

**Disease**

# Periodic fever, immunodeficiency, and thrombocytopenia syndrome<sup>1, 2</sup>

**Gene**

## WDR1

|                                                 |                                                                                                                                                                                                                         |
|-------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Inheritance</b>                              | AR                                                                                                                                                                                                                      |
| <b>Current Treatments</b>                       | Bone marrow transplant (hematopoietic stem cell transplantation (HSCT))                                                                                                                                                 |
| <b>Evidence for Treatment</b>                   | expert opinion                                                                                                                                                                                                          |
| <b>Non-Molecular Testing Availability</b>       | yes                                                                                                                                                                                                                     |
| <b>Non-Molecular Test</b>                       | T and B Lymphocyte and Natural Killer Cell Profile, peripheral smear <sup>3</sup>                                                                                                                                       |
| <b>Experimental / Investigational Therapies</b> | <a href="https://clinicaltrials.gov/ct2/results?cond=wdr1&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=">https://clinicaltrials.gov/ct2/results?cond=wdr1&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=</a> |

**References**

<sup>1</sup> <https://omim.org/entry/150550>

<sup>2</sup> <https://pubmed.ncbi.nlm.nih.gov/32960541/>

<sup>3</sup> <https://pubmed.ncbi.nlm.nih.gov/29751004/>