**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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