

Disease

Severe combined immunodeficiency, Athabascan type^{1, 2}

Gene

DCLRE1C

Inheritance	AR
Current Treatments	Hematopoietic stem cell transplantation (HSCT) - bone marrow transplant, autologous CD34+ cells, transfected with a lentiviral vector containing DCLRE1C,
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=DCLRE1C&term=&cntry=&state=&city=&dist=

References

- ¹ <https://omim.org/entry/602450>
- ² <https://pubmed.ncbi.nlm.nih.gov/36546626/>
- ³ https://www.ncbi.nlm.nih.gov/books/NBK1410/#x-scid.Differential_Diagnosis
- ⁴ www.mdpi.com/2409-515X/3/2/15/pdf