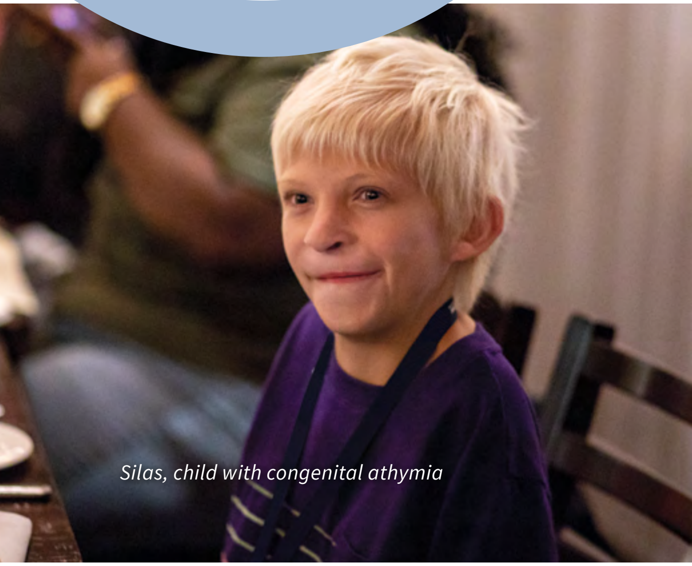
Understanding and Living With Congenital Athymia

A Guide for Parents and Caregivers



Silas, child with congenital athymia

As a parent or anyone caring for a child with congenital athymia, you naturally have questions and concerns. This guide offers some basics on the condition itself, what you need to be aware of, and what you can do to help protect children with congenital athymia. The information provided here is not intended to take the place of guidance from your physician. Talk to your doctor about any questions you may have.

athymia insights

What Is Congenital Athymia?

Congenital athymia is an ultra-rare primary immune deficiency that makes it harder for a child to fight off infections. Children with this condition are born without a functioning thymus—an organ that sits on top of the heart, behind the sternum (the long, flat bone located in the central part of your chest). The thymus plays a central role in everyone's immune system by helping important immune cells called *T cells* to develop.[1-3] You can think of the thymus as the "schoolhouse" of the immune system, where T cells mature and learn to fight infections.[2,3]

Children born without a thymus face repeated, often life-threatening infections because they do not have enough working T cells.[1] Historically, children with congenital athymia typically do not survive beyond 2 years of age.[1]

How Is Congenital Athymia Diagnosed?

Most children born with congenital athymia are first identified through a primary immune deficiency newborn screening test that is now part of standard practice in all 50 US states.[4,5] Based on the results of this newborn screening, a healthcare professional determines what additional testing may be needed. By using a technique called *flow cytometry*, a healthcare professional can then confirm if a child has congenital athymia.[4] The only way to confirm a diagnosis of congenital athymia is with this technique.

> ***“I realized his care is complex, so I needed to educate myself.”***
>
> **Lacee,**
> Mom of a child with congenital athymia

What Causes Congenital Athymia?

The underlying cause or *etiology* of congenital athymia may involve any of several rare genetic or nongenetic conditions, such as DiGeorge syndrome (also known as 22q11.2 deletion syndrome), CHARGE syndrome, or FOXN1 deficiency.[4,6,7] 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. 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 with congenital athymia, there may be no identifiable cause.[8]

How Is Congenital Athymia Treated?

Infants with congenital athymia have essentially only one viable option available to them—isolation to keep them from being infected, as well as the use of some medications to prevent infections. Even with this established standard of care, most infants diagnosed with congenital athymia and lacking a functioning immune system will eventually develop fatal infections.[1]

Once congenital athymia is diagnosed, an appropriate care plan can be put in place. Children diagnosed with this disorder are under the care of a medical specialist known as an *immunologist*.

Caring for a Child With Congenital Athymia

Children with congenital athymia are at risk of getting an infection that their body cannot fight. Even a common cold can be life threatening. The first line of defense is to limit a child's exposure to the many sources of infection.

Ways to Keep Your Home Healthy

Talk with your healthcare provider about creating a plan to keep your home healthy. From the outset, you need to inform siblings, other family members, friends, and teachers about congenital athymia and the need for taking special precautions when interacting with children with congenital athymia. Some suggestions include the following:

- *Physically isolating children with congenital athymia to avoid coming into contact with germs that can cause infections, including:*
  - *Limiting visitors to your home to minimize the introduction of germs into your house*
  - *Avoiding taking children with congenital athymia to public places, especially those with lots of other children who may have a lot of germs*
- *Using social media services such as FaceTime, Skype, or messaging apps so others can interact with you and children with congenital athymia*
- *Frequently wiping down household surfaces and items that are touched often, such as doorknobs, cell phones, tablets, car keys, and remote controls*
- *Making it a habit for everyone to frequently wash their hands*
- *Having everyone use masks, gowns, and gloves when coming into contact with a child with congenital athymia*
- *Asking schools to notify you of any outbreaks or illnesses that siblings might bring home*
- *Buying disposable paper products to simplify cleaning and disinfecting*
- *Ensuring that all family members are up to date on their vaccinations*

Other Precautions to Consider

- *Working with your healthcare providers to schedule appointments for before or after normal business hours in order to limit exposure to germs*
- *Establishing a sanitation station at the entrance to your home to allow anyone entering to disinfect, remove shoes, and put on masks, gowns, and gloves*
- *Making plans for where other members of your household can go if they are feeling sick, such as staying with a friend or family member*

Caring For the Whole Child

Every child with congenital athymia has different experiences, symptoms, and needs—especially if they have an underlying syndrome associated with the disease. And although protecting children with congenital athymia from infections serves as the first line of defense, other steps may also need to be taken. Some children, for example, need hearing aids, vision support, or physical, occupational, feeding, or speech therapy. Others may need surgery for heart or lung conditions. Your healthcare team will work with you to set up a care plan that's appropriate.

Daniel, child with congenital athymia

Resources

There are many resources available that may help you take care of yourself and your family, manage stress, deal with information overload, and more. Talk with your healthcare provider if you have any questions. The following organizations are independent of Enzyvant and may be helpful:

Jeffrey Modell Foundation

This nonprofit organization is dedicated to helping patients, families, and caregivers affected by immunodeficiency disorders find support, education, awareness, advocacy, and care. To learn more, visit info4pi.org

Immune Deficiency Foundation

Physicians, their healthcare teams, and patients can find valuable information and insights related to immunodeficiency disorders. To learn more, visit primaryimmune.org

- The Immune Deficiency Foundation's *Patient and Family Handbook for Primary Immunodeficiency Diseases* offers information and tools to enhance communication between families, caregivers, and their healthcare teams. To learn more, visit primaryimmune.org/resource/idf-patient-family-handbook

22q Family Foundation

Dedicated to raising awareness of 22q11.2 deletion syndrome (DiGeorge syndrome) and offering support and accurate information to families and caregivers affected by the disorder. To learn more, visit 22qfamilyfoundation.org

Global Genes

This organization pursues positive change and aims to connect, empower, and inspire the rare disease community. To learn more, visit globalgenes.org

> *We want to be there for other families. Nobody should have to go through what we went through.*
>
> **Dan,**
> Father of a child with congenital athymia

Don’t Forget—*You* Need Care, Too

Caring for a child with congenital athymia may change the way you interact with friends, family, and your community. Physical isolation may make you feel socially and emotionally isolated as well. Taking care of yourself is very important for you and your family.

You’re not alone. Other families living with congenital athymia understand the challenges you face and your frustration with people who may not “get it.” Many caregivers turn to social networks to find other families and organizations familiar with this condition. Having a network of people who know what you’re going through can be invaluable.

You may need to take time to manage your stress and maintain your own health—physical and mental. To cope, you may need to try new things to find out what works for you. Some people find relief and support from counseling, meditation, exercising, keeping a journal, expressing themselves through art, or joining a support group.

References

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