

DICER1-related Tumor Predisposition

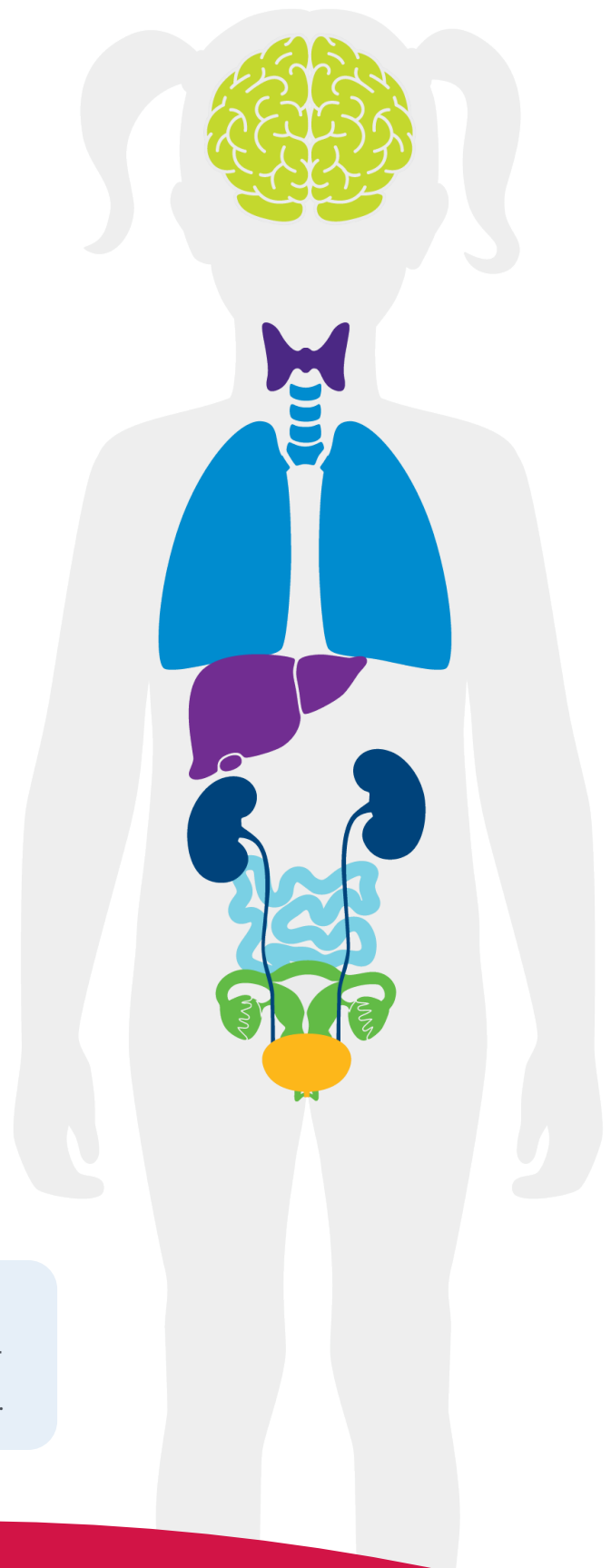
Screening Guidelines for Families

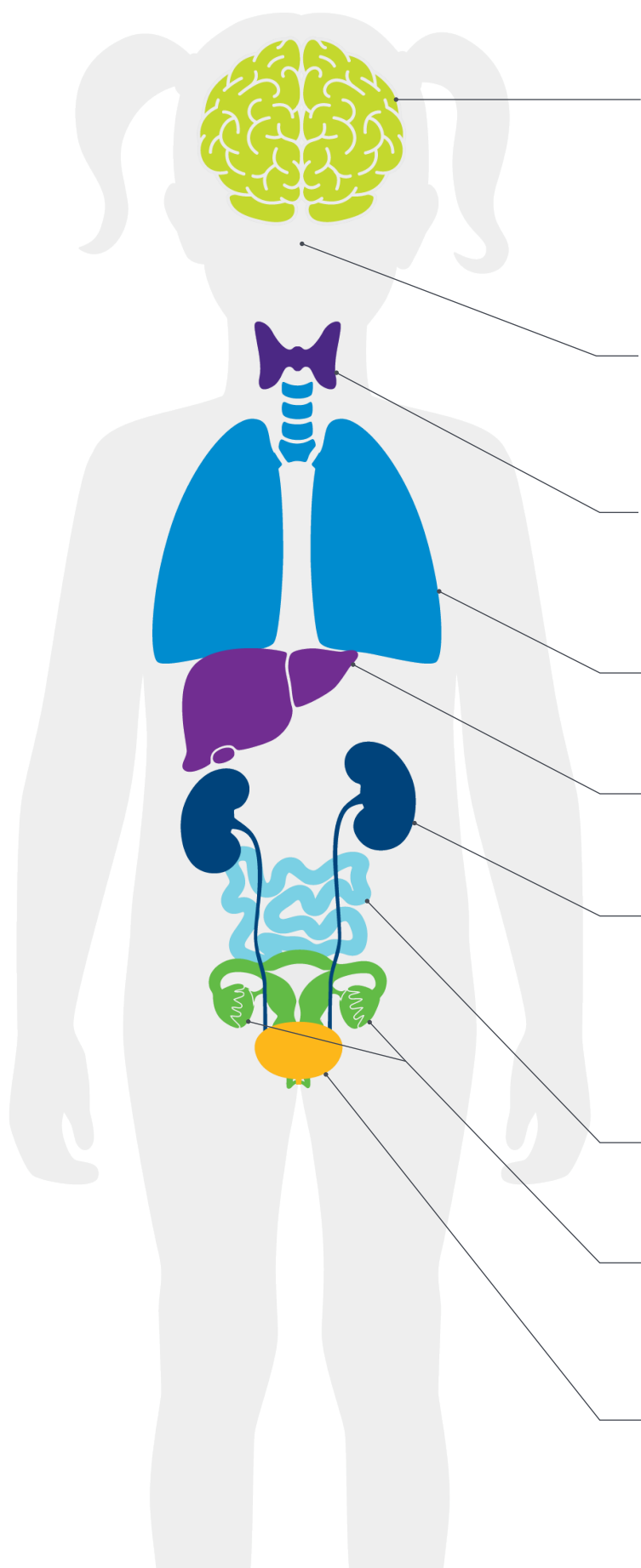
DICER1-related tumor predisposition is a genetic disorder that makes a person more likely to develop certain types of tumors or cancers. Some people will inherit the disorder from a parent. Others will be the first in their family to have it.

- **DICER1-related tumor predisposition is caused by changes in a gene known as *DICER1*.** The *DICER1* gene can prevent tumors or cancers from forming. When the gene does not work correctly, certain tumors and cancers are more likely to develop.
- **Not everyone will develop tumors.** Many people stay healthy. Some may have small health differences, such as lumps in the thyroid or small cysts in the lungs.
- **Less often, people develop tumors.** These tumors may be benign (not cancer) or malignant (cancer).



Screening guidelines are recommendations for how, when, and for how long to check for tumors known to be associated with DICER1.





Certain health conditions are linked to DICER1-related tumor predisposition. Below is a guide:

1 Brain

- **DICER1-associated primary intracranial sarcoma:** A rare cancer in the brain or spinal cord
- **Pineoblastoma:** A tumor in the pineal gland
- **Pituitary blastoma:** A tumor of the pituitary gland
- **Embryonal tumor with multi-layered rosettes:** A tumor of the central nervous system

2 Eyes and nose

- **Ciliary body medulloepithelioma:** An eye tumor
- **Nasal chondromesenchymal hamartoma:** A tumor in the nose

3 Thyroid

- **Multinodular goiter:** Lumps or enlarged areas in the thyroid gland
- **Thyroid cancer:** Cancer of the thyroid gland

4 Lungs

- **Pleuropulmonary blastoma:** A lung cyst or tumor that mostly affects young children

5 Liver

- **Mesenchymal hamartoma of the liver:** A tumor of the liver

6 Kidneys

- **Cystic nephroma:** Fluid-filled cystic tumor in the kidney
- **Anaplastic sarcoma of the kidney:** A malignant (cancer) tumor of the kidney
- **Wilms tumor:** A childhood kidney cancer

7 Gastrointestinal (GI) tract

- **Juvenile hamartomatous intestinal polyps:** Growths in the small intestines

8 Female reproductive system

- **Sertoli-Leydig cell tumor:** A tumor in the ovaries
- **Gynandroblastoma:** A tumor of the ovaries
- **Rhabdomyosarcoma:** A soft-tissue cancer

9 Bladder

- **Rhabdomyosarcoma:** A soft-tissue cancer



Screening for DICER1-related tumor predisposition:

If you know you have DICER1-related tumor predisposition, you can work with your care team to watch for tumors and other health concerns. Your care team may order tests to check for certain signs and symptoms. This helps them catch things early when they are easier to treat.

Call your care team if you notice any new symptoms or anything that worries you.

Signs and symptoms may include:



Brain tumors

- Headache
- Vomiting
- Trouble moving your eyes to look up
- Walking differently or off balance
- Seizures
- Signs of early puberty



Eye and nose tumors

- Changes in vision
- White spot in the eye
- Chronic blocked nose or nostril



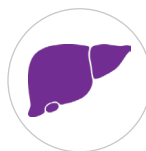
Thyroid tumors

- Swelling or lumps in the neck
- Voice changes
- Trouble swallowing
- Neck pain



Lung tumors

- Cough or difficulty breathing
- Shortness of breath, chest pain



Liver tumors

- Bloated belly area
- Belly pain
- Yellow skin and eyes



Kidney tumors

- Belly or back pain
- Lump in the belly
- Blood in the urine



Gastrointestinal (GI) tract polyps

- Nausea
- Vomiting
- Belly pain
- Bloody stool
- Changes in bowel habits (constipation or diarrhea)



Female reproductive tumors

- Belly pain or swelling
- Early puberty
- Changes in menstrual cycles (heavier bleeding or lighter/missed periods)
- Changes in bowel habits (constipation, diarrhea, bloody stool)

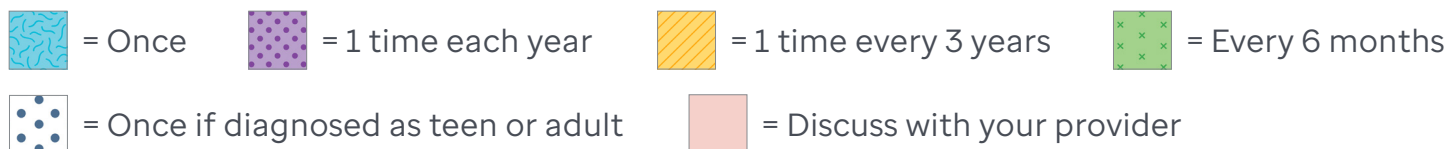


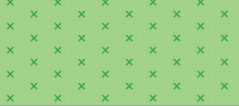
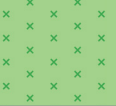
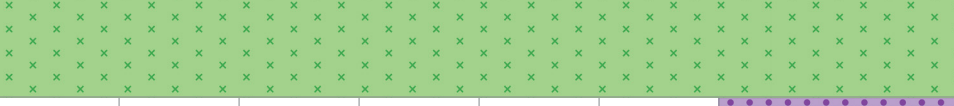
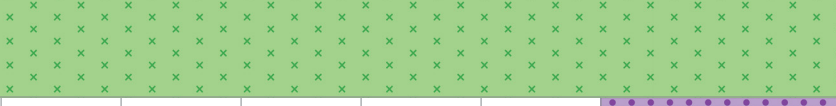
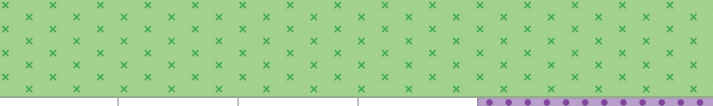
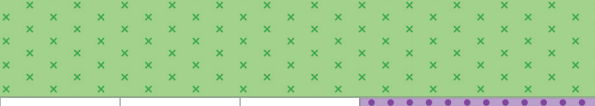
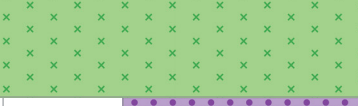
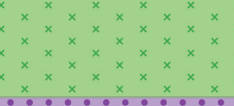
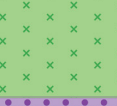
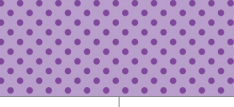
Talk with your care team about the best plan to keep up with your health. If you can, you should get care from a health care provider who is familiar with DICER1-related tumor predisposition care.

DICER1-related tumor predisposition screening recommendations

Children and teens (younger than age 18 years)

This chart outlines how often to do screening tests by age group.



System	Screening test	Birth	3-6 months	2.5-3 years	8 years	10 years	12 years	Teens	Adulthood
Lungs	Chest x-ray								
	Chest CT scan								
Kidneys	Abdominal ultrasound								
Thyroid	Neck exam								
	Thyroid ultrasound								
Female reproductive system	Pelvic ultrasound								
	Gynecology visits								
Eyes	Eye exam								
Brain	Brain MRI	If there is a family history of a DICER1-related brain tumor or concerning signs or symptoms							

Note: These recommendations are based on published guidelines. They were adapted by the Cancer Predisposition clinic team at St. Jude for the patients they care for.

DICER1-related tumor predisposition screening guidelines by age

The tests you need depend on your age and health history. The body systems below are listed in order of when screening may be needed. Those that may need the earliest or most important screening are listed first.

Children and teens (younger than age 18 years)

System	Tumor or other condition	Health care recommendations
Lungs	pleuropulmonary blastoma (PPB), lung cysts, pulmonary blastoma	Chest x-ray: Every 6 months from birth until age 8 years, then 1 time each year until age 12 years <i>If diagnosed with DICER1-related tumor predisposition at age 12 years or older: Chest x-ray at time of diagnosis</i> Chest CT scan (without contrast): Scan at age 3–6 months and another at age 2½–3 years
Kidneys	cystic nephroma, anaplastic sarcoma of the kidney, Wilms tumor	Abdominal ultrasound: Every 6 months from birth to age 8 years, then 1 time each year until age 12 years <i>If diagnosed with DICER1-related tumor predisposition at age 12 years or older: Abdominal ultrasound at time of diagnosis</i>
Thyroid	lumps or cancer in neck	Neck exam by health care provider to check for thyroid changes: 1 time each year Thyroid ultrasound: 1 time every 3 years starting at age 8 years If you get chemotherapy or radiation near the thyroid: An ultrasound at diagnosis and then 1 time each year for 5 years, then every 3 years <i>If the ultrasound shows something: Your health care provider will refer you to endocrinology (endocrine system specialists) for care based on American Thyroid Association (ATA) guidelines.</i>
Female reproductive system	ovarian or uterine tumors	Pelvic ultrasound: Every 6 months starting when DICER1-related tumor predisposition is diagnosed. Repeat sooner if symptoms appear. Gynecology visits: Usually start in the teen years or early 20s
Eyes	ciliary body medulloepithelioma	Dilated eye exam: An ophthalmologist (an eye doctor) will look inside the eye and check vision every year from ages 3 to 10. <i>If DICER1-related tumor predisposition is diagnosed after age 10 years, get a dilated eye exam with a vision check.</i>

Children and teens (younger than age 18 years) *Continued*

System	Tumor or other condition	Health care recommendations
Bladder	embryonal rhabdomyosarcoma	Watch for signs and symptoms. If you have blood in your urine, the health care provider may do an ultrasound and a test to look inside the bladder.
Nose	nasal chondromesenchymal hamartoma	Watch for signs and symptoms. If there are signs or symptoms, your care team may refer you to an ear, nose, and throat (ENT) specialist.
Brain	pituitary blastoma, pineoblastoma, embryonal tumor with multi-layered rosettes, primary intracranial sarcoma	Yearly physical exam. If there is a family history of DICER1-related brain tumor or concerning signs or symptoms: Brain magnetic resonance imaging (MRI) with contrast.
Gastro-intestinal tract	small intestine hamartomas	Watch for signs and symptoms. If there are signs or symptoms, your care team may refer you to a gastrointestinal (GI) specialist.

General care

Physical exam and a **check for signs and symptoms** (if possible by a health care provider familiar with DICER1-related tumor predisposition) each year.

Regular checkup by a pediatrician/family doctor who knows about DICER1-related tumor predisposition. Pay close attention to any medical concerns each year. These may be signs of an underlying tumor.

Note: If there is a concern for DICER1-related tumor predisposition in a patient younger than age 18 years and molecular testing is pending:

- Get a chest x-ray to look for possible pleuropulmonary blastoma (PPB)
- Get a kidney ultrasound to look for cystic nephroma

Adults (age 18 years and older)

The tests you need depend on your age and health history.

System	Tumor or other condition	Health care recommendations
Lungs	lung cysts	Chest x-ray: Done 1 time after DICER1-related tumor predisposition is diagnosed
Kidneys	renal sarcoma	Abdominal ultrasound: Done 1 time after DICER1-related tumor predisposition is diagnosed
Thyroid	nodules, multinodular goiter, carcinoma	Neck exam by health care provider to check for thyroid changes: 1 time each year Thyroid ultrasound: Every 3 years starting at diagnosis If you get chemotherapy or radiation near the thyroid: An ultrasound at diagnosis and then 1 time each year for 5 years, then every 3 years <i>If the ultrasound shows something: Your health care provider will refer you to endocrinology (endocrine system specialists) for care based on American Thyroid Association (ATA) guidelines.</i> If there are signs or symptoms, your care team may refer you to an ear, nose, and throat (ENT) specialist.
Female reproductive system	ovarian Sertoli-Leydig cell tumor; uterine, cervical or vaginal embryonal rhabdomyosarcoma, ovarian sarcoma	Gynecological exam: 1 time each year Pelvic ultrasound: Every 6 months starting when DICER1-related tumor predisposition is diagnosed. Repeat sooner if symptoms appear. Most ovarian tumors happen before age 40. After age 40, talk with your care team about whether to keep doing these ultrasounds.
Eyes	ciliary body medulloepithelioma	Dilated eye exam: Done 1 time after DICER1-related tumor predisposition is diagnosed. Repeat the test if symptoms appear.
Nose	nasal chondromesenchymal hamartoma	Watch for signs and symptoms. If there are signs or symptoms, your care team may refer you to an ear, nose, and throat (ENT) specialist.

Adults (age 18 years and older) Continued

The tests you need depend on your age and health history.

System	Tumor or other condition	Health care recommendations
Brain	pituitary blastoma; pineoblastoma	Watch for signs and symptoms. If there is a family history of DICER1-related brain tumor or concerning signs or symptoms: Brain magnetic resonance imaging (MRI) with contrast

General care

Physical exam and check for signs and symptoms once a year (preferably by a provider familiar with DICER1-related tumor predisposition)

Regular checkup by a family physician who pays close attention to any medical concerns as these may be signs of an underlying tumor

Pregnancy care

You should see a high-risk obstetrician (a doctor who cares for pregnant people).

If you are pregnant and your baby may have DICER1-related tumor predisposition, you should talk to your doctor about extra ultrasounds in the third trimester to check the baby’s lungs and kidneys. If cysts are found, your baby may need a CT scan after birth.

Notes