

Disease

Osteopetrosis type 7^{1, 2, 3}

Gene

TNFRSF11A

Inheritance	AR
Current Treatments	Bone marrow transplant (hematopoietic stem cell transplantation (HSCT))
Evidence for Treatment	guideline ⁴
Non-Molecular Testing Availability	yes
Non-Molecular Test	skeletal survey ³
Disease Frequency (per 100,000)	0.4 ⁵
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://www.ncbi.nlm.nih.gov/pubmed/30885997>
- ² <https://pubmed.ncbi.nlm.nih.gov/27187610/>
- ³ <https://pubmed.ncbi.nlm.nih.gov/33364264/>
- ⁴ <https://esid.org/layout/set/print/content/view/full/14267>
- ⁵ <https://www.ncbi.nlm.nih.gov/books/NBK1127/#clcn7.Summary>