EGFR gene

Associated Syndrome Name: *EGFR*-associated cancer risk

Important Note: The information below applies only to individuals known to have *EGFR* mutations that are germline in nature (mutations present in all cells of the body) which are typically inherited. *EGFR* mutations are commonly detected in tumors (somatic) and assist with treatment decisions in people who have a diagnosis of lung cancer. For people with somatic mutations, the information below regarding cancer risks, medical management options and information for family members may not apply.

EGFR Summary Cancer Risk Table

CANCER	GENETIC CANCER RISK
Lung	Elevated Risk

EGFR gene Overview

EGFR-associated cancer risk [1, 2, 3, 4]

- Individuals with *EGFR* mutations have an increased risk for lung cancer (non-small cell lung cancer, NSCLC). The exact risk is not known, but seems to be significantly increased over lung cancer risk in the general population.
- The lung cancer risk appears to be higher in women than in men.
- Although smoking is the most important risk factor for lung cancer in the general population, studies have found that men and women with *EGFR* mutations have a high risk for lung cancer even if they do not smoke. Some studies have even found that the risk for individuals with *EGFR* mutations could be higher in non-smokers compared to smokers. There are reports of families with *EGFR* mutations with lung cancer diagnoses at ages younger than the general population, including under age 50.
- Risk factors may further increase an individual's risk of lung cancer, including age, smoking, radon exposure, occupational exposure, personal history of cancer, family history of lung cancer, lung diseases, or secondhand smoking exposure. Shared decision making between the patient and provider and discussion of the risks and benefits of lung cancer screening is recommended.
- Although there is an increased risk for lung cancer in individuals with *EGFR* mutations, there may be interventions that can reduce this risk. Since information about *EGFR* mutations and lung cancer risk is new, and there is uncertainty about the best way to reduce the risk, it may be appropriate to interpret these results in consultation with lung cancer specialists and cancer genetics professionals who have expertise in this emerging area of knowledge.

EGFR gene Cancer Risk Table

CANCER TYPE	AGE RANGE	CANCER RISK	RISK FOR GENERAL POPULATION
Lung	To age 80[2, 4, 5]	Elevated risk	3.7%

EGFR 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)
Lung	Currently there are no specific medical management guidelines for lung cancer risk in mutation carriers. However, the increased risk may warrant consideration of individualized screening with low dose chest CT scans, as has been considered for individuals at increased risk due to various risk factors (see clinical overview).[1]	NA	NA

CANCER TYPE	PROCEDURE	AGE TO BEGIN	FREQUENCY (UNLESS OTHERWISE INDICATED BY FINDINGS)
For Patients With A Cancer Diagnosis	For patients with a diagnosis of lung cancer, the presence of this EGFR gene mutation may impact decisions about the choice of drugs for therapy.[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 EGFR 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. Wood DE et al. NCCN Clinical Practice Guidelines in Oncology®: Lung Cancer Screening. V 1.2026. Sep 16. Available at https://www.nccn.org.
2. Gazdar A, et al. Hereditary lung cancer syndrome targets never smokers with germline *EGFR* gene T790M mutations. J Thorac Oncol. 2014 9:456-63. PMID: 24736066.
3. Riely GJ et al. NCCN Clinical Practice Guidelines in Oncology®: Non-Small Cell Lung Cancer. V 3.2026. Dec 24. Available at https://www.nccn.org.
4. Yamamoto H, Inherited lung cancer syndromes targeting never smokers. Transl Lung Cancer Res. 2018 7:498-504. PMID: 30225213.
5. 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/.

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