

# RPS20 gene

## Associated Syndrome Name: RPS20-associated Cancer Risk

### RPS20 Summary Cancer Risk Table

CANCER	GENETIC CANCER RISK
Colorectal	Elevated Risk

### RPS20 gene Overview

#### RPS20-associated Cancer Risk<sup>1, 2, 3</sup>

- Mutations in *RPS20* have been found in two families which include many individuals who have had colorectal cancer. Additional *RPS20* mutations have been found in a small number of individuals suspected of having an inherited risk for colorectal cancer.
- Based on the information from the Finnish family, and the small number of individual cases, there is some evidence that individuals with *RPS20* mutations have a significantly increased risk for colorectal cancer. The exact size of this risk is unknown, and it is not certain that this risk applies to all *RPS20* mutations.
- There are currently no guidelines for the medical management of individuals with mutations in *RPS20*. Since information about the cancer risks associated with *RPS20* mutations is relatively new, and there is uncertainty about the best ways to reduce these risks, it may be appropriate to interpret these results in consultation with cancer genetics professionals who have expertise in this emerging area of knowledge.

### RPS20 gene Cancer Risk Table

CANCER TYPE	AGE RANGE	CANCER RISK	RISK FOR GENERAL POPULATION
Colorectal	To age 80 <sup>2, 3, 4</sup>	Elevated risk	2.8%

### RPS20 Cancer Risk Management Table

The overview of medical management options provided is a summary of professional society guidelines. The most recent version of each guideline should be consulted for more detailed and up-to-date information before developing a treatment plan for a particular patient.

This overview is provided for informational purposes only and does not constitute a recommendation. While the medical society guidelines summarized herein provide important and useful information, medical management decisions for any particular patient should be made in consultation between that patient and his or her healthcare provider and may differ from society guidelines based on a complete understanding of the patient's personal medical history, surgeries and other treatments.

CANCER TYPE	PROCEDURE	AGE TO BEGIN	FREQUENCY (UNLESS OTHERWISE INDICATED BY FINDINGS)
Colorectal	Currently there are no medical management guidelines for colorectal cancer risk in mutation carriers. However, the possibility of an increased risk for colorectal cancer may warrant consideration of individualized risk-reduction strategies, such as the modification of standard population screening recommendations by starting screening at younger ages and/or performing screenings at greater frequency. <sup>5</sup>	NA	NA

### Information for Family Members

The following information for Family Members will appear as part of the MMT for a patient found to have a mutation in the *RPS20* gene.

This patient's relatives are at risk for carrying the same mutation(s) and associated cancer risks as this patient. Cancer risks for females and males who have this/these mutation(s) are provided below.

Family members should talk to a healthcare provider about genetic testing. Close relatives such as parents, children, brothers and sisters have the highest chance of having the same mutation(s) as this patient. Other more distant relatives such as cousins, aunts, uncles, and grandparents also have a chance of carrying the same mutation(s). Testing of at-risk relatives can identify those family members with the same mutation(s) who may benefit from surveillance and early intervention.

## References

1. Thompson BA, et al. A novel ribosomal protein S20 variant in a family with unexplained colorectal cancer and polyposis. Clin Genet. 2020 97:943-944. PMID: 32424863.
2. Broderick P, et al. Validation of Recently Proposed Colorectal Cancer susceptibility Gene Variants in an Analysis of Families and Patients-a Systematic Review. Gastroenterology. 2017 152:75-77. PMID: 27713038.
3. Nieminen TT, et al. Germline mutation of *RPS20*, encoding a ribosomal protein, causes predisposition to hereditary nonpolyposis colorectal carcinoma without DNA mismatch repair deficiency. Gastroenterology. 2014 147:595-598. PMID: 24941021.
4. SEER\*Explorer: An interactive website for SEER cancer statistics [Internet]. Surveillance Research Program, National Cancer Institute. [Cited 2023 Mar 24]. Available from <https://seer.cancer.gov/explorer/>.
5. Ness R, et al. NCCN Clinical Practice Guidelines in Oncology<sup>®</sup> Colorectal Cancer Screening. V 3.2022. Sep 30. Available at <https://www.nccn.org>.

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