

Disease

# Cryopyrin associated periodic fever syndrome<sup>1, 2</sup>

Gene

## NLRP3

|                                          |                                                                                                                                                                                                                                   |
|------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Inheritance                              | AD                                                                                                                                                                                                                                |
| Current Treatments                       | Corticosteroids, anakinra, rilonacept and canakinumab                                                                                                                                                                             |
| Evidence for Treatment                   | guideline <sup>3</sup>                                                                                                                                                                                                            |
| Non-Molecular Testing Availability       | no                                                                                                                                                                                                                                |
| Disease Frequency (per 100,000)          | 0.19 <sup>4</sup>                                                                                                                                                                                                                 |
| Genes Included in Disease Frequency      | NLRP3                                                                                                                                                                                                                             |
| Experimental / Investigational Therapies | <a href="https://clinicaltrials.gov/ct2/results?cond=cryopyrin&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=">https://clinicaltrials.gov/ct2/results?cond=cryopyrin&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=</a> |

References

- <sup>1</sup> <https://www.ncbi.nlm.nih.gov/pubmed/31858722>
- <sup>2</sup> <https://www.ncbi.nlm.nih.gov/pubmed/30175395>
- <sup>3</sup> <https://pubmed.ncbi.nlm.nih.gov/32066461/>
- <sup>4</sup> <https://www.ncbi.nlm.nih.gov/pubmed/22527214>