UNDERSTANDING ADA-SCID

Chiesi global rare diseases

WHAT IS ADA-SCID?[1]

Adenosine deaminase severe combined immune deficiency (ADA-SCID) is an ultra-rare, inherited genetic disorder, caused by mutations in the adenosine deaminase (*ADA*) gene, that is often fatal if left untreated.[1-3]

The ADA enzyme, which is produced by the *ADA* gene, is found in all cells and most active in lymphocytes, a type of white blood cell that is a key part of the body's immune system.[1] In ADA-SCID, ADA enzyme activity is reduced or eliminated, increasing the risk of severe and recurring infections.[1]

Mutated *ADA* gene impairs the body's immune system[1]

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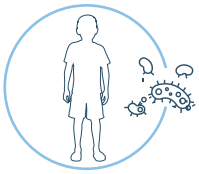

Weakened immune system cannot protect body from infections[2]

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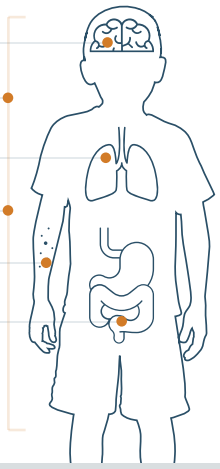
Increased risk of severe and recurring infections[1]

Most babies with ADA-SCID die by the age of 2 years unless they are diagnosed early and effective treatment is started.[3]

SIGNS AND SYMPTOMS

The most common signs and symptoms of ADA-SCID[1,3]:

- Developmental delays
- Recurrent infections
- Pneumonia[a]
- Slowed growth
- Skin rashes
- Chronic diarrhea



[a] Some lung abnormalities in ADA-SCID, as well as in other types of severe combined immune deficiency (SCID), may be caused by metabolic insufficiency, not just infections.[1]

INCIDENCE AND PREVALENCE

- Affects approximately **1** in **200,000** to **1** in **1 million** newborns around the world[3]
- Responsible for approximately **15%** of severe combined immune deficiency (SCID) cases[3]

DIAGNOSIS

ADA-SCID is typically diagnosed within the first few months of life.[1] Patients can be diagnosed in 2 ways[3]:

- A blood test that measures ADA enzyme activity
- Genetic testing for an *ADA* gene mutation

Genetic testing involves examining a person's DNA, the chemical database that carries instructions for the body's functions.[3] Genetic testing can reveal mutations in your genes that may cause illness or disease.[3]

Today, all 50 states, the District of Columbia, and Puerto Rico are currently performing newborn screening for SCID, which can also help identify cases of ADA-SCID.[4]

TREATMENT OPTIONS

Patients should always consult a physician to see which therapy is best for them.

Hematopoietic stem cell transplant (HSCT) replaces defective immune cells with healthy immune cells from a donor[2]

Enzyme replacement therapy (ERT) helps to restore immune function[2]

CHIESI GLOBAL RARE DISEASES: OUR COMMITMENT, YOUR EMPOWERMENT

Chiesi has been focused on the centrality of the patient for decades. To sharpen that focus, Chiesi created a specialty unit dedicated to the development and commercialization of products for rare diseases—**Chiesi Global Rare Diseases**.

We believe that no patient should be left behind, which is why we created a business unit specifically dedicated to those with rare diseases. Patients with rare diseases can encounter many difficulties. Chiesi Global Rare Diseases is here to help ease that burden.

References:

1. Whitmore KV, Gaspar HB. Adenosine deaminase deficiency – more than just an immunodeficiency. *Front Immunol.* 2016;7:314. doi:10.3389/fimmu.2016.00314.
2. Sauer AV, Brigida I, Carriglio N, Aiuti A. Autoimmune dysregulation and purine metabolism in adenosine deaminase deficiency. *Front Immunol.* 2012;3:265. doi:10.3389/fimmu.2012.00265.
3. Hershfield M. Adenosine Deaminase Deficiency. In: Adam MP, Ardinger HH, Pagon RA, et al, eds. *GeneReviews®*. University of Washington, Seattle; 1993-2020. Posted October 3, 2006. Updated March 16, 2017. https://www.ncbi.nlm.nih.gov/books/NBK1483/.
4. van der Burg M, Mahlaoui N, Gaspar HB, Pai S-Y. Universal newborn screening for severe combined immunodeficiency (SCID). *Front Pediatr.* 2019;7:373. doi:10.3389/fped.2019.00373.