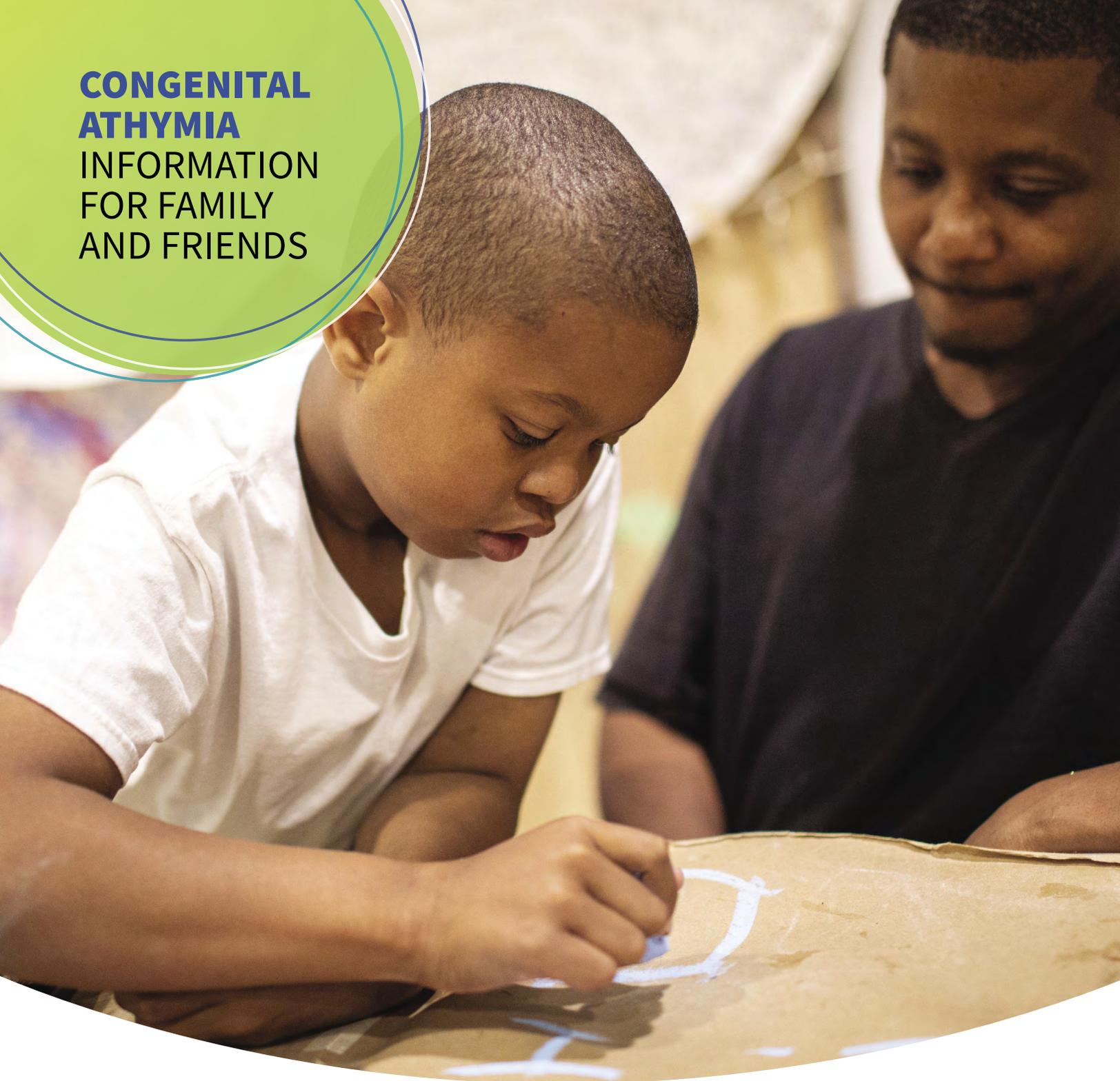


**CONGENITAL
ATHYMIA**
INFORMATION
FOR FAMILY
AND FRIENDS



This guide will explain congenital athymia, what a diagnosis means, and what you as a family member or friend can do to help the family affected by it.

WHAT IS CONGENITAL ATHYMIA?

Congenital athymia is an ultra-rare immune condition in which a child is born without a thymus.¹ This condition impacts approximately 17 to 24 infants out of every 4 million children born each year in the United States.²

Without development of a thymus prior to birth, children can face repeated infections and effects from autoimmune conditions because they do not have enough working T cells.^{1,3} These infections and autoimmune conditions can be fatal, and with only supportive care, children with congenital athymia typically do not survive beyond 2 to 3 years of age.⁴

CHILDREN WITH CONGENITAL ATHYMIA NEED SPECIAL CARE

Children with congenital athymia cannot fight infections.¹ Even a simple cold can be dangerous. Because of this, there are many practices and precautions that parents and caregivers must take on behalf of their child.

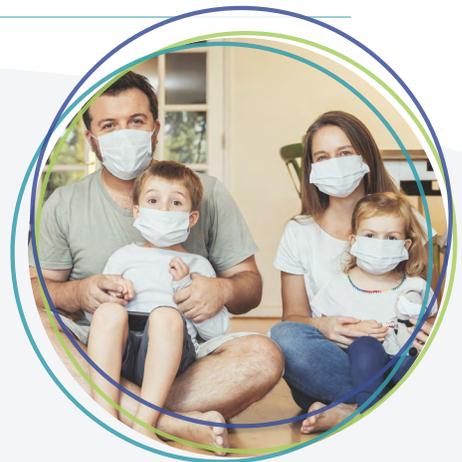
One of the most important aspects of care for children with congenital athymia is isolation. Children with congenital athymia need to be isolated from other people to avoid contact with germs that can cause infections.¹

Parents and caregivers of children with congenital athymia must also maintain strict isolation and hygiene practices within the home as well as outside. Any interactions with individuals outside the immediate household may expose the child to germs that can lead to fatal infections.¹

To keep children safe from life-threatening infections, they must be isolated from other people. This can sometimes include family members who may have been exposed to dangerous germs outside the home.

“*I can't stress how important our support system is. There will be times we need to hand over the care of our sick child for a few hours to someone we trust so we can focus on our healthy child, so they can receive that one-on-one attention.*”

MELYZA
CAREGIVER



HOW YOU CAN HELP

Close friends and relatives can help in big and small ways. The child's immediate family and caregivers are under a great deal of stress, especially while living in physical isolation. Offering empathy and showing that you care can help them feel less isolated and alone on this challenging medical journey. Any help you give will be greatly appreciated but you will need to ensure it is done in accordance with strict infection control measures as recommended by the child's physician.

Stay Informed: Educating yourself about congenital athymia helps reduce the burden on parents and caregivers of having to explain the condition and how it impacts their life.

Lend a Helping Hand: Volunteering to help can give the family much-needed relief. Ask what parents and caregivers are comfortable having you do. This might include bringing a meal, shopping for groceries, or other errands.

Learn to Socialize in New Ways: Your friends will appreciate your continued efforts to stay in touch, but their child's health status may not allow them to join social gatherings. Parents and caregivers may find it easier to simply stay home as disinfecting themselves when they return home from an outing requires time and effort. Stay in touch with phone calls or texts. You can also use social media services, including video calls for "face-to-face" visits.

Make Hospital Deliveries: The child's immediate family and caregivers may have multiple hospital stays throughout the year. During these times, parents and caregivers may appreciate receiving items that can make their stay more manageable. Listed below are some examples.

- Hand sanitizer and soap
- Face masks
- Hand lotion
- Comfy socks with grips on the bottom
- A soft blanket
- A pillow and extra pillowcases
- Dry shampoo, facial toner, lip balm, and body lotion
- Books and magazines
- An extension cord and phone charger
- Gift cards to online meal delivery services or restaurants that deliver



Parents and caregivers may appreciate it if you ask first about how their child is doing rather than about the condition itself. And remember not to take personally the restrictions they have in place.

RESOURCES AND SUPPORT

There are several patient advocacy groups that support families coping with immunodeficiency disorders like congenital athymia. The following organizations can help educate you on 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.

References:

1. Collins C, Sharpe E, Silber A, Kulke S, Hsieh EWY. Congenital athymia: genetic etiologies, clinical manifestations, diagnosis, and treatment. *J Clin Immunol*. 2021;41(5):881-895. doi.org/10.1007/s10875-021-01059-7.
2. Data on file, Enzyvant.
3. Markert ML, Gupton SE, McCarthy EA. Experience with cultured thymus tissue in 105 children. *J Allergy Clin Immunol*. Published online August 3, 2021. doi:10.1016/j.jaci.2021.06.028.
4. Hsieh EWY, Kim-Chang JJ, Kulke S, Silber A, O'Hara M, Collins C. Defining the clinical, emotional, social, and financial burden of congenital athymia. *Adv Ther*. 2021;38(8):4271-4288. doi.org/10.1007/s12325-021-01820-9.



www.enzyvant.com