

## Disease

# Diamond-Blackfan anemia 1<sup>1</sup>

## Gene

### RPS19

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

## References

<sup>1</sup> <https://www.ncbi.nlm.nih.gov/pubmed/20301769>

<sup>2</sup> <https://www.ncbi.nlm.nih.gov/books/NBK7047/>

<sup>3</sup> <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3778360/>