

## Disease

# Common variable immune deficiency 10<sup>1, 2</sup>

## Gene

### NFKB2

|                                                 |                                                                                                                                                                                                                                                             |
|-------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Inheritance</b>                              | AD                                                                                                                                                                                                                                                          |
| <b>Current Treatments</b>                       | replacement immunoglobulin treatment, cortisol                                                                                                                                                                                                              |
| <b>Evidence for Treatment</b>                   | guideline <sup>3</sup>                                                                                                                                                                                                                                      |
| <b>Non-Molecular Testing Availability</b>       | yes                                                                                                                                                                                                                                                         |
| <b>Non-Molecular Test</b>                       | immunoglobulin levels <sup>4</sup>                                                                                                                                                                                                                          |
| <b>Disease Frequency (per 100,000)</b>          | 0.4 <sup>5</sup>                                                                                                                                                                                                                                            |
| <b>Genes Included in Disease Frequency</b>      | NFKB1, TNFRSF13B, TNFRSF13C, CD19, CD81, MS4A1, CR2, LRBA, PIK3CD, NFKB2, IRF2BP2, ICOS, IL21, IKZF1                                                                                                                                                        |
| <b>Experimental / Investigational Therapies</b> | <a href="https://clinicaltrials.gov/ct2/results?cond=Common+Variable+Immunodeficiency&amp;term=&amp;cntry=&amp;state=&amp;city=">https://clinicaltrials.gov/ct2/results?cond=Common+Variable+Immunodeficiency&amp;term=&amp;cntry=&amp;state=&amp;city=</a> |

## References

- <sup>1</sup> <https://www.ncbi.nlm.nih.gov/pubmed/31417880>
- <sup>2</sup> <https://pubmed.ncbi.nlm.nih.gov/30941118/>
- <sup>3</sup> <https://pubmed.ncbi.nlm.nih.gov/23730886/>
- <sup>4</sup> <https://omim.org/entry/615577>
- <sup>5</sup> <https://ghr.nlm.nih.gov/condition/common-variable-immune-deficiency#genes>