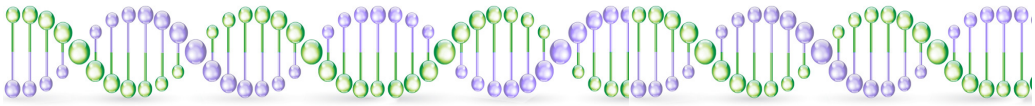
Information Sheet



DICER1 syndrome and the DICER1 gene

DICER1 syndrome is caused by a mutation in the DICER1 gene. It is very rare.

What does the DICER1 gene do?

The DICER1 gene is involved in controlling cell activities. When damaged, specific types of benign and malignant tumours can grow.

The tumours in DICER1 syndrome usually arise from tissues that developed in the embryo but didn't reach their mature state. They are often called blastomas (so... pleuroplumonary blastoma, pineoblastoma, medulloblastoma or pituitary blastoma)

How is DICER1 syndrome diagnosed?

Some of these rare tumours are associated with more than a 50% chance of detecting a germline DICER1 mutation on genetic testing. These include:

- pleuropulmonary blastoma
- cystic nephroma
- Sertoli-Leydig cell tumour of the ovary

Multinodular thyroid disease is also common in DICER1 syndrome although this is also common in the general population.

DICER1 syndrome is diagnosed when a germline DICER1 mutation (pathogenic variant) is detected or when an individual has a combination of rare DICER1-related tumours. For a complete list, see eviQ's Cancer Genetics Paediatric Genetic Testing page.

How is DICER1 syndrome managed?

Most people who carry a DICER1 mutation do not develop tumours or cancers. The likelihood is believed to be 5% by age 10 and 10% by age 20. However, because most of the tumours occur in childhood, screening is recommended to detect the tumours before they can cause problems. Exactly when to start and what to look for is still debated and patients are encouraged to enrol in research studies.

Do DICER1 mutations cause Wilms tumours?

DICER1 mutations have been detected in infants and children with Wilms tumour, a type of blastoma of the kidney. The likelihood of a DICER1 mutation is 1% to 5% in familial Wilms tumour (that is, a family with 2 or more close relatives with Wilms tumours). As a result, under the eviQ guidelines, genetic testing is not recommended in an individual with a personal or family history of Wilms tumour if there no other features of DICER1 syndrome.

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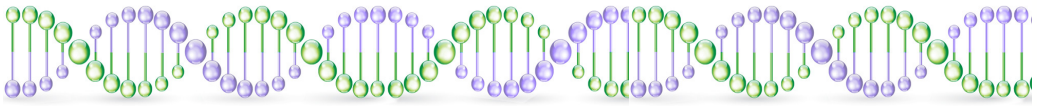

SYDNEY CANCER GENETICS

Is DICER1 syndrome inherited?
Yes. DICER1 is a hereditary cancer syndrome caused by a germline DICER1 mutation. There is a 50% chance of a person who carries a germline DICER1 mutation, whether male or female, passing the mutation to their son or daughter. If a mutation is identified, then predictive testing is available for blood relatives.

Does this sound like you or your family?
Has a rare tumour such as a pleuropulmonary blastoma, cystic nephroma or Sertoli-Leydig cell tumour of the ovary been diagnosed in you or has a DICER1 mutation been detected in a blood relative?

Make an appointment with Dr Hilda High at Sydney Cancer Genetics. It is a confidential opportunity to discuss your personal and family history of cancer and genetic testing can be organised, if needed.

More information is available on our website, including links to the following:

- The DICER1 Syndrome Information Network has easy to read information about this syndrome for families
- The Cancer Genetics section of the Cancer Institute's eviQ website provides up-to-date Australian-based management guidelines
- The International Pleuropulmonary Blastoma (PPB) / DICER1 Registry has information for families and doctors

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