

**Disease**

# Poikiloderma with neutropenia<sup>1, 2</sup>

**Gene**

## USB1

|                                                 |                                                                                                                                                                                     |
|-------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Inheritance</b>                              | AR                                                                                                                                                                                  |
| <b>Current Treatments</b>                       | Granulocyte-Colony Stimulating Factor (GCSF)                                                                                                                                        |
| <b>Evidence for Treatment</b>                   | expert opinion                                                                                                                                                                      |
| <b>Non-Molecular Testing Availability</b>       | yes                                                                                                                                                                                 |
| <b>Non-Molecular Test</b>                       | complete blood count <sup>1</sup>                                                                                                                                                   |
| <b>Experimental / Investigational Therapies</b> | <a href="https://clinicaltrials.gov/search?cond=Congenital%20Neutropenia&amp;viewType=Table">https://clinicaltrials.gov/search?cond=Congenital%20Neutropenia&amp;viewType=Table</a> |

**References**

<sup>1</sup> <https://omim.org/entry/604173>

<sup>2</sup> <https://pubmed.ncbi.nlm.nih.gov/28353165/>