DICER1 gene

Associated Syndrome Name: *DICER1* tumor predisposition

DICER1 Summary Cancer Risk Table

CANCER	GENETIC CANCER RISK
Endocrine	High Risk
Lung	High Risk
Ovarian	High Risk
Other	Elevated Risk
Renal	Elevated Risk

DICER1 gene Overview

DICER1 tumor predisposition [1, 2, 3, 4]

- Individuals with *DICER1* mutations have *DICER1* tumor predisposition.
- Individuals assigned female sex at birth with *DICER1* mutations have a high risk for ovarian cancer, particularly for a subtype called Sertoli-Leydig ovarian cell tumor (SLCT). Most are diagnosed before age 40.
- Individuals with *DICER1* mutations have a high risk for lung cysts and lung cancer, particularly for a subtype called pleuropulmonary blastoma (PPB). Symptoms may include difficulty breathing and typically present in early childhood.
- Individuals with *DICER1* mutations have a high risk for thyroid-related conditions, including goiter, nodules, and cancer.
- Individuals with *DICER1* mutations have an elevated risk for tumors of the kidney, most of which are benign and occur in childhood.
- Other clinical features and tumors have been observed in individuals with *DICER1* mutations, including tumors of the central nervous system, head and neck, and soft tissue. However, further studies are needed to determine if these are conclusively associated with *DICER1* mutations.
- Although there are increased risks for cancer in individuals with *DICER1* mutations, there are interventions that may reduce these risks. Guidelines from the American Association for Cancer Research (AACR) are listed below. Since information about the cancer risks associated with *DICER1* mutations is relatively new, and there is still some uncertainty about the best ways to reduce these risks, it may be appropriate to interpret these results in consultation with cancer genetics experts in this emerging area of knowledge.

DICER1 gene Cancer Risk Table

CANCER TYPE	AGE RANGE	CANCER RISK	RISK FOR GENERAL POPULATION
Ovarian	To age 50[2, 5]	7%	0.2%
Lung	To age 5[2, 5]	18%	<0.1%
Thyroid	To age 80[3, 5]	16%	1.1%
Renal	To age 80[5]	Elevated risk	1.4%
Other - Non-malignant features of *DICER1* tumor predisposition	All ages	*DICER1* tumor predisposition is associated with non-malignant clinical features, some of which may require medical intervention in early childhood.	NA

DICER1 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)
Ovarian	Pelvic ultrasound[4]	From birth	Every 6 months until at least age 40
Lung	Chest X-ray[4]	From birth	Every 6 months until age 8. Every 12 months until age 12.
	Consider chest CT[4]	Once at 3 months and 2.5 years	NA
Thyroid	Thyroid ultrasound[4]	8 years	Every 3 years
Renal	Abdominal ultrasound[4]	From birth	Every 6 months until age 8. Every 12 months until age 12.
Other - Non-malignant features of *DICER1* tumor predisposition	Monitor for symptoms. Additional screenings recommended, including physical, neurological, and eye exams.[4]	Varies	Annually

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 DICER1 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.

Since risks and screening recommendations begin in infancy, genetic testing for children at risk of inheriting a *DICER1* mutation is appropriate.

There are special considerations for pregnant individuals, including third trimester ultrasound screening of the lung and kidneys for fetuses at risk of inheriting a *DICER1* mutation.[4]

References

1. Schultz KAP, et al. *DICER1* Tumor Predisposition. 2014 Apr 24 [Updated 2020 Apr 30]. In: Adam MP, et al., editors. GeneReviews® [Internet]. PMID: 24761742.

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4. Schultz KAP, et al. Update on Pediatric Surveillance Recommendations for PTEN Hamartoma Tumor Syndrome, DICER1-Related Tumor Predisposition, and Tuberous Sclerosis Complex. Clin Cancer Res. 2025 Jan 17;31(2):234-244. PMID: 39540884.

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