Rx-Genes
Treatment Guidance at the Point of Care

**Disease**

# Diamond-blackfan anemia 3[1]

**Gene**

## RPS24

| | |
|---|---|
| **Inheritance** | AD |
| **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/