Ambry Genetics®

Clinician Management Resource for Germline Pathogenic or Likely Pathogenic *NF1* Variants

This overview of clinical management guidelines is based on this patient's positive test result for a germline pathogenic or likely pathogenic variant in the *NF1* gene. Unless otherwise stated, medical management guidelines used here are limited to those issued by the National Comprehensive Cancer Network® (NCCN®)[1] in the U.S. Please consult the referenced guideline for complete details and further information.

Clinical correlation with the patient's past medical history, treatments, surgeries and family history may lead to changes in clinical management decisions; therefore, other management recommendations may be considered. Genetic testing results and medical society guidelines help inform medical management decisions but do not constitute formal recommendations. Discussions of medical management decisions and individualized treatment plans should be made in consultation between each patient and his or her healthcare provider, and may change over time.

SCREENING/SURGICAL CONSIDERATIONS[1]	AGE TO START	FREQUENCY
Female Breast Cancer		
Breast Screening ▪ Mammography ▪ Consider breast MRI with and without contrast*	30 years old or 5-10 years before the earliest known breast cancer in the family**	Every 12 months, until age 50
Evidence insufficient for risk-reducing mastectomy recommendation. Manage based on family history.	Individualized	N/A
Other		
Recommend referral to neurofibromatosis specialist for evaluation and management of malignant peripheral nerve sheath tumors, GIST, others	Individualized	N/A

* Consider possibility of false-positive MRI results due to presence of breast neurofibromas.

** At this time, there are no data to suggest an increased breast cancer risk after age 50.

1. Referenced with permission from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) for Genetic/Familial High-Risk Assessment: Breast, Ovarian, Pancreatic, and Prostate. v2.2025. © National Comprehensive Cancer Network, Inc. 2024. All rights reserved. Accessed November 7, 2024. To view the most recent and complete version of the guideline, go online to NCCN.org. NCCN makes no warranties of any kind whatsoever regarding their content, use or application and disclaims any responsibility for their application or use in any way.

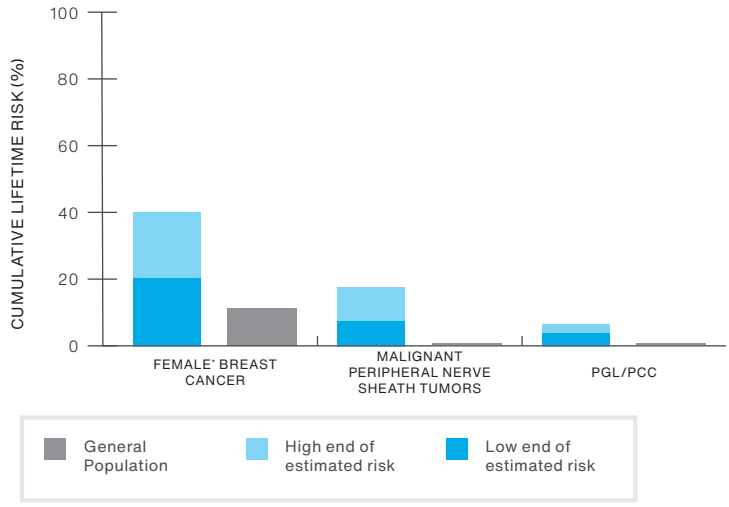
Understanding Your Positive *NF1* Genetic Test Result

INFORMATION FOR PATIENTS WITH A PATHOGENIC OR LIKELY PATHOGENIC VARIANT

5 Things To Know

1	Result	Your testing shows that you have a pathogenic or likely pathogenic variant in the *NF1* gene.
2	Neurofibromatosis type 1	People with a germline pathogenic or likely pathogenic *NF1* variant have neurofibromatosis type 1 (NF1).
3	Cancer risks	You have an increased chance to develop female* breast cancer and possibly other types of cancer such as gastrointestinal stromal tumors (GIST), malignant peripheral nerve sheath tumors (MPNSTs), or paragangliomas and/or pheochromocytomas (PGL/PCC).
4	What you can do	Risk management decisions are very personal. There are options to detect cancer early or lower the risk to develop cancer. It is important to discuss these options with your healthcare provider and decide on a plan that works for you.
5	Family	Up to 50% of the time, a person is born with an *NF1* variant that was not inherited from either parent. Testing family members for the pathogenic or likely pathogenic *NF1* variant found in you could help to determine who in your family may or may not be at increased risk. It is recommended that you share this information with your family members so they can learn more and discuss with their healthcare providers.

NF1 Cancer Risks**



* Refers to sex assigned at birth

** Risk estimates in this graph are only applicable to people with germline pathogenic or likely pathogenic variants in the *NF1* gene. Because risk estimates vary in different studies, only approximate risks are given. Cancer risks will differ based on individual and family history.

Germline *NF1* Variants in the Family^

There is up to a 50/50 random chance to pass on the pathogenic or likely pathogenic *NF1* variant to each of your children.

^ People with somatic *NF1* variants cannot pass them on to their children.

RESOURCES

- Bright Pink brightpink.org
- Children's Tumor Foundation ctf.org
- Imerman Angels imermanangels.org
- Neurofibromatosis Network nfnetwork.org
- Susan G. Komen Foundation komen.org
- National Society of Genetic Counselors nsgc.org
- Canadian Society of Genetic Counsellors cagc-accg.ca

Please discuss this information with your healthcare provider. The cancer genetics field is continuously evolving, so updates related to your *NF1* result, medical recommendations, and/or potential treatments may be available over time. This information is not meant to replace a discussion with a healthcare provider, and should not be considered or interpreted as medical advice.

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MKT-ONCO-FLYR-10027-EN v3 01.30.25