

Disease

Bone marrow failure syndrome 4¹

Gene

MYSM1

Inheritance	AR
Current Treatments	Hematopoietic stem cell transplantation (HSCT) - bone marrow transplant
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	WBC and RBC counts. immunoglobulin level, T and B Lymphocyte and Natural Killer Cell Profile ²
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Bone+Marrow+Failure+Syndrome&term=&cntry=&state=&city=

References

¹ <https://omim.org/entry/618116>

² <https://pubmed.ncbi.nlm.nih.gov/32640305/>