

Disease

Hermansky-Pudlak syndrome 10^{1, 2}

Gene

AP3D1

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	NK- and T-cell degranulation ³
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/30472485/>

² <https://pubmed.ncbi.nlm.nih.gov/20301464/>

³ <https://www.omim.org/entry/617050#1>