AXIN2 gene

Associated Syndrome Name: *AXIN2*-associated cancer risk

AXIN2 Summary Cancer Risk Table

CANCER	GENETIC CANCER RISK
Colorectal	Elevated Risk

AXIN2 gene Overview

AXIN2-associated cancer risk [1, 2, 3, 4, 5]

- Mutations in *AXIN2* have been found in families where many individuals have large numbers of colorectal polyps and/or colorectal cancer. Individuals with *AXIN2* mutations are believed to have a significantly increased risk for colorectal cancer, but the exact risk is not known.
- The colorectal polyps in individuals with *AXIN2* mutations may be of different types, including adenomas and hyperplastic polyps.
- Individuals with *AXIN2* mutations may also have missing adult teeth (oligodontia) and reduced amounts of hair on the skin and scalp.
- Although there is an increased risk for colorectal cancer in individuals with mutations in *AXIN2*, it may be possible to reduce this risk with appropriate medical management. Guidelines for the medical management of patients with *AXIN2* mutations have been developed by the National Comprehensive Cancer Network (NCCN). These are listed below. These guidelines will evolve as we learn more about individuals with *AXIN2* mutations, and it is recommended that patients with a mutation be managed by a multidisciplinary team with expertise in medical genetics and the care of patients with hereditary cancer syndromes.

AXIN2 gene Cancer Risk Table

CANCER TYPE	AGE RANGE	CANCER RISK	RISK FOR GENERAL POPULATION
Colorectal	To age 80[2, 3, 6]	Elevated risk	2.8%

AXIN2 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	Colonoscopy[4]	25 to 30 years	Every 2 to 3 years
	Colorectal surgical evaluation and counseling.[4]	Based on cancer diagnosis and/or polyp number, size and histology	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 AXIN2 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. Marvin ML, Mazzoni SM, Herron CM, Edwards S, Gruber SB, et al. *AXIN2*-associated autosomal dominant ectodermal dysplasia and neoplastic syndrome. Am J Med Genet A. 2011 155A:898-902. PMID: 21416598.

2. Lammi L, et al. Mutations in *AXIN2* cause familial tooth agenesis and predispose to colorectal cancer. Am J Hum Genet. 2004 74:1043-50. PMID: 15042511.

3. Rohlin A, et al. Expanding the genotype-phenotype spectrum in hereditary colorectal cancer by gene panel testing. Fam Cancer. 2017 16):195-203. PMID: 27696107.

4. Gupta S, et al. NCCN Clinical Practice Guidelines in Oncology® Genetic/Familial High-Risk Assessment: Colorectal, Endometrial, and Gastric. V 1.2025. Jun 13. Available at https://www.nccn.org.

5. Rivera B, et al. A novel *AXIN2* germline variant associated with attenuated FAP without signs of oligondontia or ectodermal dysplasia. Eur J Hum Genet. 2014 22:423-6. PMID: 23838596.

6. SEER*Explorer: An interactive website for SEER cancer statistics [Internet]. Surveillance Research Program, National Cancer Institute. [Cited 2025 Aug 12]. Available from https://seer.cancer.gov/explorer/.

Last Updated on 10-Mar-2026