

## Disease

# RAC2 associated severe combined immunodeficiency<sup>1, 2</sup>

## Gene

### RAC2

|                                                 |                                                                                                                                                                                                                                                                |
|-------------------------------------------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Inheritance</b>                              | AR and AD                                                                                                                                                                                                                                                      |
| <b>Current Treatments</b>                       | Hematopoietic stem cell transplantation (HSCT) - bone marrow transplant, immunoglobulin replacement                                                                                                                                                            |
| <b>Evidence for Treatment</b>                   | expert opinion                                                                                                                                                                                                                                                 |
| <b>Non-Molecular Testing Availability</b>       | yes                                                                                                                                                                                                                                                            |
| <b>Non-Molecular Test</b>                       | T and B Lymphocyte and Natural Killer Cell Profile, immunoglobulin levels <sup>2</sup>                                                                                                                                                                         |
| <b>Disease Frequency (per 100,000)</b>          | 1.72 <sup>3</sup>                                                                                                                                                                                                                                              |
| <b>Genes Included in Disease Frequency</b>      | JAK3, IL2RG, LCK, NHEJ1, FOXN1, PNP, PRKDC, ORAI1, IL7R, PTPRC, ADA, RAG1, RAG2, DCLRE1C, CD3D, CD3E, CD247, AK2, CORO1A, CHD7, CIITA, RFXANK, RFX5, RFXAP, DOCK8, DOCK2, LIG4, MTHFD1, NBN, SLC46A1, PGM3, RMRP, STIM1, TTC7A, ZAP70, RAC2, PAX1, LAT, NUDCD3 |
| <b>Experimental / Investigational Therapies</b> | <a href="https://clinicaltrials.gov/ct2/results?cond=rac2&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=">https://clinicaltrials.gov/ct2/results?cond=rac2&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=</a>                                        |

## References

<sup>1</sup> <https://www.ncbi.nlm.nih.gov/pubmed/30723080>

<sup>2</sup> <https://pubmed.ncbi.nlm.nih.gov/32542921/>

<sup>3</sup> [www.mdpi.com/2409-515X/3/2/15/pdf](http://www.mdpi.com/2409-515X/3/2/15/pdf)