

Disease

X-linked immunodeficiency with hyper-IgM type 1¹

Gene

CD40LG

Inheritance	XL
Current Treatments	Hematopoietic Stem Cell Transplantation (HSCT) Bone marrow transplant. Prophylaxis for pneumonia secondary to Pneumocystis jiroveci. immunoglobulin replacement
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	immunoglobulin levels, flow cytometric analysis ²
Disease Frequency (per 100,000)	0.1 ³
Genes Included in Disease Frequency	CD40LG
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Hyper+IgM&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/books/NBK1402/#xlhi.Summary>

² <https://www.ncbi.nlm.nih.gov/books/NBK1402/>

³ <https://www.ncbi.nlm.nih.gov/pubmed/14663287>