

Disease

Immunodeficiency 24¹

Gene

CTPS1

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 ²
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=CTPS1&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/pubmed/24870241>

² <https://pubmed.ncbi.nlm.nih.gov/32161190/>