

Disease

## Pseudo-TORCH syndrome 2<sup>1, 2</sup>

Gene

### USP18

|                                                 |                                                                                                                                                                                                                           |
|-------------------------------------------------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Inheritance</b>                              | AR                                                                                                                                                                                                                        |
| <b>Current Treatments</b>                       | Ruxolitinib                                                                                                                                                                                                               |
| <b>Evidence for Treatment</b>                   | expert opinion                                                                                                                                                                                                            |
| <b>Non-Molecular Testing Availability</b>       | yes                                                                                                                                                                                                                       |
| <b>Non-Molecular Test</b>                       | Interferon signature <sup>3</sup>                                                                                                                                                                                         |
| <b>Experimental / Investigational Therapies</b> | <a href="https://clinicaltrials.gov/ct2/results?cond=USP18&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=">https://clinicaltrials.gov/ct2/results?cond=USP18&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=</a> |

References

<sup>1</sup> <https://www.ncbi.nlm.nih.gov/pubmed/31940699>

<sup>2</sup> <https://omim.org/entry/617397>

<sup>3</sup> <https://pubmed.ncbi.nlm.nih.gov/31940699/>