

Disease

Ataxia-pancytopenia syndrome^{1, 2, 3}

Gene

SAMD9L

Inheritance	AD
Current Treatments	Hematopoietic stem cell transplantation (HSCT) - bone marrow transplant
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	no
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=SAMD9L&term=&cntry=&state=&city=&dist=

References

- ¹ <https://omim.org/entry/159550>
- ² <https://pubmed.ncbi.nlm.nih.gov/31306780/>
- ³ <https://pubmed.ncbi.nlm.nih.gov/36880537/>