

Disease

# Monocarboxylate transporter 8 deficiency<sup>1, 2, 3</sup>

Gene

## SLC16A2

|                                                 |                                                                                                                                                                                                                                   |
|-------------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Inheritance</b>                              | XL                                                                                                                                                                                                                                |
| <b>Current Treatments</b>                       | Emcitate (tiratricol/triiodothyroacetic acid/TRIAC)                                                                                                                                                                               |
| <b>Evidence for Treatment</b>                   | expert opinion                                                                                                                                                                                                                    |
| <b>Non-Molecular Testing Availability</b>       | yes                                                                                                                                                                                                                               |
| <b>Non-Molecular Test</b>                       | TSH, free T3, free T4 <sup>1</sup>                                                                                                                                                                                                |
| <b>Experimental / Investigational Therapies</b> | <a href="https://clinicaltrials.gov/ct2/results?cond=Monocarboxylate+Transporter+8+%28Mct8%29+Deficiency&amp;term=">https://clinicaltrials.gov/ct2/results?cond=Monocarboxylate+Transporter+8+%28Mct8%29+Deficiency&amp;term=</a> |

References

- <sup>1</sup> <https://www.ncbi.nlm.nih.gov/books/NBK26373/>
- <sup>2</sup> <https://pubmed.ncbi.nlm.nih.gov/34679181/>
- <sup>3</sup> <https://pubmed.ncbi.nlm.nih.gov/40549098/>