

Disease

Griscelli syndrome, type 2^{1, 2}

Gene

RAB27A

Inheritance	AR
Current Treatments	Emapalumab ,Hematopoietic stem cell transplantation (HSCT) - bone marrow transplant
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	natural killer cell activity, cytotoxic T lymphocyte activity ¹
Disease Frequency (per 100,000)	2 ³
Genes Included in Disease Frequency	PRF1, UNC13D, STX11, STXBP2, RAB27A
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Griscelli+Syndrome&term=&cntry=&state=&city=&dist=

References

- ¹ <https://pubmed.ncbi.nlm.nih.gov/32107531/>
- ² <https://pubmed.ncbi.nlm.nih.gov/32374962/>
- ³ <https://www.ncbi.nlm.nih.gov/books/NBK1444/#top>