

Disease

Radioulnar synostosis with amegakaryocytic thrombocytopenia 2^{1, 2}

Gene

MECOM

Inheritance	AD
Current Treatments	Hematopoietic stem cell transplantation (HSCT) - bone marrow transplant
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	Complete blood count, bone marrow aspiration and biopsy ¹
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Amegakaryocytic+Thrombocytopenia&term=&cntry=&state=&city=

References

¹ <https://omim.org/entry/616738>

² <https://pubmed.ncbi.nlm.nih.gov/32064714/>