

Disease

Immunodeficiency-centromeric instability-facial anomalies syndrome 1^{1, 2, 3}

Gene

DNMT3B

Inheritance	AR
Current Treatments	replacement immunoglobulin treatment, Bone marrow transplant (hematopoietic stem cell transplantation (HSCT))
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	immunoglobulin levels, cytogenetic analysis for centromeric instability, DNA methylation studies ⁴
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=DNMT3B&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/pubmed/30511102>

² <https://www.ncbi.nlm.nih.gov/pubmed/30353301>

³ <https://pubmed.ncbi.nlm.nih.gov/33544358/>

⁴ <https://pubmed.ncbi.nlm.nih.gov/33082427/>