TERT gene

Associated Syndrome Name: *TERT*-associated cancer risk

TERT Summary Cancer Risk Table

CANCER	GENETIC CANCER RISK
Skin	Elevated Risk

TERT gene Overview

TERT-associated cancer risk [1, 2, 3]

- Individuals with the *TERT* variant found in this patient have *TERT*-associated cancer risk.
- Individuals with this *TERT* variant have been reported in two unrelated families including many individuals who have had melanoma. Based on the information from these cases, there is evidence that individuals with this *TERT* variant have a significantly increased risk for melanoma, including at young ages and non-UV-exposed sites. The exact size of this risk is unknown.
- Studies have suggested that individuals with this *TERT* variant could also have an increased risk for other cancers. However, these studies are not conclusive and there are currently no medical management guidelines to address these possible risks.
- Other pathogenic variants in the *TERT* gene are associated with dyskeratosis congenita and idiopathic pulmonary fibrosis. The variant found in this patient does not cause these conditions.
- Although there are increased cancer risks for patients with the *TERT* 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 *TERT* mutations is relatively new, and there is uncertainty about the best way 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.

TERT gene Cancer Risk Table

CANCER TYPE	AGE RANGE	CANCER RISK	RISK FOR GENERAL POPULATION
Melanoma	To age 80[2, 3, 4]	Elevated risk	1.6%

TERT 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 *TERT* 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

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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. Cancer.Net, American Society of Clinical Oncology, Melanoma: Risk Factors and Prevention 12/2021 Available at http://www.cancer.net/cancer-types/melanoma/risk-factors-and-prevention.
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7. Swetter SM, et al. NCCN Clinical Practice Guidelines in Oncology®: Melanoma: Cutaneous. V 2.2023 Mar 10. Available at https://www.nccn.org.

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