

Disease

Osteopetrosis type 4¹

Gene

CLCN7

Inheritance	AR and AD
Current Treatments	Bone marrow transplant (hematopoietic stem cell transplantation (HSCT))
Evidence for Treatment	guideline ²
Non-Molecular Testing Availability	yes
Non-Molecular Test	skeletal survey ¹
Disease Frequency (per 100,000)	0.4 ¹
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/books/NBK1127/#clcn7.Summary>

² <https://esid.org/layout/set/print/content/view/full/14267>