

Disease

# Choreoathetosis and congenital hypothyroidism with or without pulmonary dysfunction<sup>1, 2</sup>

Gene

## NKX2-1

|                                                 |                                                                                                                                                                                                                                                                   |
|-------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Inheritance</b>                              | AD                                                                                                                                                                                                                                                                |
| <b>Current Treatments</b>                       | levolevothyroxine, levodopa, tetrabenazine                                                                                                                                                                                                                        |
| <b>Evidence for Treatment</b>                   | guideline <sup>3</sup>                                                                                                                                                                                                                                            |
| <b>Non-Molecular Testing Availability</b>       | yes                                                                                                                                                                                                                                                               |
| <b>Non-Molecular Test</b>                       | TSH, 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/610978>

<sup>2</sup> <https://www.ncbi.nlm.nih.gov/books/NBK185066/>

<sup>3</sup> <https://pubmed.ncbi.nlm.nih.gov/33272083/>