

Disease

Immunodeficiency-centromeric instability-facial anomalies syndrome 4^{1, 2}

Gene

HELLS

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 ³
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=hells&term=&cntry=&state=&city=&dist=

References

- ¹ <https://omim.org/entry/616911>
- ² <https://pubmed.ncbi.nlm.nih.gov/26216346/>
- ³ <https://pubmed.ncbi.nlm.nih.gov/33082427/>