

Disease

Osteopetrosis type 1^{1, 2}

Gene

TCIRG1

Inheritance	AR
Current Treatments	Bone marrow transplant (hematopoietic stem cell transplantation (HSCT))
Evidence for Treatment	guideline ³
Non-Molecular Testing Availability	yes
Non-Molecular Test	skeletal survey ¹
Disease Frequency (per 100,000)	0.4 ⁴
Genes Included in Disease Frequency	TCIRG1, CLCN7, CA2, SNX10, TNFRSF11A
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Osteopetrosis&term=&cntry=&state=&city=&dist=

References

- ¹ <https://pubmed.ncbi.nlm.nih.gov/31859718/>
- ² <https://www.ncbi.nlm.nih.gov/pubmed/24753205>
- ³ <https://esid.org/layout/set/print/content/view/full/14267>
- ⁴ <https://www.ncbi.nlm.nih.gov/books/NBK1127/#clcn7.Summary>