

Disease

# Central hypothyroidism and testicular enlargement<sup>1</sup>

Gene

## IGSF1

|                                                 |                                                                                                                                                                                                                                                                   |
|-------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Inheritance</b>                              | XL                                                                                                                                                                                                                                                                |
| <b>Current Treatments</b>                       | levolevothyroxine                                                                                                                                                                                                                                                 |
| <b>Evidence for Treatment</b>                   | guideline <sup>2</sup>                                                                                                                                                                                                                                            |
| <b>Non-Molecular Testing Availability</b>       | yes                                                                                                                                                                                                                                                               |
| <b>Non-Molecular Test</b>                       | TSH, free T4 <sup>1</sup>                                                                                                                                                                                                                                         |
| <b>Experimental / Investigational Therapies</b> | <a href="https://clinicaltrials.gov/ct2/results?cond=Congenital+Hypothyroidism&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=">https://clinicaltrials.gov/ct2/results?cond=Congenital+Hypothyroidism&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=</a> |

References

<sup>1</sup> <https://omim.org/entry/300888>

<sup>2</sup> <https://pubmed.ncbi.nlm.nih.gov/33272083/>