

Disease

Agammaglobulinemia 7^{1, 2, 3}

Gene

PIK3R1

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	no
Disease Frequency (per 100,000)	0.75 ⁴
Genes Included in Disease Frequency	BTK, IGHM, IGLL1, CD79A, CD79B, BLNK, PIK3R1, TCF3
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Activated+PI3Kdelta+Syndrome&term=&cntry=&state=&city=&rank=

References

- ¹ <https://www.ncbi.nlm.nih.gov/pubmed/31111319>
- ² <https://pubmed.ncbi.nlm.nih.gov/33401995/>
- ³ <https://pubmed.ncbi.nlm.nih.gov/34033842/>
- ⁴ <https://www.ncbi.nlm.nih.gov/pubmed/30359632>