

Disease

Cartilage-hair hypoplasia^{1, 2, 3}

Gene

RMRP

Inheritance	AR
Current Treatments	Hematopoietic stem cell transplantation (HSCT) - bone marrow transplant
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	no
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=Cartilage-hair+Hypoplasia&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK84550/>
- ² <https://www.ncbi.nlm.nih.gov/pubmed/20375313>
- ³ <https://pubmed.ncbi.nlm.nih.gov/31379817/>
- ⁴ www.mdpi.com/2409-515X/3/2/15/pdf