PDGFRA gene

Associated Syndrome Name: *PDGFRA*-associated cancer risk

PDGFRA Summary Cancer Risk Table

CANCER	GENETIC CANCER RISK
Other	Elevated Risk

PDGFRA gene Overview

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

- Mutations in the *PDGFRA* gene have been identified in a few families with gastrointestinal stromal tumors (GIST).
- Based on these individual cases, there is some evidence that individuals with *PDGFRA* mutations have an increased risk for GIST. The exact size of this risk is unknown.
- Some individuals with *PDGFRA* mutations have been noted to have coarse facial features and large hands or feet.
- Non-cancerous growths (e.g. inflammatory fibroid polyps, fibroid tumors) in the gastrointestinal tract have been reported in some individuals with a *PDGFRA* mutation.
- There are currently no guidelines for the medical management of individuals with mutations in *PDGFRA*. Since information about the cancer risks associated with *PDGFRA* 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.

PDGFRA gene Cancer Risk Table

CANCER TYPE	AGE RANGE	CANCER RISK	RISK FOR GENERAL POPULATION
Gastrointestinal Stromal Tumors (GIST)	To age 80[1, 2, 3]	Elevated risk	<0.1%

PDGFRA 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)
Gastrointestinal Stromal Tumors (GIST)	Currently there are no medical management guidelines for gastrointestinal stromal tumor risk in mutation carriers. However, it is important to have awareness of and seek medical attention for symptoms such as GI bleeding, obstruction, abdominal pain, early satiety, and fatigue related to anemia.[3]	NA	NA
For Patients With A Cancer Diagnosis	For patients with a *PDGFRA* mutation and a GIST diagnosis, targeted therapies may be available as a treatment option (e.g., tyrosine kinase inhibitors).[3]	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 PDGFRA 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. Postow MA, et al. Inherited gastrointestinal stromal tumor syndromes: mutations, clinical features, and therapeutic implications. Clin Sarcoma Res. 2012 Oct 4;2(1):16. PMID: 23036227.

2. Manley PN, et al. Familial *PDGFRA*-mutation syndrome: somatic and gastrointestinal phenotype. Hum Pathol. 2018 Jun;76:52-57. PMID: 29486293.

3. Von Mehren M, et al. NCCN Clinical Practice Guidelines in Oncology®: Gastrointestinal Stromal Tumors. V 1.2026. Jan 13. Available at https://www.nccn.org.

Last Updated on 15-Sep-2026