UT Southwestern Harold C. Simmons Comprehensive Cancer Center | NCI Designated Comprehensive Cancer Center

DICER1 Mutations

What You Should Know About *DICER1* Mutations

Individuals with a mutation in the *DICER1* gene have an increased lifetime risk for cancerous (malignant) and noncancerous (benign) tumors of the lungs, kidneys, ovaries, eyes, thyroid and nasal passages. Some individuals with a *DICER1* mutation never develop a tumor or a cancer.

Cancer Risks Associated with a *DICER1* Mutation

If an individual has a *DICER1* mutation, their risk to develop a tumor is currently unknown, but is thought to be low. The most commonly seen tumors in individuals with a *DICER1* mutation are pleuropulmonary blastoma (PPB) and cystic nephroma. *DICER1*-related tumors typically develop before the age of 40, with many tumors occurring in childhood.

- Pleuropulmonary Blastoma (PPB): A rare childhood lung tumor that can be benign or malignant.
- Cystic Nephroma and Wilms Tumor: Cystic nephroma is a benign kidney tumor (fluid filled cysts). Individuals with a *DICER1* mutation also have an increased risk of Wilms tumor, a type of cancer that starts in the kidneys.
- Ovarian Sertoli-Leydig Tumors (SLCT): A testosterone-secreting ovarian tumor. Females with these tumors have symptoms that include menstrual irregularities (few to no periods), acne, extra body hair, and deepening of the voice. About 10-30% of SLCTs are cancerous (malignant).
- Ciliary Body Medulloepithelioma: Eye tumors that can be benign or malignant. They may cause redness of the eye, change in iris color, visible masses on the iris, visual impairment, or glaucoma.
- Nasal Chondromesenchymal Hamartoma (NCMH): Benign tumors that grow high inside of the nose.
- Thyroid Goiter, Cysts and Hyperplasia: *DICER1* mutations are associated with an increased risk for thyroid cysts, multi-nodular goiter, and hyperplasia (abnormal growth). These are all benign growths on the thyroid gland.

Risks to Family Members

Mutations in the *DICER1* gene are inherited in an autosomal dominant fashion. This means that children, brothers, sisters, and parents of individuals with a *DICER1* mutation have a 1 in 2 (or 50%) chance of having the mutation as well. Individuals with a *DICER1* mutation may develop one or more of these tumors, or none of the above. Both males and females can inherit a familial *DICER1* mutation and both males and females can pass it on to their children.

Managing Cancer and Tumor Risks

The following recommendations are based on expert consensus guidelines from the 2016 American Association of Cancer Research (AACR).

- Initial chest CT between 3 and 6 months of age, with follow-up determined by initial findings
- Consider 6-month chest radiographs until 8 years of age and annual chest radiograph from age 8 to 12 years
- Consider biannual abdominal ultrasound until age 8 and annually thereafter
- Consider pelvic ultrasound every 1-2 years beginning in early childhood
- Evaluation by an ophthalmologist
- Ear, nose, and throat (ENT) evaluation with nasal endoscopy for persistent symptoms of nasal obstruction
- Consider thyroid ultrasound starting at age 8

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UT Southwestern Harold C. Simmons Comprehensive Cancer Center | 2201 Inwood Rd., Dallas, Texas 75390-9323
Phone: (214) 645-2563 | Fax: (214) 645-2562 | utswmed.org/cancergenetics