Rx-Genes
Treatment Guidance at the Point of Care

Disease

Shwachman-Diamond syndrome[1]

Gene

SBDS

Inheritance	AR
Current Treatments	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[2]
Non-Molecular Testing Availability	no
Disease Frequency (per 100,000)	1.3[3]
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://pubmed.ncbi.nlm.nih.gov/22191555/

[3] https://www.ncbi.nlm.nih.gov/books/NBK1756/#sds.Summary