piece of the information that tells your body how to grow and develop. People with Cockayne syndrome have changes on specific genes, so their bodies grow and develop differently from most people's.

Geneticist: A doctor who specializes in how genes work. They might be part of your child's CS team.

Genetic counselor: A health care professional who talks with people and their families about genes and the conditions that gene changes can cause. Your genetic counselor can talk with you about testing, which gene changes your child has, and more.

Multi-gene panel: A test that looks for changes in several genes, not just one at a time.

Mutation: A change or mistake in a gene. People with CS have mutations, or changes, on certain genes.

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Kidneys

Caring for Your Child's Kidneys

How CS Affects the Kidneys

Many children with Cockayne syndrome have problems with their kidneys.¹ These are the organs that filter waste out of the blood. The kidney problems may develop because CS causes the body to age quickly. Many kidney problems in Cockayne syndrome are similar to those that adults develop in old age.¹⁻³

Kidney problems can include:¹⁻³

- Losing protein in the urine (pee). This can lead to a condition called **nephrotic syndrome (neff-ROT-ick syndrome)**
- Too much of a substance called uric acid in the blood
- Kidneys that are damaged
- Having just one kidney, or having two kidneys, one of which is very small or does not work well
- Infections in the urine system (urinary tract), which sometimes happen again and again
- High blood pressure

Alert

If your child needs an IV, be sure the nurse knows that your child's CS affects how much fluid their body can handle. Volumes of IV fluids that other children can handle may overload your child's kidneys. This can be dangerous.⁴

You can find more information in the [Emergency Medicine](#) section.

Meet Your Specialists

Nephrologist Urologist

A **nephrologist** is a doctor who specializes in treating the kidneys. A **urologist** is a surgeon who treats urinary problems.

Nephrology is the branch of medicine and science that deals with research and treatment on the kidneys.

Tests for Kidney Problems

Your child should have blood and urine tests at least once a year. The results tell your child's doctor how well their kidneys are working. If the results are abnormal, your child might need tests more often or more detailed evaluations.⁵

Your doctor and health care team should know:

- Comparing your child's test results to their own results from an earlier test can be more helpful than comparing them to results for children without CS.
- Children with CS may have high blood pressure. Your child needs their blood pressure checked often. They might need to take medication to keep blood pressure from getting too high. This needs to be monitored regularly. If hypertension is an issue, medication may be

- necessary. Be sure the health care team uses a child-sized blood pressure cuff to check, even if your child is older.
- Your child probably needs urine tests each time they go to the kidney specialist. They may also need an ultrasound of their kidneys from time to time. This test uses sound waves to create pictures of the inside of the body.

Treating Kidney Problems

Your child might have one or more of the following treatments:¹

- Medications to lower blood pressure
- Medications to slow down kidney disease
- Medications to control the amount of a substance called uric acid in the blood

How Often Should My Child See a Specialist?

That depends on your child's condition and any problems they have. However, it is a good idea to have kidney tests at least once a year. Your child may need to see a kidney specialist periodically.

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Medical Terms You Might Need

Fluid overload: A condition that can happen to children with CS if they receive a dose of fluid that is normal for children without CS. The kidneys of children with CS cannot handle a normal amount of fluid. Make sure the doctor, nurse or other health care provider knows your child has CS.

Hypertension: High blood pressure.

Nephrotic syndrome (neff-ROT-ick syndrome): A kidney disease that happens when the kidneys are damaged and do not filter waste properly.

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Nervous System and Brain (Neurology)

Helping Your Child With the Effects of CS

How CS Affects the Brain and Nervous System

CS affects the brain, which may shrink or develop abnormalities. CS also affects the way your child's nerves function. The nerves work less and less normally over time. Your child could also have a stroke, or "brain attack."

Any of these changes can affect your child's ability to move, think, speak, and do regular activities. Your child's specific problems depend on which nerves or areas of the brain are most affected by CS.

Problems can include:¹⁻⁶

- Learning to talk or do certain motor activities later than other children
- Loss of mobility at certain joints, caused by tight tendons and muscles (contractures)
- Tremors (shaking)
- Difficulty walking, standing, or doing other physical activities
- Seizures²
- Bladder control problems³

Other Nervous System Problems

Other problems include a condition called **peripheral neuropathy (purr-IF-er-ul nur-AW-puth-ee)**, meaning irritation or injury to the nerves, which can cause numbness and tingling in the hands or feet.⁴ CS can also affect the parts of the nervous system that work on their own to control breathing and other body functions. The medi-

Meet Your Specialist Neurologist

A **neurologist** is a doctor who specializes in finding and treating problems with the brain, spinal cord, and nerves.

Neurology is the branch of science and medicine that involves studying and treating these areas of the body.

cal term for this is **autonomic nervous system dysfunction (aw-to-NOM-ick nervous system dis-FUNK-shun)**.

Finally, CS can cause sleep problems. These include leg movements (restless legs syndrome) and pauses in breathing during sleep (sleep apnea).

Tests for Nervous System and Brain Problems

Tests for specific problems may include:

- **EEG:** If your child has seizures or spells concerning for seizures, they might have this test to measure brain activity and detect signs of seizures.
- **EMG** or **nerve conduction test:** These tests measure muscle and nerve activity.
- **CT** or **MRI:** These tests create pictures of the inside of the body. Your child's doctor might suggest one or both tests to look at your child's brain or other areas.

- **Sleep study:** This test can help find the cause of sleep problems. Your child spends the night in a “sleep lab” where the health care team can watch them and measure their activity during sleep. This test can help diagnose restless legs syndrome and sleep apnea.

Treating Nervous System and Brain Problems

Treatments for nervous system and brain problems include the following.

- A medication called carbidopa-levodopa for shaking (tremors).⁵ This may also help your child’s muscles work better.
- Medications for seizures. Talk with your child’s doctor about which ones are the safest for your child’s liver.
- Medication for numbness and tingling in the hands and feet.
- An operation called deep brain stimulation. This may help with tremors, movement problems, and other concerns.

Stretching and exercise are important to help your child stay as flexible and strong as possible.

If your child goes to school, they will probably need a specialized learning plan called an **individualized education program**, or **IEP**. Talk with the social worker on your child’s health care team or your child’s teacher or principal about an IEP.

How Often Should My Child See a Neurologist?

Your child should see a neurologist as soon as you learn they have Cockayne syndrome, if possible. The neurologist

Medical Terms You Might Need

Autonomic nervous system dysfunction (aw-to-NOM-ick nervous system dis-FUNK-shun): Problems with the nerves that usually work on their own, without conscious control. These nerves control body functions you do not need to think about, such as breathing.

Peripheral neuropathy (purr-IF-er-ul nur-AW-puth-ee): A nerve condition that causes numbness and tingling in the hands or feet. It can happen when the nerves are damaged, such as by disease or medications.

will check for nervous system problems. They may recommend that your child have a brain MRI if they have not had one already. After that, your child should see the neurologist at least once a year. They may need to go more often if they have new problems.

If your child takes carbidopa-levodopa or other medications, they need to see a neurologist at least twice a year. The doctor will check how well the medication is working and look for any side effects.

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Nutrition

How CS Affects Nutrition

Cockayne syndrome can cause feeding and **nutrition (noo-TRI-shun)** problems. Many children with CS have the condition called acid reflux or acid indigestion. The medical term for this is **gastrointestinal reflux disease** or **GERD**. The discomfort of GERD can make it difficult for your child to eat enough. Your child might sometimes not want to eat. This might be related to GERD, another CS symptom, or something else.

Problems with eating can lead to poor growth or weight loss. Other nutrition problems can include:

- Fast metabolism¹
- Constipation and diarrhea²
- Low weight or losing weight³
- Not getting enough nutrition from foods and drinks³

Treating Nutrition Problems

Your child might have the following treatments:¹

- A nutritionist will help to develop a feeding plan for your child that has the right calories for good growth.
- Treatment to help with eating or swallowing problems, from a specialist in feeding therapy.
- Some children will need a feeding tube to help them get enough nutrition and fluids. One type of tube goes through the nose into the stomach. This is called a **nasogastric tube (nay-zo-GASS-trick tube)** or **NGT (en-gee-tee)**.

Meet Your Specialists

Nutrition specialist Feeding specialist

A **nutrition specialist** is also called a **nutritionist (noo-TRI-shun-ist)** or **dietitian (die-uh-TISH-un)**. They are trained to help people know what to eat to stay as healthy as possible.

A **feeding specialist** helps people who have problems chewing, eating, and swallowing food and drinks.

Another type goes into the stomach through an opening in the belly. This is called a **gastrosomy (gass-TRAW-stuh-mee)** tube or **G-tube (gee tube)**.

- Diaper rash treatment and other skin care to prevent problems from diarrhea.
- Medications to help with reflux or constipation.
- An operation to help with GERD. This is called fundoplication.

Other Important Things to Know

- Your child might lose the fat layer under their skin over time. This could cause their feeding tube to leak. If that happens, they might not get all the nutrients they need. Liquid leaking from the tube could also irritate your child's skin. You may want

- to ask your doctor or nurse to check how well the feeding tube fits over time.²
- Losing fat under the skin is also a sign your child is not getting enough nutrition.²
 - Children with CS are at risk for blood sugar problems. The [Endocrinology](#) chapter has more information about this. Talk with your doctor about checking your child's blood sugar regularly.⁵
 - Children with CS do not usually grow in line with child development growth charts. Your child's doctor will measure BMI (body mass index) to make sure that your child is growing in a way that is right for them.

Alert

Children with CS should not take in too much food or liquid at once. This includes regular food, tube feeding, and fluid from an IV.⁴ With food, they need small amounts more often rather than large amounts at once.

Alert

Your child should not receive the medication called Flagyl (metronidazole).⁶ It can cause serious problems, including liver failure.

Medical Terms You Might Need

Acid reflux: Also called gastrointestinal (gas-trow-in-TEST-tuh-null) reflux disease, GERD (gurd), reflux, and heartburn. This condition happens when acid from the stomach comes back up into the throat. It causes pain and discomfort. You might have had it yourself, and it is common in children with CS.

Gastrostomy tube (gass-TRAW-stuh-mee tube): Also called a G tube ("gee tube") or written as GT. This is a small tube a doctor puts in your child's stomach through an opening in their belly. It allows your child to get nutrition from outside their body if they cannot eat or swallow well.

Nasogastric tube ("nay-zo-GASS-trick" tube): Also called an NGT (en-gee-tee). This is a small tube a doctor puts into the nose. It carries nutrition from outside the body into the stomach. Your child might have one if they cannot eat or swallow well.

How Often Should My Child See a Specialist?

That depends on your child's condition and any problems they have. Every child with Cockayne syndrome is different. If you are concerned about something, talk with your doctor or someone else on your child's health care team. You can also talk with a CS support group for suggestions and encouragement in getting care for your child.

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Eyes and Vision (Ophthalmology)

Making the Most of Your Child's Sight

How CS Can Affect Your Child's Eyes

Many children with Cockayne syndrome have eye problems. These often include **cataracts (CAT-uh-rackts)**, a condition where the lens inside your child's eye is cloudy instead of clear.¹

Other eye problems can include:¹

- Problems with the cornea, the clear outer covering of the front of the eye
- Disease in the retina, the area where the eye forms images²
- Dry eyes from not making enough tears or sleeping with the eyes open³
- Problems with the eye muscles or abnormal eye movements³
- Farsightedness⁴
- Eyes that are small or look different from other children's¹
- An eye disease called **glaucoma (glow-COE-muh, with "ow" pronounced like the word "ow")**

CS can also affect the nerve that connects the eye to the brain, called the optic nerve.

Special Situations

- **Sunburn of the eye:** Your child is at high risk for sunburn, and this includes their eyes.
- **Sleeping with eyes open:** If your child is starting to sleep with their eyes open or incompletely closed, you can put drops or gel in their eyes. This keeps them from getting too dry. This com-

Meet Your Specialist Ophthalmologist

An **ophthalmologist** is a doctor who specializes in finding and treating eye problems.

Ophthalmology is the branch of science and medicine that involves studying and treating eye diseases and injuries.

monly happens as the fat behind the eye breaks down and the eyes sink beyond the eyelid covering, so the eyes dry out.

- **Tunnel vision and night blindness:** If your child has difficulty seeing at night or seeing things that are off to the side (peripheral vision problems), talk with your doctor. These can be signs of a condition called **retinitis pigmentosa (reh-tin-EYE-tus pig-men-TOE-suh)**, or RP. This condition is a complication of Cockayne syndrome.² If your child has RP, doctors may check for Cockayne syndrome if they do not already know your child has it.

Testing for Eye and Vision Problems

The ophthalmologist (eye doctor) will check your child's ability to see and look for eye problems. This may include testing your child's retina to look for problems. Some retina tests are done with sedation, where your child gets

medication to help them relax and feel sleepy or even sleep through the test. This allows the doctor to examine the retina closely.

Treating Eye and Vision Problems

- An ophthalmologist might do surgery to remove cataracts.
- Your child might need glasses, contact lenses or both.³
- Have your child wear sunglasses or other eye protection in bright light. They should wear eye protection even when the weather is cold or they are riding in a car.⁵
- Talk with your child's eye doctor (ophthalmologist) about the best drops or gel to keep your child's eyes from getting dry. You can buy some kinds without a prescription. If they do not work well, the doctor can prescribe some that may be more effective.

How Often Should My Child See an Ophthalmologist?

Your child should see an ophthalmologist as soon as you learn they have Cockayne syndrome, if possible. The doctor can check your child's eyes to learn if they have cataracts or any other problems. After that, your child should see an ophthalmologist at least once a year. Their annual examination should include checking the retina for changes.⁴

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2. Hamel C. Retinitis pigmentosa. *Orphanet J Rare Dis*. 2006;1:40.

Medical Terms You Might Need

Cataracts (CAT-uh-rackts): A condition where the lens inside the eye becomes cloudy and difficult to see through.

Cornea (CORE-nee-uh): The clear tissue that covers the front of the eye.

Glaucoma (glow-COE-muh, with the "ow" said like the word "Ow): A disease that makes pressure build up inside the eye.

Optic nerve: The nerve that connects the eye with the brain.

Retina (RET-uh-nuh): The tissue at the back of the eye where images form.

Retinitis pigmentosa (reh-tin-EYE-tus pig-men-TOE-suh): An eye disease that makes the retina slowly break down over time. It can lead to blindness.

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Bones, Nerves, and Muscles

Helping Your Child Move and Do Activities

How CS Affects the Nerves, Muscles, and Movements

Cockayne syndrome causes the nerves to lose the ability to function. Their function usually gets worse as time goes on, and affects your child's ability to move, speak, eat, and do other activities. Specific problems depend on which nerves and parts of the brain CS is affecting the most.

Problems moving and speaking can include:^{1,2}

- Difficult or unsteady walking
- Tremor (shaking)
- Slow and awkward-sounding speech
- Difficulty speaking and swallowing

Your child might also have some of the following conditions:

- A curved back (scoliosis or kyphosis)
- Stiff or bent arms or legs caused by tight tendons and muscles (contractures)
- Joints that get out of position (dislocated joints)
- Small or weak muscles

Tests for Nerve and Muscle Problems

Your child may have tests for specific problems. This may include an EMG or nerve conduction test to measure muscle and nerve activity.

Meet Your Specialists Orthopedic surgeon Physiatrist

An **orthopedic surgeon** is a doctor who specializes in treating problems with the bones and joints. A **physiatrist** is a doctor who helps people go back to normal activities after an illness or injury. This is called rehabilitation. Some physiatrists specialize in disorders of nerves and muscles.

Orthopedics is the branch of medicine and science that deals with studying and treating the bones and joints. **Physiatry** deals with nerve and muscle problems related to normal movement and activity.

Treating Muscle and Nerve Problems

Treatments for muscle and nervous system problems include the following:

- Medication called carbidopa-levodopa for shaking (tremors), which may also help your child's muscles work better³
- Braces (orthoses) to help stabilize joints and stretch out contractures
- Physical therapy for numbness, tingling, and movement problems.⁴ Your child might use equipment such as a wheelchair, special stroller or seat, or another item⁵

Tips From a CS Parent

A home stretching program is important!

Even if your child cannot walk, they need to keep their muscles as relaxed and flexible as possible. This will help with symptoms.

It will also help them stay as physically active as they can.

Plan some time to stretch with your child every day, if possible. Your physical therapist can help you create a stretching plan.

- Occupational therapy to help your child eat, feed themselves, get dressed, and do other everyday activities
- Speech therapy to help with speaking and swallowing problems

Treating Contractures

Things that can help with tight, short tendons and tight muscles include:

- Regular stretching. All children with CS should stretch every day if possible. Ask your child's physical therapist about stretches that can help
- Wearing a cast or splint
- Taking medication, by mouth or in a shot (injection)
- Surgery

How Often Should My Child See an Orthopedist or Physiatrist?

Your child should see these specialists when they need them. Your regular doctor may ask you to see them for specific problems. After you start

Medical Terms You Might Need

Carbidopa-levodopa: A medication that can help with shaking (tremors) and movement problems in children with CS. Doctors also use it to treat other diseases that cause movement problems and shaking, such as Parkinson disease.

Contractures (con-TRACT-shurs): Tightness in the tendons, muscles, joints and other tissues of the body. Your child's body might look bent. They might have difficulty moving or walking because of tightness.

Kyphosis (ky-FOE-sus): A problem with the spine. It makes the back bend upward, or "hunch." Your child's back might look rounded instead of straight up and down.

Scoliosis (sko-lee-OH-sus): A problem with the spine. It makes the spine curve sideways at certain points instead of going straight up and down. If your child has scoliosis, they might have one shoulder that looks higher than the other. Or the child might look normal but have back pain and tightness.

treatment, the specialist will talk with you about how often they would like to check your child. If they do not mention this recommendation, you should ask them.

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1. Wilson BT, Stark Z, Sutton RE, et al. The Cockayne Syndrome Natural History (CoSyNH) study: clinical findings in 102

- individuals and recommendations for care. *Genet Med*. 2016;18(5):483-493.
2. Karikkineth AC, Scheibye-Knudsen M, Fivenson E, Croteau DL, Bohr VA. Cockayne syndrome: clinical features, model systems and pathways. *Ageing Res Rev*. 2017;33:3-17.
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 4. Rapin I, Weidenheim K, Lindenbaum Y, et al. Cockayne syndrome in adults: review with clinical and pathologic study of a new case. *J Child Neurol*. 2006;21(11):991-1006.
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Lungs (Pulmonology)

Helping Your Child Breathe Better

How CS Can Affect Your Child's Lung Health

Some children with Cockayne syndrome develop infections that affect their breathing. These may happen when your child accidentally breathes food or fluid into the lungs when they swallow.¹ The medical term for this problem is **aspiration (ass-puh-RAY-shun)**. If this happens, your child may need to see a doctor who specializes in lung problems, called a pulmonologist.

Other lung problems related to CS can include:¹

- Asthma
- Stiffness or tightness of the lung tissue or chest, which can make it hard to breathe normally²

Testing for Lung Problems

Your child's doctor may want to do tests to learn how healthy your child's lungs are. Tests of how they swallow may be especially important. Lung tests can include:

- **Bronchoscopy (bron-KOSS-cup-ee)**: This test looks inside your child's lungs with a thin tube that goes in through their nose. Your child will have medication to help them relax and feel sleepy or even sleep through the test. The doctor can also take a small sample of lung tissue during the test, if needed.

Meet Your Specialist Pulmonologist

A **pulmonologist** is a doctor who specializes in finding and treating lung and breathing problems.

Pulmonology is the branch of science and medicine that involves studying and treating conditions that affect the lungs and breathing.

- **High-altitude test**: Your child might have trouble breathing if they travel on a plane. Your doctor can test how well your child breathes when there is less oxygen in the air around them.
- **Sleep study**: To check for breathing problems during sleep, including sleep apnea. This condition causes breathing to stop for short periods during sleep.
- **Swallowing study**: To check the way your child swallows. This helps the doctor make sure your child is safe when swallowing their food and drinks.

Treating Lung and Breathing Problems

Treatment depends on the specific problem. For example, if your child has asthma, the doctor may recommend breathing treatments, an inhaler for emergencies, or daily medication.

If your child tends to get lung infections (pneumonia), the doctor will look for the cause, including whether your child is getting food or water in the lungs.

Wearing a small device during sleep can help your child with sleep apnea. This device is called a **BiPap (BYE-pap)**.³ It helps your child keep breathing in and out normally, even when they are asleep.

How Often Should My Child See a Pulmonologist?

That depends on their lung health and any breathing problems. If they do not have breathing problems, they may not need to see a pulmonologist at all. If they have asthma or another long-lasting lung problem, they may need to see the pulmonologist every few weeks or months. Alternatively, you might see a technician to have breathing tests.

References

1. Wilson BT, Stark Z, Sutton RE, et al. The Cockayne Syndrome Natural History (CoSyNH) study: clinical findings in 102 individuals and recommendations for care. *Genet Med*. 2016;18(5):483-493.
2. Johns Hopkins Medicine. Restrictive lung disease. Accessed July 1, 2020. [### **Medical Terms You Might Need**](https://www.hop-</div><div data-bbox=)

Aspiration (ass-puh-RAY-shun):

Breathing food or fluid into the lungs. This can damage your child's lungs and raise the risk of getting a lung infection, such as pneumonia.

BiPap (BYE-pap): A small device your child wears when they sleep. It helps them keep breathing normally.

Bronchoscopy (bron-KOSS-cup-ee): A way to look inside the lungs. The doctor uses a thin tube that goes in through the nose.

[kinsmedicine.org/health/conditions-and-diseases/restrictive-lung-disease](https://www.kinsmedicine.org/health/conditions-and-diseases/restrictive-lung-disease).

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Research and New Treatments

Learning More About CS Should You or Your Child Join a Research Study?

Research studies can give you or your child a chance to try a new treatment that is not yet available to the public. Researchers need volunteers to learn how well these new treatments work. There are also studies to learn about genetics, which is how diseases like Cockayne syndrome are passed from parents to children.

Cockayne syndrome is a rare disease, so there are fewer studies than for some other diseases. However, researchers are studying several topics. These include:

- How CS affects the way the body uses energy (metabolism)¹
- Skin problems that can happen in children with CS (xeroderma pigmentosum)²
- How CS develops over time³
- Eye disease in CS (retinitis pigmentosa)⁴
- Possible medications to treat CS⁵

Finding a Research Study for CS

Ask your child's doctor or someone else on the health care team about finding a research study. They may be able to help you find one. You can also contact an organization that supports children and families with Cockayne syndrome. The next section lists several of these.

Meet Your Specialist Researcher

A **researcher** is a doctor, nurse, or other health care or science professional who specializes in learning how to prevent and treat diseases and keep people healthier.

Researchers follow strict rules to make sure the people who take part in research studies are safe and receive the treatment they need.

Being in a research study can give you a chance to try a treatment that is not available to the public yet. The treatment may help you, or it may help others later.

Medical Term You Might Need

Clinical trial: Another name for a research study. It is used for studies that include people. The people in a clinical trial are all volunteers.

Support for Children and Families With CS

- **National Initiative for Cockayne Syndrome:** <http://nics-online.org/>
- **National Institutes of Health Genetic and Rare Disease Information Center:** <https://rarediseases.info.nih.gov/diseases/6122/cockayne-syndrome>
- **Share and Care Cockayne Syndrome Network:** <http://cockaynesyndrome.org/>
- **Amy and Friends:** <https://www.amyandfriends.org/>
- **National Organization for Rare Diseases:** <https://rarediseases.org/rare-diseases/cockayne-syndrome/>
- **Orphanet:** <https://www.orpha.net/consor/cgi-bin/index.php>

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Glossary

Medical Terms You Might Need

Acid reflux: Also called gastrointestinal (gas-trow-in-TEST-tuh-null) reflux disease, GERD (gurd), reflux, and heartburn. This condition happens when acid from the stomach comes back up into the throat. It causes pain and discomfort. You might have had it yourself, and it is common in children with CS.

Aspiration (ass-puh-RAY-shun): Breathing food or fluid into the lungs. This can damage your child's lungs and raise the risk of getting a lung infection, such as pneumonia.

Atherosclerosis (ath-uh-row-scluh-ROW-sis): The medical term for hardening of the arteries. Arteries are the blood vessels that carry blood from the heart to the rest of the body. Old age and diseases such as CS can make them stiff and hard, and this can lead to problems.

Audiologist (odd-ee-OLL-uh-jist): A health care provider who tests how well someone can hear. They can recommend hearing aids and other things to help with hearing loss. They work with ear, nose, and throat doctors.

Auditory brainstem response or brainstem auditory evoked response (odd-it-ORRY brainstem response or EE-voak-ed response): Also called ABR or BAER. The audiologist tests how the hearing part of your child's brain responds to

sound. Your child might need medication for this test to make them relaxed and sleepy. This is because they need to be very still during the test.

Autonomic nervous system dysfunction (aw-to-NOM-ick nervous system dis-FUNK-shun): Problems with the nerves that usually work on their own, without conscious control. These nerves control body functions you do not need to think about, such as breathing.

Barium (BEAR-ee-um): A thick liquid your child might drink before having X-rays. Barium can help your child's doctor see more in the X-ray image.

BiPap (BYE-pap): A small device your child wears when they sleep. It helps them keep breathing normally.

Bone densitometry (den-sit-AH-met-ree): A type of X-ray that measures how healthy the bones are. If your child is losing bone, their bones could become weak and brittle. They are more likely to break a bone.

Bronchoscopy (bron-KOSS-cup-ee): A way to look inside the lungs. The doctor uses a thin tube that goes in through the nose.

Carbidopa-levodopa: A medication that can help with shaking (tremors) and movement problems in children with CS. Doctors also use it to treat other

diseases that cause movement problems and shaking, such as Parkinson disease.

Cataracts (CAT-uh-rackts): A condition where the lens of the eye becomes cloudy and difficult to see through.

Clinical trial: Another name for a research study. It is used for studies that include people. The people in a clinical trial are all volunteers.

Cochlear implant (COCK-lee-ur implant): A small device a doctor can put inside a child's ear to help them hear.

Contractures (con-TRACKT-shurs): Tightness in the tendons, muscles, joints and other tissues of the body. Your child's body might look bent. They might have difficulty moving or walking because of tightness.

Cornea (CORE-nee-uh): The clear tissue that covers the front of the eye.

Echocardiogram (eck-oh-CAR-dee-oh-gram): Also called an echo. This test uses sound waves to create a picture of the heart.

Electrocardiogram (ee-leck-tro-CAR-dee-oh-gram): Also called an EKG or ECG. This test measures electrical activity in your child's heart.

Enamel hypoplasia (uh-NAM-ul hi-PLAY-shuh): Thin tooth enamel. The enamel is the hard white material that covers each tooth.

Endocrine (EN-do-crin) system: The system of glands that make hormones to control the body. Hormones are a type of chemical. They send signals to the brain and other parts of the body.

Endoscopy (en-DAW-scu-h-pee): A test that uses a thin tube with a camera and light to examine the throat and upper part of the digestive system, such as the stomach.

Flagyl or metronidazole (met-row-NYE-dazoll): A medication that can be dangerous for children with CS. Let the doctor, nurse, or other health care provider know that your child has CS and should not take this medication.

Fluid overload: A condition that can happen to children with CS if they receive a normal IV dose of fluid. Their kidneys cannot handle a normal amount of fluid. Make sure the doctor, nurse or other health care provider knows your child has CS.

Gastrostomy tube (gass-TRAW-stuh-mee tube): Also called a G tube ("gee tube") or written as GT. This is a small tube a doctor puts in your child's stomach through an opening in their belly. It allows your child to get nutrition from outside their body if they cannot eat or swallow well.

Gene: A small piece of the information that tells your body how to grow and develop. People with Cockayne syndrome have changes on specific genes, so their bodies grow and develop differently from most people's. A condition related to changes on specific genes is called a genetic (juh-NET-ick) condition.

Geneticist: A doctor who specializes in how genes work. They might be part of your child's CS team.

Genetic counselor: A health care professional who talks with people and their

families about genes and the conditions that gene changes can cause. Your genetic counselor can talk with you about testing, which gene changes your child has, and more.

Glaucoma (glow-COE-muh, with the “ow” said like the word “ow”): A disease that makes pressure build up inside the eye.

Gut dysmotility (diss-mow-TILL-uh-tee): A condition where your child’s digestive system does not move food and liquids through the body normally.

Hypertension: High blood pressure.

Kyphosis (ky-FOE-sus): A problem with the spine. It makes the back bend upward, or “hunch.” Your child’s back might look rounded instead of straight up and down.

Metronidazole (met-row-NYE-da-zoll): Also called Flagyl. A medication that can be dangerous for children with CS. Let the doctor, nurse, or other health care provider know that your child has CS and should not take this medication.

Multi-gene panel: A test that looks for changes in several genes, not just one at a time.

Mutation: A change or mistake in a gene. People with CS have mutations, or changes, on certain genes.

Nasogastric tube (nay-zo-GASS-trick tube) or NGT (en-gee-tee): A small tube a doctor puts into the nose. It carries nutrition from outside the body into the stomach. Your child might have one if they cannot eat or swallow well.

Nephrotic syndrome (neff-ROT-ick syndrome): A kidney disease that happens when the kidneys are damaged and do not filter waste properly.

Optic nerve: The nerve that connects the eye with the brain.

Optoacoustic test: Also called an OAE test. The audiologist tests the nerves inside your child’s ear to see how well they respond.

Otolaryngologist (oh-toe-larr-in-GOLL-uh-jist): A doctor who specializes in treating ear, nose, and throat conditions. Some of them also specialize in treating children. This type of doctor is called a pediatric otolaryngologist.

Peripheral neuropathy (purr-IF-er-ul nur-AW-puth-ee): A nerve condition that causes numbness and tingling in the hands or feet. It can happen when the nerves are damaged, such as by disease or medications.

Photosensitivity (foe-toe-sen-suh-TIV-uh-tee): Being very sensitive to bright light. Children with CS often have this condition. They can get a sunburn easily, even through a car window.

Puberty (PEW-bur-tee): The time when a child’s body turns into an adult body.

Retina (RET-uh-nuh): The tissue at the back of the eye where images form.

Retinitis pigmentosa (reh-tin-EYE-tus pigmen-TOE-suh): An eye disease that makes the retina slowly break down over time. It can lead to blindness.

Scoliosis (sko-lee-OH-sus): A problem with the spine that makes the spine curve sideways at certain points in-

stead of going straight up and down. Your child might have one shoulder that looks higher than the other. Or they might look normal but have back pain and tightness.

Swallow study: A test that shows what happens in the throat and esophagus (ee-SOF-uh-gus) when your child swallows.

Triglycerides (try-GLISS-err-ides): Fat in the bloodstream. It is normal to have this, but it can be dangerous if the level goes too high.

Appendix. Medications Used to Treat Symptoms in Cockayne Syndrome

This table was created from the clinical experiences of children with Cockayne syndrome who were prescribed these medications. Much of the content in this chart is based on case studies and parent or caregiver reports. It should be noted that not all children will react to medications in the same manner.

The table is constructed to have the physician discuss the information regarding safety and dosing concerns with individual choice of medications.

Drug class	Medication	Used for/clinical indication	Known reactions
Anesthesia	Propofol	Sedative	Cases of extreme drops in blood pressure reported in CS children
	Vecuronium	General anesthesia	Not to be used in people with liver/kidney issues; a case report of severe drop in blood pressure reported
Angiotensin II receptor blockers (ARBs)	Losartan	Used to lower blood pressure and reduce the risk of stroke Also used to slow long term kidney damage in patients with type 2 diabetes	No adverse reactions reported

Drug class	Medication	Used for/clinical indication	Known reactions
Antibiotics	Azithromycin	Antibiotic	No adverse reactions reported
	Cephalexin	Antibiotic	Diarrhea and vomiting reported in one case
	Clarithromycin	Antibiotic Also used with other meds to treat stomach ulcers	No adverse reactions reported
	Doxycycline	Antibiotic	Can cause permanent yellowing or greying of teeth
	Metronidazole	Antibiotic	AVOID! Can be fatal
	Trimethoprim	Antibiotic	Diarrhea and vomiting reported
	Trimoxazole	Antibiotic	No adverse reactions reported
Anticonvulsants	Levetiracetam	Antiepileptic	No adverse reactions reported
Antidiarrheals	Loperamide	Motility agent, antidiarrheal agent	No adverse reactions reported
Antihistamines	Cetirizine	Antihistamine	No adverse reactions reported
Bile acid sequestrants	Cholestyramine	Cholesterol-lowering agent	No adverse reactions reported
Calcium channel blockers	Amlodipine	Antihypertensive, treatment for angina, and other conditions caused by coronary artery disease	No adverse reactions reported

Drug class	Medication	Used for/clinical indication	Known reactions
D2 receptor antagonist	Domperidone	Motility agent	Regular heart monitoring and ECG recording should be performed when prescribed this medication
Decarboxylase inhibitors	Carbidopa-levodopa	Used for movement disorders and tremors	No adverse reactions reported
Gabapentinoids	Gabapentin	Anti epileptic	No adverse reactions reported
Histamine H2-receptor antagonist	Ranitidine	Gastric acid blocker	No adverse reactions reported
Human B-type natriuretic peptide	Nesiritide	Antihypertensive Afterload reducer for congestive heart failure	No adverse reactions reported
Insulin	Insulin aspart or glargine	Antidiabetogenic	No adverse reactions reported
NSAID	Naproxen	Antiinflammatory	No adverse reactions reported

Drug class	Medication	Used for/clinical indication	Known reactions
Opioids	Paracetamol-codeine	Analgesic	Prescribing doses are to be used with caution; reactions noted of increased analgesia and nonresponsive children due to dosage
	Morphine	Analgesic	Prescribing doses are to be used with caution; reactions noted of increased analgesia and nonresponsive children due to dosage
	Tramadol, hydrocodone, and other opioids	Analgesic	To be used with extra vigilance; CS children recorded to have an exaggerated response, ranging from respiratory depression to blunted affect that can last several days
Proton-pump inhibitors	Omeprazole	Gastric acid blocker	No adverse reactions reported
	Lansoprazole	Gastric acid blocker	No adverse reactions reported
Sedatives			To be used with extra vigilance; children with CS have been reported to have issues keeping warm and taking longer to wake up from anesthetics

Drug class	Medication	Used for/clinical indication	Known reactions
Skeletal muscle relaxants	Baclofen	Used to treat muscle symptoms including spasm and stiffness	No adverse reactions reported
Supplements	Melatonin	Helps to regulate sleep, commonly prescribed to treat insomnia	No adverse reactions reported
Thyroid hormones	Levothyroxine	Used to treat hypothyroidism (low thyroid hormone). It is also used to treat or prevent goiter (enlarged thyroid gland), which can be caused by hormone imbalances, radiation treatment, surgery, or cancer	No adverse reactions reported
Tricyclic antidepressants	Amitriptyline	Antidepressant	No adverse reactions reported