

Disease

X-linked severe combined immunodeficiency^{1, 2}

Gene

IL2RG

Inheritance	XL
Current Treatments	Hematopoietic stem cell transplantation (HSCT) - bone marrow transplant, gene therapy (MB-107 - clinical trial)
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	T and B Lymphocyte and Natural Killer Cell Profile ³
Disease Frequency (per 100,000)	1.72 ⁴
Genes Included in Disease Frequency	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
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=il2rg&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK1410/>
- ² <https://www.ncbi.nlm.nih.gov/pubmed/30995372>
- ³ https://www.ncbi.nlm.nih.gov/books/NBK1410/#x-scid.Differential_Diagnosis
- ⁴ www.mdpi.com/2409-515X/3/2/15/pdf