

**Disease**

# Immunodeficiency-centromeric instability-facial anomalies syndrome 2<sup>1, 2</sup>

**Gene**

## ZBTB24

|                                                 |                                                                                                                                                                                                                             |
|-------------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Inheritance</b>                              | AR                                                                                                                                                                                                                          |
| <b>Current Treatments</b>                       | replacement immunoglobulin treatment, Bone marrow transplant (hematopoietic stem cell transplantation (HSCT))                                                                                                               |
| <b>Evidence for Treatment</b>                   | expert opinion                                                                                                                                                                                                              |
| <b>Non-Molecular Testing Availability</b>       | yes                                                                                                                                                                                                                         |
| <b>Non-Molecular Test</b>                       | immunoglobulin levels, cytogenetic analysis for centromeric instability, DNA methylation studies <sup>3</sup>                                                                                                               |
| <b>Experimental / Investigational Therapies</b> | <a href="https://clinicaltrials.gov/ct2/results?cond=ZBTB24&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=">https://clinicaltrials.gov/ct2/results?cond=ZBTB24&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=</a> |

**References**

<sup>1</sup> <https://omim.org/entry/614069>

<sup>2</sup> <https://pubmed.ncbi.nlm.nih.gov/28128455/>

<sup>3</sup> <https://pubmed.ncbi.nlm.nih.gov/33082427/>