

Disease

# Hereditary nonpolyposis colorectal cancer-4<sup>1, 2</sup>

Gene

## PMS2

|                                                 |                                                                                                                                                                                                                                                                                           |
|-------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Inheritance</b>                              | AD                                                                                                                                                                                                                                                                                        |
| <b>Current Treatments</b>                       | colonoscopy - polypectomy, aspirin                                                                                                                                                                                                                                                        |
| <b>Evidence for Treatment</b>                   | guideline <sup>2</sup>                                                                                                                                                                                                                                                                    |
| <b>Non-Molecular Testing Availability</b>       | no                                                                                                                                                                                                                                                                                        |
| <b>ClinGen</b>                                  | <a href="https://actionability.clinicalgenome.org/ac/Adult/ui/stg2SummaryRpt?doc=AC069">https://actionability.clinicalgenome.org/ac/Adult/ui/stg2SummaryRpt?doc=AC069</a>                                                                                                                 |
| <b>Disease Frequency (per 100,000)</b>          | 160 <sup>3, 4</sup>                                                                                                                                                                                                                                                                       |
| <b>Genes Included in Disease Frequency</b>      | MLH1, MSH2, MSH6, PMS2, EPCAM                                                                                                                                                                                                                                                             |
| <b>Experimental / Investigational Therapies</b> | <a href="https://clinicaltrials.gov/ct2/results?cond=Hereditary+Non-polyposis+Colon+Cancer&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=">https://clinicaltrials.gov/ct2/results?cond=Hereditary+Non-polyposis+Colon+Cancer&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=</a> |

References

- <sup>1</sup> <https://www.ncbi.nlm.nih.gov/books/NBK1211/>
- <sup>2</sup> [https://www.nccn.org/professionals/physician\\_gls/pdf/genetics\\_colon.pdf](https://www.nccn.org/professionals/physician_gls/pdf/genetics_colon.pdf)
- <sup>3</sup> <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3566622/>
- <sup>4</sup> <https://www.nature.com/articles/ncomms14755>