

Disease

Immunodeficiency 76^{1, 2}

Gene

FCHO1

Inheritance	AR
Current Treatments	Hematopoietic stem cell transplantation (HSCT) - bone marrow transplant
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	T and B Lymphocyte and Natural Killer Cell Profile, immunoglobulin levels ²
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=fcho1&term=&cntry=&state=&city=&dist=

References

¹ <https://omim.org/entry/619164>

² <https://pubmed.ncbi.nlm.nih.gov/33950325/>