

Disease

Immunodeficiency 14^{1, 2, 3}

Gene

PIK3CD

Inheritance	AR and AD
Current Treatments	Replacement immunoglobulin treatment, rapamycin, leniolisib phosphate (Joenja), Bone marrow transplant (hematopoietic stem cell transplantation (HSCT)) - treatment for AR and AD forms differ
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	immunoglobulin levels, T and B Lymphocyte and Natural Killer Cell Profile ⁴
Disease Frequency (per 100,000)	0.4 ⁵
Genes Included in Disease Frequency	NFKB1, TNFRSF13B, TNFRSF13C, CD19, CD81, MS4A1, CR2, LRBA, PIK3CD, NFKB2, IRF2BP2, ICOS, IL21, IKZF1
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=pik3cd&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/pubmed/30911953>
- ² <https://www.ncbi.nlm.nih.gov/pubmed/31111319>
- ³ <https://pubmed.ncbi.nlm.nih.gov/34033842/>
- ⁴ <https://pubmed.ncbi.nlm.nih.gov/33080915/>
- ⁵ <https://ghr.nlm.nih.gov/condition/common-variable-immune-deficiency#genes>