

Disease

Common variable immune deficiency 13^{1, 2, 3}

Gene

IKZF1

Inheritance	AD
Current Treatments	replacement immunoglobulin treatment, Bone marrow transplant (hematopoietic stem cell transplantation (
Evidence for Treatment	guideline ⁴
Non-Molecular Testing Availability	yes
Non-Molecular Test	immunoglobulin levels ⁵
Disease Frequency (per 100,000)	0.4 ⁶
Genes Included in Disease Frequency	NFKB1, TNFRSF13B, TNFRSF13C, CD19, CD81, MS4A1, CR2, LRBA, PIK3CD, NFKB2, IRF2BP2, ICOS, IL21, IKZF1
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Common+Variable+Immunodeficiency&term=&cntry=&state=&city=

References

- ¹ <https://www.ncbi.nlm.nih.gov/pubmed/31089937>
- ² <https://www.ncbi.nlm.nih.gov/pubmed/30965037>
- ³ <https://pubmed.ncbi.nlm.nih.gov/33392855/>
- ⁴ <https://pubmed.ncbi.nlm.nih.gov/23730886/>
- ⁵ <https://omim.org/entry/616873>
- ⁶ <https://ghr.nlm.nih.gov/condition/common-variable-immune-deficiency#genes>