

## Disease

# Bone marrow failure syndrome 3<sup>1</sup>

## Gene

### DNAJC21

|                                                 |                                                                                                                                                                                                                                       |
|-------------------------------------------------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Inheritance</b>                              | AR                                                                                                                                                                                                                                    |
| <b>Current Treatments</b>                       | oral pancreatic enzymes, fat-soluble vitamins, blood and/or platelet transfusions, granulocyte-colony stimulating factor, Hematopoietic Stem Cell Transplantation (HSCT) Bone marrow transplant                                       |
| <b>Evidence for Treatment</b>                   | guideline <sup>2</sup>                                                                                                                                                                                                                |
| <b>Non-Molecular Testing Availability</b>       | no                                                                                                                                                                                                                                    |
| <b>Disease Frequency (per 100,000)</b>          | 1.3 <sup>3</sup>                                                                                                                                                                                                                      |
| <b>Genes Included in Disease Frequency</b>      | DNAJC21, EFL1, SBDS, SRP54                                                                                                                                                                                                            |
| <b>Experimental / Investigational Therapies</b> | <a href="https://clinicaltrials.gov/ct2/results?cond=Bone+Marrow+Failure+Syndromes%2C+Inherited&amp;term=&amp;cntry=">https://clinicaltrials.gov/ct2/results?cond=Bone+Marrow+Failure+Syndromes%2C+Inherited&amp;term=&amp;cntry=</a> |

## References

- <sup>1</sup> <https://www.ncbi.nlm.nih.gov/books/NBK1756/>
- <sup>2</sup> <https://pubmed.ncbi.nlm.nih.gov/22191555/>
- <sup>3</sup> <https://www.ncbi.nlm.nih.gov/books/NBK1756/#sds.Summary>