8 Frequently Asked Questions About
MCT8 DEFICIENCY

This resource provides key questions and answers about MCT8 deficiency (Allan–Herndon–Dudley syndrome [AHDS]) for you and for family or friends you want to educate about the condition. For more information and resources, please visit **lifewithmct8deficiency.com**.

Q1. What is MCT8 deficiency?

MCT8 deficiency is a rare genetic disorder that makes it hard for the brain to get enough T3 thyroid hormone while the rest of the body gets too much. This imbalance can lead to serious developmental impairment, feeding and growth problems, heart concerns, and muscle weakness or stiffness and overall health concerns.

Q2. What causes MCT8 deficiency?

MCT8 deficiency is caused by changes in a gene called *SLC16A2*, which makes a protein called MCT8. This protein helps move thyroid hormones (especially one called T3) in and out of the cells, including the brain.

Thyroid hormones are very important for growth and development and for controlling how the body uses energy. When MCT8 doesn’t work properly, the body and brain are affected in different ways.

Q3. Who can have MCT8 deficiency?

MCT8 deficiency mostly affects boys because the gene is located on the X chromosome. Boys have only 1 X chromosome, so if the gene is changed, they will show symptoms.

Girls have 2 X chromosomes, so they can carry the gene change without being as severely affected. Some girls may have mild symptoms, but it is much less common for them to experience the full effects of the condition.

Q4. What are the main signs of MCT8 deficiency?

In the first 2-3 months of life, symptoms of MCT8 deficiency may begin to appear, and may include:

- Trouble holding up the head
- Delays in reaching developmental milestones
- High T3 hormone levels
- Ongoing loss of body weight
- Sleep problems

Over time, these signs and symptoms may worsen and additional symptoms may appear, including:

- Delays in movement and daily activities
- Feeding challenges
- Problems with muscles
- Unusual movements
- Heart-related issues
- Increased fussiness
- Frequent infections
- Overall impact on daily life

Q5. How is MCT8 deficiency diagnosed?

Boys who show developmental impairment, low muscle tone, fast resting heart rate, and ongoing loss of body weight should have thyroid hormone testing that includes measuring levels of the hormones T3, T4, and TSH.

The testing result associated with MCT8 deficiency is:

- High T3
- Low to low-normal free T4
- Normal or slightly elevated TSH

To confirm the diagnosis, testing should also include a genetic test to look for mutations in the *SLC16A2* gene.

Q6. Can MCT8 deficiency be treated?

Currently, there are no approved treatments for MCT8 deficiency in the United States. Care focuses on managing symptoms, preventing complications, and supporting comfort and quality of life. Most people need ongoing medical support, and care plans are tailored to each person's needs.

Q7. What is it like to live with MCT8 deficiency?

People with MCT8 deficiency can experience a wide range of symptom severity. Those living with MCT8 deficiency typically need significant, ongoing assistance, either from devoted family caregivers or in a specialized care setting.

Q8. Where can families find support?

Below is a list of organizations that provide education and support for families living with disorders like MCT8 deficiency. The websites listed are not owned or operated by Egetis Therapeutics or its affiliates. Egetis Therapeutics does not control, endorse, or guarantee the content of third-party websites.

MCT8-AHDS Foundation (Nonprofit Organization)
www.mct8.info

Child Neurology Foundation
www.childneurologyfoundation.org

RareConnect
www.rareconnect.org

Patient Advocate Foundation
www.patientadvocate.org

Caring for someone with MCT8 deficiency can feel overwhelming, but you are not alone. With the right care team and support resources, families can help children live as comfortably and fully as possible. Staying informed and connected is an important part of managing this disorder.

For more detailed information and resources, visit lifewithmct8deficiency.com

EGETIS
THERAPEUTICS