

**Laboratory tests** are studies that require a sample of blood, urine, body tissue, or other bodily fluids (obtained by a laboratory technician) to analyze the health and functioning of the body. The results of these tests can help diagnose disease or illness, plan or evaluate treatment, and monitor disease progression. Not all tests are necessary; your child's physician will decide which tests are right for your child.

## Common Serum (Blood) and Urine Studies

Blood samples are usually obtained through venipuncture (a small needle used to puncture a blood vessel and collect a blood sample). Urine studies can be obtained voluntarily, but younger children and infants may require catheterization (a small tube inserted into the urethra to collect urine).



- **Complete blood cell count (CBC)**: Measures the number of red blood cells and white blood cells, hemoglobin, and hematocrit. This common lab study helps diagnose infections, detects anemia or blood-clotting problems, and evaluates red blood cell production or destruction.
- **Comprehensive metabolic panel (CMP)**: A common lab study that evaluates electrolyte levels in the body, acid/base balance (pH), blood sugar levels, and liver and kidney function.
- **Thyroid studies**: Test the level of different thyroid hormones (TSH, free T4, and T3) in the bloodstream. Can help identify hypothyroidism or hyperthyroidism.
- **Inflammatory markers**: Includes erythrocyte sedimentation rate (ESR) and C-reactive protein (CRP). Both are tests for inflammation in the body. The results are nonspecific, meaning the test cannot determine where the source of inflammation is located.
- **Liver function tests (LFTs)**: A panel of tests that evaluate how well the liver is working.
- **Vitamin levels**: Measure the level of certain nutrients in the body. Vitamin D and B12 levels are most commonly ordered, but tests for folic acid and vitamin E levels are not uncommon.
- **Iron studies**: This group of tests includes iron, iron saturation, ferritin, and TIBC (total iron-binding capacity). These look at levels of iron carried in the bloodstream and stored in your body. They can help diagnose iron deficiencies.
- **Toxicology screening**: Measures the level of prescription and/or illegal drugs in the body, as well as ethanol (alcohol) levels. Can be obtained through blood or urine.
- **Drug levels**: Monitor the levels of prescription medication in the bloodstream. Can help providers adjust medications for best therapeutic effect and evaluate compliance with medication regimens.

# Common Lab Studies

- **Heavy metal testing:** Used to test for the presence of heavy metals, such as lead, iron, mercury, or copper. A related test is a ceruloplasmin test, which measures levels of a copper-containing protein. Heavy metal tests may be individual or in a panel of tests, and they may involve both blood and urine samples. Sometimes urine must be collected over a 24-hour period.



## Common Metabolic and Genetic Studies

Don't hesitate to discuss the potential risks and benefits of genetic testing with your child's neurology provider. If the results of a genetic study are abnormal, your child may be referred to a geneticist or genetic counselor for further interpretation, information, and guidance.

- **Chromosomal studies:** Include a karyotype, which evaluates the number and structure of the chromosomes, and a chromosomal microarray (CMA), which looks for small changes in chromosomes. Both are used to screen for genetic disease. Sometimes abnormal results can indicate a specific condition, and other times abnormal results have unknown significance.
- **Gene-specific tests:** These tests are used to look for specific changes (or mutations) in chromosomes. They are ordered when a specific genetic disorder is suspected. They can be obtained by blood or buccal (cheek) swabs. These tests are sent to outside facilities and may need prior authorization by your insurance company before they are completed.
- **Other metabolic studies:** These include lactate and pyruvate levels, acylcarnitine profile, and very long chain fatty acids (VLCFAs), among others. Each test is meant to evaluate for metabolic disease. Metabolic testing is often combined with genetic testing.

## Common Cerebral Spinal Fluid (CSF) Studies

The fluid that surrounds the brain and spinal cord (CSF) is collected during a lumbar puncture (a sterile procedure in which a needle is inserted into a space at the base of the spine). The CSF is collected and sent to a laboratory for analysis. Sometimes the pressure of the CSF (called opening pressure, or OP), is measured.

- **Routine CSF studies:** Measure the number of red and blood white cells, glucose levels, and protein levels in the CSF. In children, this test most often screens for infection or bleeding.
- **Bacterial cultures and viral studies:** Tests that identify the presence of bacteria or viruses in the CSF. Can take several days to be processed and for results to become available.
- **CSF neurotransmitters:** Used to screen for rare neurometabolic disorders.

## Resources and References:

<https://medlineplus.gov/> (use the Search MedlinePlus function to view more information about the tests listed above)