Neuroblastoma - Childhood - Diagnosis [1]

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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 find, or diagnose, a tumor. They also do tests to learn if cancer has spread to another part of the body from where it started. If this happens, it is called metastasis. For example, imaging tests can show if the cancer has spread. Imaging tests show pictures of the inside of the body. Doctors may also do tests to learn which treatments could work best.

For most tumors, a biopsy is the only sure way for the doctor to know whether an area of the body has cancer. In a biopsy, the doctor takes a small sample of tissue for testing in a laboratory. If a biopsy is not possible, the doctor may suggest other tests that will help make a diagnosis.

Your child's doctor may consider these factors when choosing a diagnostic test:

- The type of cancer suspected
- Your signs and symptoms
- Your age and medical condition
- The results of earlier medical tests

According to international criteria developed by an International Neuroblastoma Risk Group task force, a diagnosis of neuroblastoma can be made if:

- Neuroblastoma cells are detected in the bone marrow and higher than normal levels of one of the main chemicals produced by the nervous system, called catecholamine are present in the urine, or

- By a tumor biopsy showing neuroblastoma cells

In addition to a physical examination, the following tests may be used to evaluate patients with neuroblastoma. Not all tests listed will be used for every person.

- **Blood tests and urine tests.** Complete blood counts (CBC) [3]are tested to find out if the child has signs of anemia, which is having low levels of red blood cells in the blood. To evaluate liver and kidney function, additional blood tests will be done. In addition, blood clotting tests may also be recommended.

  Urine is collected to test for tumor markers produced by a neuroblastoma tumor. A tumor marker is a substance found in higher than normal amounts in the blood, urine, or body tissues of people with certain kinds of cancer. Urinary catecholamine metabolites are detected in more the 85% of patients with neuroblastoma.

  Catecholamines are organic compounds that include the hormones epinephrine (adrenaline), norepinephrine (noradrenaline) and dopamine. Release of the hormones epinephrine and norepinephrine from the adrenal medulla of the adrenal glands is part of the fight-or-flight response. The fight-or-flight response is the body's natural reaction to a harmful situation, preparing itself to either quickly leave the area or stay to fight. Eventually the body breaks down the catecholamine molecules into smaller pieces, called metabolites, which are then passed out of the body in the urine.

  The 2 catecholamine metabolites most often measured are homovanillic acid (HVA) or vanillylmandelic acid (VMA).

- **Biopsy.** A biopsy [4] is the removal of a small amount of tissue for examination under a microscope. A pathologist then analyzes the sample(s). A pathologist is a doctor who specializes in interpreting laboratory tests and evaluating cells, tissues, and organs to diagnose disease. The type of biopsy performed depends on the location of the tumor. If the surgeon determines that the entire tumor can be removed, the whole tumor is commonly removed instead of doing a separate biopsy.
- **Genetic studies.** Tests of neuroblastoma cell DNA are used to find a change in the oncogene *MYCN*, a gene responsible for cell growth. More than 10 copies of the gene,

called amplification, is associated with a tumor that grows and spreads quickly, making it more difficult to treat. A non-amplified *MYCN*gene is linked to less aggressive tumors, which grow and spread more slowly.

Additional tests are done to find out if the tumor has changes in the numbers of whole chromosome or parts of chromosomes. A series of studies have shown that segmental chromosomal aberrations are associated with more aggressive disease, while whole chromosome gains are seen in tumors with more favorable outcomes. DNA sequencing tests to find out if mutations of *ALK* are also commonly done. Sequencing studies have shown that a small subset of neuroblastoma tumors have gene mutations, and some of these mutations, like *ALK*, can be targeted with new drugs. Studies have shown that tumors that come back, called recurrent or relapsed, often have increased gene mutation frequency.

For patients with a family history of neuroblastoma, genetic tests to determine if germline mutations in the *PHOX2B* or *ALK* genes are commonly done.

- **Bone marrow aspiration and biopsy.** These 2 procedures [5]are similar and often done at the same time to examine the bone marrow. 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.

  A pathologist then analyzes the samples. 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. Other types of anesthesia, which is medication to block the awareness of pain, may also be used.

- **Computed tomography (CT or CAT) scan.** A CT scan [6] creates a 3-dimensional picture of the inside of the body using x-rays taken from different angles. 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. A special dye called a contrast medium is usually given before the scan to provide better detail on the image. This dye is injected into a patient's vein.

- **Magnetic resonance imaging (MRI).** An MRI [7] 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 is injected into a patient's vein. An MRI is better at showing tumors around the spine, and it is essential to look at a tumor that is near where nerves leave the spinal column, which can press on the spinal cord.

- **MIBG scan.** MIBG stands for meta-iodobenzylguanidine. Neuroblastoma cells absorb this protein. When the protein is linked to a small amount of radioactive iodine, it can be used to find neuroblastoma in the bone, bone marrow and other parts of the body. Because the child's thyroid gland will also absorb radioactive iodine, regular iodine is taken by mouth for several days before the scan to protect the thyroid.

- **Positron emission tomography (PET) or PET-CT scan.** A PET scan is usually combined with a CT scan (see above), called a PET-CT scan [8]. However, you may hear your doctor refer to this procedure just as a PET scan. A PET scan is a way to create pictures of organs and tissues inside the body. A small amount of a radioactive substance is injected into a patient's body. This substance is absorbed mainly by organs and tissues that use the most energy. Because cancer tends to use energy actively, it absorbs more of the radioactive substance. A scanner then detects this substance to produce images of the inside of the body. This test is usually performed for patients who have tumors that do not take up MIBG (see above).

After diagnostic tests are done, your child's doctor will review all of the results with you. If the diagnosis is neuroblastoma, these results also help the doctor describe the cancer; this is called stages and risk grouping [9].

The next section in this guide is Stages and Groups. [9] *It explains the system doctors use to describe the extent of the disease. Or, use the menu to choose another section to continue reading this guide.*

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[3] http://www.cancer.net/node/24716
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