

MCT8 DEFICIENCY

EMERGENCY MEDICAL INFORMATION

This guide is provided for general educational purposes only and is not medical advice. It does not replace the judgment of your child's healthcare team. In an emergency, call 911 or your local emergency number. Always follow the instructions of your child's treating clinicians.

What Is Monocarboxylate Transporter 8 (MCT8) Deficiency?

MCT8 deficiency (Allan–Herndon–Dudley syndrome [AHDS]) is a rare X-linked disorder caused by pathogenic mutations in the *SLC16A2* gene.¹ This gene encodes MCT8, which is essential for transporting thyroid hormones—particularly triiodothyronine (T3)—into cells and across the blood-brain barrier.² When MCT8 function is impaired, 2 concomitant clinical presentations occur:

- **Central effect** – Insufficient T3 in the brain leads to severe neurodevelopmental impairment and intellectual disability¹
- **Peripheral effect** – Persistent thyrotoxicosis leads to systemic complications such as failure to thrive, irritability, sleep disturbance, muscle wasting and hypermetabolism, and cardiovascular symptoms including tachycardia^{1,3}

Emergency Situation Considerations

Chronic peripheral thyrotoxicosis	• Results from elevated serum T3^{1,4,5} • Monitor T3 levels
Cardiac Monitoring	• Predisposes to tachycardia, hypertension, and arrhythmias^{4,5} • Sudden death, likely related in part to underlying cardiac arrhythmias, is a leading cause of mortality^{1,6}
Respiratory Risk	• Recurrent pulmonary infections are a common cause of death, likely related to cardiac etiologies as listed above^{5,6} • Truncal hypotonia, reduced muscle mass, and scoliosis impair cough and ventilatory reserve^{1,4}
Aspiration Risk	• High baseline aspiration risk from dysphagia, poor head and neck control, axial hypotonia, and gastroesophageal reflux^{1,4} • Many patients require gastrostomy tube (G-tube) feeding because of severe feeding difficulties and aspiration risk⁵ • Aspiration pneumonia is a documented cause of early death^{1,4}
Seizures	• True seizures: present in approximately 25% of patients⁴; often drug resistant⁴ • Changes in barometric pressure may trigger seizure episodes⁷ • Spastic activity during sleep-to-wake transitions may be mistaken for seizures⁸ • Paroxysmal kinesigenic dyskinesias: brief stereotyped episodes of body extension, mouth opening, and limb stretching or flexing^{4,9} – Typically lasting 1 to 2 minutes⁴ – May be triggered by physical stimuli such as diaper changes, clothing changes, or lifting^{4,9} – Consciousness preserved; resolve spontaneously^{4,10}

References: 1. Bauer AJ, Auble B, Clark AL, et al. *Front Pediatr.* 2024;12:1444919. 2. van Geest FS, Gunhanlar N, Groeneweg S, Visser WE. *Front Endocrinol (Lausanne).* 2021;12:723750. 3. Groeneweg S, van Geest FS, Peeters RP, Heuer H, Visser WE. *Endocr Rev.* 2020;41(2):146–201. 4. Sarret C, Oliver Petit I, Tonduti D. In: Adam MP, Feldman J, Mirzaa GM, et al, eds. *GeneReviews® [Internet]*. University of Washington, Seattle; 1993–2026. Updated January 16, 2020. Accessed May 26, 2026. <https://www.ncbi.nlm.nih.gov/books/NBK26373> 5. Children's Hospital of Philadelphia. Accessed May 26, 2026. <https://www.chop.edu/conditions-diseases/mct8-deficiencyallan-herndon-dudley-syndrome-ahds> 6. Groeneweg S, van Geest FS, Abaci A, et al. *Lancet Diabetes Endocrinol.* 2020;8(7):594–605. 7. Rakers F, Walther M, Schiffner R, et al. Weather as a risk factor for epileptic seizures: a case-crossover study. *Epilepsia.* 2017;58(7):1287–1295. doi:10.1111/epi.13776 8. Boelaert K, Bauer A, Cappola AR, et al. Thyrotoxicosis in MCT8 deficiency. *J Clin Endocrinol Metab.* Published online January 9, 2026. doi:10.1210/clinem/dgaf707 9. Ramon-Gomez JL, Cortés-Rojas MC, Polania-Puentes MJ, Guerrero-Ruiz GDP. *Mov Disord Clin Pract.* 2024;11(5):567–570. 10. Garone G, Capuano A, Travaglini L, et al. *Int J Mol Sci.* 2020;21(10):3603.

MCT8 DEFICIENCY (AHDS) PATIENT INFORMATION

Name: _____ Date of Birth: _____

Diagnosis Date: _____ Primary Caregiver: _____

Phone: _____

Insurance Provider Name: _____ Subscriber Name: _____

Member/ID Number: _____ Group Number: _____

Additional Insurance: _____

Allergies: _____

Independently Ambulatory ☐ Nonambulatory ☐

Ambulatory With: Assisted Device ☐ _____ Another Person's Assistance ☐

Do-Not-Resuscitate (DNR): ☐ Yes ☐ No DNR Active: ☐ Yes ☐ No

This is a caregiver record only and is not a valid Do-Not-Resuscitate order. Please provide your child's official DNR/POLST/MOLST documentation to emergency providers separately.

Feeding Method: ☐ Oral ☐ G-tube ☐ G-J tube ☐ Nothing by Mouth

Feeding Method Details (frequency, times, volume, delivery device, rate): _____

G-tube Surgery Date: _____ Last Change Date: _____

Other Surgeries: _____

Verbal: ☐ Yes ☐ No Communication Method: ☐ Cards ☐ AAC Device Other: _____

Specify approaches to handling cardiac issues, respiratory distress, aspiration, and/or seizures experienced by your child:

MCT8 DEFICIENCY (AHDS) PATIENT INFORMATION

Current Medication and Dosing (name, dose, route, frequency, reason): _____

Symptoms: _____

MCT8 DEFICIENCY (AHDS) PATIENT INFORMATION

Medical Team Contacts

Medical team	Name	Office contact	Phone	Email
Primary care provider				
Pediatric neurologist				
Pediatric endocrinologist				
Genetic counselor				
Cardiologist				
Gastroenterologist				
Nutrition specialist				
Pulmonologist				
Orthopedic specialist				
Physical therapist				
Occupational therapist				
Speech-language pathologist				
Feeding therapist				
Nursing agency				
Hospice agency				
Clinical geneticist				