

Disease

B-cell immunodeficiency, distal limb anomalies, and urogenital malformations¹

Gene

TOP2B

Inheritance	AD
Current Treatments	replacement immunoglobulin treatment
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	immunoglobulin levels, T and B Lymphocyte and Natural Killer Cell Profile ²
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=top2b&term=&cntry=&state=&city=&dist=

References

¹ <https://omim.org/entry/609296>

² <https://pubmed.ncbi.nlm.nih.gov/33459963/>