Rx-Genes
Treatment Guidance at the Point of Care

Disease

Diamond-Blackfan anemia 14 with mandibulofacial dysostosis[1]

Gene

TSR2

Inheritance	XL
Current Treatments	corticosteroids, red blood cell transfusion, hematopoietic stem cell transplantation (HSCT) - bone marrow transplant
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	Complete blood count with reticulocyte count, erythrocyte adenosine deaminase activity, fetal hemoglobin, bone marrow aspiration and biopsy[2]
Disease Frequency (per 100,000)	0.75[3]
Genes Included in Disease Frequency	RPL5, RPL11, RPL35A, RPS10, RPS17, RPS19, RPS24, RPS26, GATA1, RPL15, RPL18, RPL26, RPL27, RPL31, RPL35, RPS7, RPS15A, RPS27,RPS28, RPS29, TSR2
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Diamond+Blackfan+Anemia&term=&cntry=&state=&city=&dist=

References

[1] https://www.ncbi.nlm.nih.gov/pubmed/20301769

[2] https://www.ncbi.nlm.nih.gov/books/NBK7047/

[3] https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3778360/