



**UNDERSTANDING
CONGENITAL
ATHYMIA**
A GUIDE FOR PARENTS
AND CAREGIVERS

MARCUS
Child with congenital athymia

UNDERSTANDING CONGENITAL ATHYMIA

WHAT IS CONGENITAL ATHYMIA?

Congenital athymia is an ultra-rare immune condition in which a child is born without a thymus.¹ Congenital athymia is a primary immunodeficiency, which occurs when part of the body's immune system is missing or doesn't function properly.²

The thymus is an organ that sits on top of the heart and plays a critical role in helping the immune system work.¹ The immune system is made of organs, cells, and proteins that work together throughout the body to fight infections.³ One important cell is the T cell. This is a type of white blood cell that attacks and remembers foreign invaders such as viruses, bacteria, fungi, and parasites.⁴ T cells begin in the bone marrow as immature cells, then travel to the thymus, where they become functioning T cells. The thymus selects T cells to fight infections and releases them into the bloodstream as part of an infection-fighting army.¹

Children with congenital athymia can face repeated infections because they do not have enough working T cells to fight the invaders off.¹ These infections can be fatal.¹ Without working T cells, children can also be affected by autoimmune conditions, when the body's immune system attacks and destroys healthy body tissue by mistake.⁵ With only supportive care, children with congenital athymia typically do not survive beyond 2 to 3 years of age.⁶

WHAT CAUSES CONGENITAL ATHYMIA?

The underlying cause or etiology of congenital athymia may involve any of several rare genetic or nongenetic conditions, such as^{1,7}:

- DiGeorge syndrome (also known as 22q11.2 deletion syndrome)
- CHARGE syndrome
- FOXP1 deficiency

Not everyone with congenital athymia has a related syndrome or genetic condition. And not everyone with a related syndrome or genetic condition has congenital athymia.⁸ If a child is diagnosed with congenital athymia, doctors will check to see if there are any additional underlying rare syndromes or genetic conditions. For some patients, there is no known cause for their congenital athymia.

HOW IS **CONGENITAL ATHYMIA** DIAGNOSED?

Congenital athymia is usually first detected in newborn screening for severe combined immune deficiency (SCID). Newborn screening is required in all 50 US states.¹ Congenital athymia is not SCID, but the test for SCID will show if there is a low number of T cells, which is seen in congenital athymia.¹

Based on the results of this newborn screening, your child's doctor determines what additional testing may be needed. Another test is needed after screening to confirm the diagnosis. After a positive newborn SCID screening result, a laboratory technique called flow cytometry is used to verify low naive T cells and strengthen the diagnosis of congenital athymia.¹

Early detection is key. The sooner congenital athymia is diagnosed, the sooner isolation and other measures designed to decrease the risk of infection can be started.⁹



SCREENING AND DIAGNOSIS

Primary immunodeficiencies occur when part of the body's immune system is missing or functions improperly.^{1,10}

SCID is a group of rare diseases caused by a different underlying immunodeficiency. Care and treatment plans are different for congenital athymia and SCID, so screening to determine the correct condition is important.¹

HOW DO I CARE FOR MY CHILD WITH CONGENITAL ATHYMIA?

Children with congenital athymia need special care. One of the most important things about caring for children with congenital athymia is isolation—both in the hospital and at home. Children with congenital athymia need to be isolated to avoid contact with germs that can cause infections.¹

Parents and caregivers of children with congenital athymia must also maintain strict isolation and hand-washing practices. Any interactions with individuals may expose the patient to germs that can lead to fatal infections.¹

Children diagnosed with congenital athymia are cared for by a specialized team of doctors. The pediatric clinical immunologist is the key member of this team. Other doctors may also play an important role, including pediatric endocrinologists (hormone specialists), cardiologists (heart specialists), otolaryngologists (ear, nose, and throat doctors), pediatric dermatologists (skin specialists), and infectious disease specialists.⁹

To keep children safe from life-threatening infections, they must be isolated from other people outside the home.¹ This can sometimes include family members, because they may be exposed to germs that may be fatal to a patient with congenital athymia.

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Anyone else who came into the home had to wear a mask, gown, and gloves, and of course, had to be healthy. No one could be sick and be around my child.”

LACEE

MOTHER | CAREGIVER



HOW CAN I **PROTECT MY CHILD** WITH CONGENITAL ATHYMIA?

Talk with your child’s doctor about creating a plan that is right for your family. Also, consider requesting doctor appointments before or after normal business hours to limit your child’s exposure to germs.

Your physician may suggest you establish a sanitation station at the home entrance. You should restrict visitors to the home; however, if you have visitors coming into the home, have them disinfect with hand sanitizer, remove shoes, and put on masks, gowns, and gloves if available through your doctor.¹

Educate everyone you know. It is important to educate siblings, other family members, friends, and teachers about congenital athymia soon after you receive the diagnosis. Make sure they understand what special precautions are needed, even though they are unlikely to be interacting with your child.

Have a “sick plan” in place. When another member of your household feels unwell or becomes sick, make sure they have a destination to go to (such as staying with a friend or another family member) until they are well enough to come home.

Your physician may suggest that your child not receive vaccinations until deemed appropriate. This includes vaccines with either live virus or inactivated virus. A vaccine could trigger a fatal infection or immune response.⁸ Talk to your physician prior to your child or any household members receiving vaccinations.



Creating a plan to limit the risk of germs in your home is crucial to protecting your child with congenital athymia



A SIMPLE COLD CAN BE A DANGER

Talk to your doctor to find out what is right for you and your family, which may include some of these potential guidelines:

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- Restrict visitors in the home
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- Encourage frequent handwashing in the home
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- Wipe down any items coming into the home
-
- Remind family members that they should shower and change clothes upon re-entry to the home from work or school
-
- Homeschool other children in the family and work from home, if possible
 - If siblings must attend school, ask the school to notify you of any outbreaks or illnesses that siblings might bring home
-
- Work with doctors to prevent exposure to sick children at medical appointments
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DON'T FORGET **SELF-CARE**

Children with congenital athymia need a lot of love, attention, and support—but so do you! Caring for a child with a life-threatening immune disorder can create feelings of extreme isolation, stress, and anxiety. Taking care of yourself is very important for you and your family.

You're not alone. Other families living with congenital athymia may better understand the challenges you face. Turning to social networks to connect with people who know what you are going through can be invaluable.

If possible, ask for help and establish a good support system with your family, friends, and healthcare team members to help you better cope with the complexities and challenges that come with the disease.

Take time for yourself. Remember to take the time you need to effectively manage your stress and maintain your own health—both physical and mental. Consider self-care from counseling, meditation, exercising, journaling, art, or becoming active within a support group.

Take time for others. Caring for a child with congenital athymia can feel overwhelming and all-consuming. Be sure to take the time you need to keep other interpersonal relationships—children, partner, family members, friends—close.

“

Lean on your family and friends and advocate for your child as best you can.”

AMANDA

MOTHER | CAREGIVER



RESOURCES AND SUPPORT

There are several patient advocacy groups that offer support for families dealing with the effects of immunodeficiency disorders.

The following organizations may help you cope with the ongoing challenges of caring for a child with congenital athymia:



Jeffrey Modell Foundation | info4pi.org

This nonprofit organization is dedicated to helping patients, families, and caregivers affected by immunodeficiency disorders find support, education, awareness, advocacy, and care.



Immune Deficiency Foundation | primaryimmune.org

This organization provides patients, families, and caregivers with valuable information related to immunodeficiency disorders.



National Organization for Rare Disorders | rarediseases.org

This patient organization, with over 300 patient organization members, is committed to the identification, treatment, and cure of rare disorders through programs of education, advocacy, research, and patient services.



Global Genes | globalgenes.org

This organization is focused on bringing positive change to the rare disease community and aims to connect, empower, and inspire affected patients, families, and caregivers.

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