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AXIN2 Mutations

Cancer Risks and General Management Recommendations

Individuals with a pathogenic variant (mutation) in *AXIN2* have an autosomal dominant condition called oligodontia-colorectal cancer syndrome. Pathogenic variants in *AXIN2* can cause oligodontia (the absence of six or more permanent teeth).[1,2] Individuals may also have features of ectodermal dysplasia including sparse eyebrows, scalp, and body hair.[3] We encourage patients to discuss appropriate treatment and management concerns with their dentist and other medical providers.

Recent evidence also suggests that individuals with *AXIN2* pathogenic variants are predisposed to developing adult-onset colon adenomas, polyps, and colorectal cancer.[1,3-5]

***AXIN2* Mutation Carrier Cancer Risks**	**General Population Lifetime Cancer Risks**	**Surveillance/Management Recommendations[6]**

Colon Cancer[1,3-5]
Increased (lifetime risk not established)
	4.5%	• Begin colonoscopy at age 25-30.
		  ○ Repeat every 2-3 years if no polyps are found.
		  ○ Repeat every 1-2 years if polyps are found.
		• Surgical evaluation is recommended if appropriate. Consider surgery if the polyp burden becomes unmanageable by colonoscopy.

Implications for Family Members/ Reproductive Considerations

- First-degree relatives (i.e., parents, siblings, and children) have a 50% chance to have the familial *AXIN2* mutation. Second-degree relatives (i.e., nieces/nephews, aunts/uncles, and grandparents) have a 25% chance to have the familial mutation.
- For carriers of a known mutation, assisted reproduction (with or without egg or sperm donation), pre-implantation genetic testing, and prenatal diagnosis options exist.
- All family members are encouraged to pursue genetic counseling to clarify their risks. Family members can visit www.FindAGeneticCounselor.com to find genetic services near them.

References

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