

MITF gene

Associated Syndrome Name: MITF-associated cancer risk

MITF Summary Cancer Risk Table

CANCER	GENETIC CANCER RISK
Skin	Elevated Risk

MITF gene Overview

MITF-associated cancer risk^{1,2,3}

- Individuals with the *MITF* variant identified in this patient have an increased risk for melanoma. Although there is no exact estimate of this risk, it is believed to be significantly increased over that in the general population.
- Individuals with this *MITF* variant are likely to have dozens to hundreds of moles (nevi).
- There have been some studies suggesting that individuals with the *MITF* variant identified in this patient have an increased risk for renal (kidney) cancer. However, these studies are not conclusive and there are currently no medical management recommendations to address this possible risk.
- Other types of variants in the *MITF* gene cause conditions that can include albinism, or absence of pigment in the skin, hair, and eyes, as well as hearing loss. The variant found in this patient is not believed to be associated with these conditions.
- Although there are increased cancer risks for patients with the *MITF* variant found in this patient there may be interventions that can reduce these risks. General recommendations for melanoma prevention from the National Comprehensive Cancer Network (NCCN) and the American Society of Clinical Oncology (ASCO) are listed below. Since information about the cancer risks associated with this *MITF* variant 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.

MITF gene Cancer Risk Table

CANCER TYPE	AGE RANGE	CANCER RISK	RISK FOR GENERAL POPULATION
Melanoma	To age 80 ^{1,2,3,4}	Elevated risk	1.6%

MITF 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)
Melanoma	Currently there are no specific medical management recommendations for melanoma risk in mutation carriers. However, the increased risk for melanoma warrants consideration of risk-reduction strategies, including frequent self-examination of the skin, consideration of clinical skin examinations, and minimizing exposure to the sun and other sources of UV radiation. ^{5,6,7}	Individualized	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 MITF 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. Potrony M, et al. Prevalence of *MITF* p.E318K in Patients With Melanoma Independent of the Presence of *CDKN2A* Causative Mutations. *JAMA Dermatol*. 2016 152:405-12. PMID: 26650189.
2. Yokoyama S, et al. A novel recurrent mutation in *MITF* predisposes to familial and sporadic melanoma. *Nature*. 2011 480:99-103. PMID: 22080950.
3. Bertolotto C, et al. A SUMOylation-defective *MITF* germline mutation predisposes to melanoma and renal carcinoma. *Nature*. 2011 480:94-8. PMID: 22012259.
4. 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/>.
5. Swetter SM, et al. NCCN Clinical Practice Guidelines in Oncology®: Melanoma: Cutaneous. V 2.2025 Jan 28. Available at <https://www.nccn.org>.
6. National Cancer Institute. Skin Cancer (Including Melanoma)-Patient Version. Accessed 2025 Apr 1. Available at <https://www.cancer.gov/types/skin>.
7. American Cancer Society. Risk Factors for Melanoma Skin Cancer. Updated 2023 Oct 27. Available at <https://www.cancer.org/cancer/types/melanoma-skin-cancer/causes-risks-prevention/risk-factors.html>.

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