Rx-Genes
Treatment Guidance at the Point of Care

Disease

Bare lymphocyte syndrome, type II, complementation group D[1, 2]

Gene

RFXAP

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	flow cytometric analysis of HLA-DR expression on monocytes and B cells [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, CD3 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=Bare+Lymphocyte+Syndrome&term=&cntry=&state=&city=&dis

References

[1] https://pubmed.ncbi.nlm.nih.gov/22390233/

[2] https://omim.org/entry/209920

[3] https://pubmed.ncbi.nlm.nih.gov/25001848/

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