

Disease

Hermansky-Pudlak syndrome 2^{1, 2, 3}

Gene

AP3B1

Inheritance	AR
Current Treatments	Hematopoietic Stem Cell Transplantation (HSCT) Bone marrow transplant, granulocyte-colony stimulating factor (G-CSF)
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	natural killer cell activity, cytotoxic T lymphocyte activity, ⁴
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Hermansky+Pudlak+Syndrome&term=&cntry=&state=&city=&d

References

¹ <https://pubmed.ncbi.nlm.nih.gov/31820501/>

² <https://omim.org/entry/608233>

³ <https://www.ncbi.nlm.nih.gov/books/NBK1287/>

⁴ <https://pubmed.ncbi.nlm.nih.gov/32107531/>