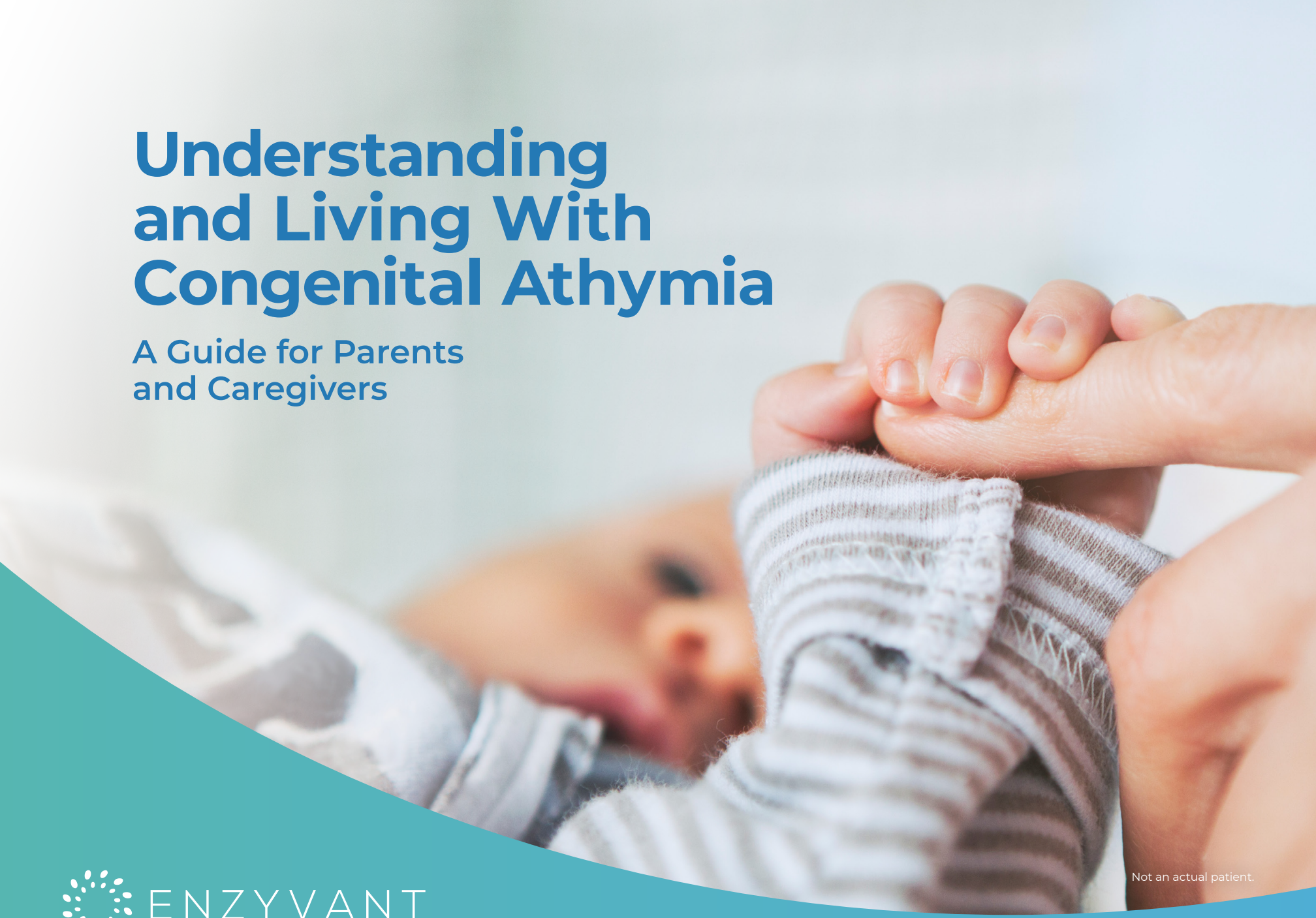
Understanding and Living With Congenital Athymia

A Guide for Parents and Caregivers



Not an actual patient.

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Introduction

As someone who cares for a child with congenital athymia, you naturally have questions and concerns. This guide offers some basics on the condition, what you need to be aware of, and what you can do to help protect your child.

The information provided here is not intended to take the place of guidance from your child's healthcare provider, whom you should talk to about any questions you may have.

Congenital athymia is a rare immune condition that requires children and often their families to live in isolation[1]

Congenital athymia is a primary immunodeficiency, which is a type of immune condition that occurs when part of the immune system is missing.[2,3]

Children with congenital athymia are born without a thymus. The thymus is an organ that sits on top of the heart and plays an important role in how the immune system works.[2]

Without a thymus, these children can face life-threatening infections because they are unable to produce enough naive T cells—white blood cells that help fight off the organisms that cause infections. They can also be affected by autoimmune conditions, which occur when the body's immune system attacks and destroys healthy tissue by mistake.[2,4]

Approximately 17 to 24 infants are born with congenital athymia in the US each year.[1]

Diagnosing congenital athymia

Congenital athymia is often detected through newborn screening for severe combined immunodeficiency (SCID), a test that is required in all 50 states in the US.[2]

While these two conditions are not the same, the test for SCID will likely indicate to your child's healthcare provider that further testing and examination are needed. After a positive screening result, an immunologist will use a laboratory technique called flow cytometry to verify low T cells and possibly lead to a diagnosis of congenital athymia.[2]

The sooner congenital athymia is identified, the sooner isolation and infection prevention measures can begin.[2]

Congenital athymia is associated with other genetic conditions

Congenital athymia has previously been referred to as complete DiGeorge anomaly, but it is now known to be associated with multiple genetic conditions, congenital syndromes, and environmental exposures. However, for some children there is no known cause.[2,5,6]

Prenatal testing may detect genetic abnormalities that are associated with congenital athymia, but congenital athymia is usually not detected until birth. Some of these associated conditions include[2,7]:

- **Complete DiGeorge syndrome**
- **CHARGE syndrome**
- ***FOXN1* deficiency**
- **Diabetic embryopathy**

Your child's healthcare provider may also test for additional underlying rare syndromes or genetic conditions.[2]

Protecting your child from infection requires constant care and attention[2]

Children with congenital athymia need special care to minimize the risk of life-threatening infections. One of the most important things that your child's healthcare provider will discuss with you is **isolation—both in the hospital and in the home.[2]**

Always talk to your child's healthcare provider to create a plan that is right for you and your family. Some steps they may recommend are:

- Limiting or restricting visitors in the home[2]
- Working with healthcare providers to prevent exposure to sick people at medical appointments[6]
- If possible, homeschooling other children in your family and/or working from home[8]
- Having a "sick plan" in place for when a member of your household feels ill
- Making sure everyone you know understands the severity of the diagnosis and that special precautions and isolation are needed to protect your child[2]
- Showering and changing clothes any time you leave and return home[2]
- Frequent handwashing[2]
- Wiping down any items, such as groceries, brought into the home
- Obtaining protective supplies like masks, gloves, and gowns[2]

There are several organizations for children with immune system diseases and their families that provide valuable support and education.

Scan the QR code to learn more, or visit congenital-athymia.com/resources

Your child may be cared for by a team of healthcare providers

Every child with congenital athymia is different. **Some may have symptoms and needs that must be addressed in addition to protecting them from infections.** For example, children may need hearing aids, vision support, or physical, occupational, feeding, or speech therapy. Others may need surgery for heart or lung conditions. **Your child's team of pediatric clinicians and specialists may include but are not limited to[2,6,8]:**

IMMUNOLOGISTS
for immune system disorders

CARDIOLOGISTS
for heart conditions

ENDOCRINOLOGISTS
for thyroid and parathyroid functions

OTOLARYNGOLOGISTS
for ear, nose, and throat conditions

GENETICISTS
for genetic screenings

INFECTIOUS DISEASE SPECIALISTS
for infections

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CONNECT is here to help

Enrolling in the Enzyvant CONNECT® Patient Support Program will give you and your family access to **educational resources** and, if eligible, **financial assistance** as you navigate the congenital athymia journey. Enzyvant CONNECT is available to patients with any type of insurance—including commercial plans, Medicare, or Medicaid—as well as patients who are underinsured or have no insurance coverage.

Dedicated care team

- Your Support Liaison will help you understand your child's diagnosis
- Your Access Specialist can help you navigate insurance benefits and financial assistance

Access to exclusive resources

- Document organizer
- *Sadie's Search*, a storybook written specifically with your child in mind
- Interactive T-cell progress tracker
- Activity book
- And more!

Co-pay program

- The Enzyvant CONNECT® Commercial Co-Pay Program can help caregivers of eligible commercially-insured patients in the US and US territories
- You may receive co-pay assistance for medication-related out-of-pocket costs for a treatment for congenital athymia

Call 844-ENZCNCT (844-369-2628) today to get connected to personalized support.
Support is available Monday–Friday, 8:00 AM to 8:00 PM ET.

Scan the QR code to start your enrollment, or visit EnzyvantCONNECT.com/get-started

Enzyvant and Enzyvant CONNECT are not responsible for treatment decisions or timing for treatment.

References: 1. 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:10.1007/s12325-021-01820-9 **2.** 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:10.1007/s10875-021-01059-7 **3.** Immune Deficiency Foundation. *Patient & family handbook for primary immunodeficiency diseases.* 6th ed. 2019. **4.** NCI Dictionary of Cancer Terms. T cell. National Cancer Institute. Accessed March 11, 2023. https://www.cancer.gov/publications/dictionaries/cancer-terms/def/t-cell **5.** Markert ML, Gupton SE, McCarthy EA. Experience with cultured thymus tissue in 105 children. *J Allergy Clin Immunol.* 2022;149(2):747-757. doi:10.1016/j.jaci.2021.06.028 **6.** Markert ML. Defects in thymic development. In: Sullivan KE, Stiehm ER, eds. *Stiehm's Immune Deficiencies: Inborn Errors of Immunity.* 2nd ed. Elsevier; 2020:357-379. **7.** Mustillo PJ, Sullivan KE, Chinn IK, et al. Clinical practice guidelines for the immunological management of chromosome 22q11.2 deletion syndrome and other defects in thymic development. *J Clin Immunol.* 2023;43(2):247-270. doi: 10.1007/s10875-022-01418-y **8.** Gupton SE, McCarthy EA, Markert ML. Care of children with DiGeorge before and after cultured thymus tissue implantation. *J Clin Immunol.* 2021;41(5):896-905. doi:10.1007/s10875-021-01044-0