

Disease

X-linked lymphoproliferative syndrome 1^{1, 2}

Gene

SH2D1A

Inheritance	XL
Current Treatments	Emapalumab ,Hematopoietic stem cell transplantation (HSCT) - bone marrow transplant
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	invariant natural killer T-cell quantitation ¹
Disease Frequency (per 100,000)	0.1 ¹
Genes Included in Disease Frequency	SH2D1A and XIAP
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=SH2D1A&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/books/NBK1406/#x-lpd.Summary>

² <https://pubmed.ncbi.nlm.nih.gov/32374962/>