Congenital Athymia

Information for Family and Friends

Mitchel, child with congenital athymia

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

athymia insights

What Is Congenital Athymia?

Congenital athymia is an ultra-rare immune disorder in which a child is born without a functioning thymus. The thymus—which sits on top of the heart, behind the sternum (the long, flat bone located in the central part of your chest)—is the organ that "teaches" your T cells how to defend your body against infections.[1,2] T cells are a type of white blood cell that are responsible for attacking and remembering foreign invaders such as viruses, bacteria, fungi, and parasites.[2] Without a thymus, a child can face repeated, often life-threatening infections because she or he does not have enough working T cells.[3] About 17 to 24 infants out of every 4 million babies born each year in the United States are diagnosed with congenital athymia, and historically, without treatment they do not survive beyond 2 years of age.[3,4]

Because congenital athymia occurs along with different underlying conditions, athymic children can have a range of other problems—such as heart disease, underactive parathyroid, eye or hearing abnormalities, or other physical deformities—that differ from child to child.[5,6] For this reason, every family is faced with a unique, often complicated medical journey that requires multiple specialists, ongoing appointments, and special care. And because they lack a fully functioning immune system, babies diagnosed with this disorder need to be isolated to keep them from getting infections.

What Causes Congenital Athymia and How Is It Diagnosed?

The most common cause of congenital athymia is DiGeorge syndrome, also referred to as 22q11.2 deletion syndrome.[7] Other known causes include genetic disorders known as CHARGE syndrome and FOXN1 deficiency.[5,8,9]

These rare conditions are first revealed through newborn screening. Every baby in the United States undergoes these screenings after birth.[5,10] Based on the results of that screening, the doctor overseeing the baby's care then determines what additional testing may be needed. A technique called *flow cytometry* is used by healthcare providers to diagnose congenital athymia.[5] A child is typically diagnosed with this condition within the first few weeks of life.

Living With Congenital Athymia

Because babies with this disorder cannot fight infections, there are practices and precautions of which everyone who comes in contact with them needs to be aware. Parents and caregivers should talk to their healthcare provider about creating a plan to keep their home healthy. Guidance available to families and caregivers generally includes but isn't limited to:

- *Isolating the affected child to avoid coming into contact with germs that can cause infections, including:*

  - *Avoiding taking the child to public places, especially those frequented by other children who may be carrying infectious germs*
  - *Restricting visits by anyone outside the immediate household*

- *Wiping down any items coming into the home with disinfectant products*
- *Making it a habit for everyone in the household to frequently wash their hands with antibacterial products*
- *Having family members and visitors coming into contact with the child use masks, gowns, and gloves*
- *Encouraging the community of family, other caregivers, and teachers to learn more about the very serious nature of the child's condition*
- *Staying informed about any illnesses at school that siblings might bring home*
- *Establishing a sanitation station at the entrance to the home to allow visitors to disinfect, remove shoes, and put on masks, gowns, and gloves*
- *Frequently cleaning household surfaces and items touched a lot such as cell phones, tablets, car keys, and remote controls*
- *Making plans for where other members of the household can go if they are feeling sick, such as staying with a friend or family member*

How Family and Friends Can Help

Close friends and relatives can help in big and small ways. Parents and caregivers may appreciate your asking first about how the child is doing rather than asking about the disease itself. And remember not to take the restrictions they have in place personally. 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. Helping out can take several forms, including:

- *Educating yourself about the disorder, which can reduce the burden on parents and caregivers of having to explain*
- *Offering to take in a family member who is ill to help avoid putting the child with athymia at risk for infection*
- *Running errands and asking which ones would be most helpful. These might include picking up kids from school or driving them to activities, bringing a meal, shopping for groceries, or babysitting so the caregivers can get some much-needed time away to exercise or relax*

Silas, child with congenital athymia

- *Socializing in new ways. Your loved ones may appreciate your continuing to stay in touch, but their child's health status may not allow them to join social gatherings. If parents or caregivers leave home, disinfecting themselves when they return takes time and effort—making it simpler for them to just "stay in." In these situations, technology such as Skype or FaceTime can help you stay connected*

- *Making hospital deliveries. The child's immediate family and caregivers may have many hospital stays and may appreciate having meals delivered. And for longer stays, families and caregivers appreciate:*
  - *Nice-smelling hand sanitizer and soap*
  - *Comfy socks with grips on the bottom*
  - *A soft blanket*
  - *A pillow and extra pillowcases*
  - *Dry shampoo, facial toner, lip balm, and lotion*
  - *An extension cord or a long phone charger cord*
  - *Gift cards to restaurants that deliver or online delivery services*

- *Holding or promoting events that can help to create awareness of congenital athymia in your community*

> ***"Being newly diagnosed was really overwhelming."***
>
> **Lacee,**
> Mom of a child with congenital athymia

Resources

There are several organizations independent from Enzyvant such as those listed that offer support, resources, education, and encouragement to patients, families, and caregivers living with immune disorders.

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

References

1. Lewis DE, Blutt SE. Organization of the immune system. In: Rich RR, Fleisher TA, Shearer WT, Schroeder HW, Frew AJ, Weyand CM, eds. *Clinical Immunology: Principles and Practice*. 5th ed. New York, NY: Elsevier; 2019:19-38.e1.

2. Alberts B, Johnson A, Lewis J, et al. *Molecular Biology of the Cell*. 6th ed. New York, NY: Garland Science; 2015.

3. Markert ML, Devlin BH, Chinn IK, McCarthy EA. Thymus transplantation in complete DiGeorge anomaly. *Immunol Res*. 2009;44(1-3):61-70.

4. Data on file, Enzyvant.

5. Markert ML. Defects in thymic development: DiGeorge/CHARGE/chromosome 22q11.2 deletion. In: Sullivan KE, Stiehm ER, eds. *Stiehm's Immune Deficiencies*. New York, NY: Elsevier; 2014:221-242.

6. Tang KL, Antshel KM, Fremont WP, Kates WR. Behavioral and psychiatric phenotypes in 22q11.2 deletion syndrome. *J Dev Behav Pediatr*. 2015;36(8):639-650.

7. Markert ML. Thymus transplantation. In: Sullivan KE, Stiehm ER, eds. *Stiehm's Immune Deficiencies.* New York, NY: Elsevier; 2014:1059-1067.

8. Davies EG. Immunodeficiency in DiGeorge syndrome and options for treating cases with complete athymia. *Front Immunol*. 2013;4:322.

9. Rota IA, Dhalla F. FOXN1 deficient nude severe combined immunodeficiency. *Orphanet J Rare Dis*. 2017;12(1):6.

10. All 50 states now screening newborns for severe combined immunodeficiency (SCID). Immune Deficiency Foundation (IDF) website. https://primaryimmune.org/news/all-50-states-now-screening-newborns-severe-combined-immunodeficiency-scid. Accessed October 25, 2019.

ENZYVANT

www.enzyvant.com

athymia insights

© 2020 Enzyvant Therapeutics, Inc. All rights reserved.
US--2000006