

Disease

Neutropenia, severe congenital, 8, autosomal dominant^{1, 2}

Gene

SRP54

Inheritance	AD
Current Treatments	granulocyte-colony stimulating factor (G-CSF) therapy, oral pancreatic enzymes, fat-soluble vitamins, blood and/or platelet transfusions, granulocyte-colony stimulation factor, Hematopoietic Stem Cell Transplantation (HSCT) Bone marrow transplant
Evidence for Treatment	guideline ³
Non-Molecular Testing Availability	no
Disease Frequency (per 100,000)	1.3 ⁴
Genes Included in Disease Frequency	DNAJC21, EFL1, SBDS, SRP54
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Shwachman-diamond+Syndrome&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK1756/>
- ² <https://www.ncbi.nlm.nih.gov/pubmed/29914977>
- ³ <https://pubmed.ncbi.nlm.nih.gov/22191555/>
- ⁴ <https://www.ncbi.nlm.nih.gov/books/NBK1756/#sds.Summary>