Rx-Genes
Treatment Guidance at the Point of Care

Disease

JAK3 associated T cell-negative, B cell-positive, natural killer cell-negative severe combined immunodeficiency[1]

Gene

JAK3

Inheritance	AR
Current Treatments	Hematopoietic stem cell transplantation (HSCT) - bone marrow transplant
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	T and B Lymphocyte and Natural Killer Cell Profile[2]
Disease Frequency (per 100,000)	1.72[3]
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=jak3&term=&cntry=&state=&city=&dist=

References

[1] https://omim.org/entry/600802

[2] https://www.ncbi.nlm.nih.gov/books/NBK1410/#x-scid.Differential_Diagnosis

[3] www.mdpi.com/2409-515X/3/2/15/pdf