Retinoblastoma - Childhood - Diagnosis [1]

This section has been reviewed and approved by the Cancer.Net Editorial Board [2], 08/2014

ON THIS PAGE: You will find a list of the common tests, procedures, and scans that doctors can use to find out what?s wrong and identify the cause of the problem. To see other pages, use the menu on the side of your screen.

Doctors use many tests to diagnose cancer and find out if it has spread to another part of the body, called metastasis. Some tests may also determine which treatments may be the most effective. Although a biopsy, which is the removal of a small amount of tissue for examination under a microscope, is the way to make a definitive diagnosis for most types of cancer, this is usually not possible for retinoblastoma, and the doctor will suggest other ways to make a diagnosis. Imaging tests may be used to find out whether the cancer has spread. This list describes options for diagnosing this type of cancer, and not all tests listed will be used for every person. Your doctor may consider these factors when choosing a diagnostic test:

- Age and medical condition
- Type of cancer suspected
- Signs and symptoms
- Previous test results

The next step after observing any symptoms (see Symptoms [3]) is to have the child examined by a specialist, who will do a thorough ophthalmic examination to check the retina for a tumor. Depending on the child?s age, either a local or general anesthetic is used during the eye examination. Anesthetic is a medication that blocks the awareness of pain.

The specialist will make a drawing or take a photograph of the tumor in the eye to provide a record for future examinations and treatment. Additional tests may also be done to locate or confirm the presence of a tumor.

In addition to a physical examination, the following tests may be used to diagnose retinoblastoma:

Ultrasound [4]**.** An ultrasound uses sound waves to create pictures of the internal organs. A transmitter that emits sound waves is moved over the child?s body. A tumor generates different echoes of the sound waves than normal tissue does, so when the waves are bounced back to a computer and changed into images, the doctor can locate a mass inside the body. The procedure is painless.

Computed tomography (CT or CAT) scan [5]**.** A CT scan creates a three-dimensional picture of the inside of the child?s body with an x-ray machine. A computer then combines these images into a detailed, cross-sectional view that shows any abnormalities or tumors. A CT scan can also be used to measure the tumor?s size or help the doctor find cancer outside of the eye. Sometimes, a special dye called a contrast medium is given before the scan to provide better detail on the image. This dye can be injected into a patient?s vein or given as a pill to swallow.

Magnetic resonance imaging (MRI) [6]**.** An MRI uses magnetic fields, not x-rays, to produce detailed images of the brain and spinal column. MRI can also be used to measure the tumor?s size. A special dye called a contrast medium is given before the scan to create a clearer picture. This dye can be injected into a patient?s vein or given as a pill to swallow.

MRI or CT scan of the brain. These tests may be recommended to find out if there is an abnormality of the pineal gland, which is a small gland in the brain that regulates the body?s response to light. It is recommended that these scans be performed once every six months until age five for children with the genetic form of retinoblastoma, which includes those with bilateral disease and those with unilateral disease with a family history of the disease. Very young children with a tumor in one eye who do not have a family history of the disease may also be at risk, and these tests may be recommended. Scans may also be recommended years after treatment for children who have received external-beam radiation therapy (see Treatment [7]), either as a baseline in case of problems in the future, or to determine the cause of a new symptom or sign of a problem.

If there are any additional signs found during the physical examination, the doctor may recommend additional tests to determine if the cancer has spread elsewhere in the body.

Blood tests. These tests evaluate the blood and check for problems with the liver and kidneys. The doctor may also look at the blood for changes in chromosome 13. Chromosomes are the parts of a cell that contain genes, and in a few cases of retinoblastoma, these genes are either missing or nonfunctional. Molecular analysis of the gene is now possible in a few medical centers to determine changes that are not visible on ordinary chromosome analysis.

Lumbar puncture (spinal tap). A lumbar puncture is a procedure in which a doctor takes a sample of cerebral spinal fluid (CSF) to look for cancer cells, blood, or tumor markers, which are substances found in higher than normal amounts in the blood, urine, or body tissues of people with certain kinds of cancer. CSF is the fluid that flows around the brain and the spinal cord. Doctors generally give an anesthetic to numb the child?s lower back before the procedure.

Bone marrow aspiration and biopsy [8]**.** These two procedures are performed to determine if any retinoblastoma cells have spread to the bone marrow and are often done at the same time. Bone marrow has both a solid and a liquid part. A bone marrow aspiration removes a sample of

the fluid with a needle. A bone marrow biopsy is the removal of a small amount of solid tissue using a needle. The sample(s) are then analyzed by a pathologist, which is a doctor who specializes in interpreting laboratory tests and evaluating cells, tissues, and organs to diagnose disease. A common site for a bone marrow aspiration and biopsy is the pelvic bone, which is located in the lower back by the hip. The skin in that area is usually numbed with medication beforehand, and other types of anesthesia may be used.

Hearing test. Children with retinoblastoma taking specific types of chemotherapy (see Treatment [7]) may have their hearing tested (called an audiology test) to make sure the drugs are not causing hearing loss.

After diagnostic tests are done, your doctor will review all of the results with you. If the diagnosis is cancer, these results also help the doctor describe the cancer; this is called staging.

The next section helps explain the different stages for this type of cancer. Use the menu on the side of your screen to select Stages, or you can select another section, to continue reading this guide.

Links:
[1] http://www.cancer.net/cancer-types/retinoblastoma-childhood/diagnosis
[2] http://www.cancer.net/about-us
[3] http://www.cancer.net/node/19580
[4] http://www.cancer.net/node/24714
[5] http://www.cancer.net/node/24486
[6] http://www.cancer.net/node/24578
[7] http://www.cancer.net/node/19583
[8] http://www.cancer.net/navigating-cancer-care/diagnosing-cancer/tests-and-procedures/bone-marrow-aspiration-and-biopsy