

Children with Holoprosencephaly often have many additional complications, so chances are your child will require some or many of these tests based on their individual needs

BMP

The basic metabolic panel (BMP) is a set of seven to eight blood tests that measure certain nutrients and electrolytes essential for basic body functions. The basic metabolic panel typically measures the serum or plasma levels of sodium, potassium, chloride, and bicarbonate in the body. The BMP is also used to test other electrolytes such as: blood urea nitrogen (BUN), creatinine, and glucose to check for or rule out diabetes or kidney disease.

CBC

A complete blood count (CBC) is a common blood test used to evaluate overall health and detect a wide range of disorders, including anemia and infection. The test measures:

- Hemoglobin, the oxygen-carrying protein in red blood cells
- Hematocrit, the proportion of red blood cells to plasma in the blood
- White blood cells, which fight infection
- Platelets, which help with blood clotting

A complete blood count may be done as part of a routine medical examination. Abnormal increases or decreases in cell counts may indicate an underlying medical condition that warrants further evaluation.

CT Scan

A "CT" or "CAT" scan are the common terms for "computerized tomography" (or computed axial tomography). The CT scanner is a doughnut-shaped machine that uses advanced x-ray technology to take cross-section pictures of your body, called "slices." A CT scan can visualize the brain and other parts of the body that cannot be seen on regular x-ray examinations.

What to expect? Your child may need to be sedated for this test, because they need to remain extremely still, and an IV may need to be started. You may be able to watch the procedure from an area shielded from radiation.

EEG

An electroencephalogram (EEG) is a test that measures the electrical activity in the brain, called brain waves. An EEG measures brain waves through small button electrodes that are placed on your child's scalp. An EEG is a painless recording of the brain's electrical activity. Small gold cups, called electrodes, will be used to "listen" to your child's brain waves. These will NOT shock your child.

What to expect? Your child's physician may request that your child be sleep deprived for this test. The technologist measures and marks 30 spots on your child's head. This is done through the hair, and no hair needs to be removed. These marks indicate where the 30 electrodes will be applied. The electrode sites need to be very clean in order to get the best recording possible, so the sites are rubbed with a mild cleanser and a paste or glue will be used to apply the electrodes to your child's head. The application procedure takes around 30 minutes, and your child's head needs to be still for the whole procedure. The length of time the EEG is recorded depends on what test your child's doctor has ordered. At the end of the recording, the technologist will painlessly remove the electrodes with water or solvent, depending on what was used to put on the electrodes.

Echocardiogram

An echocardiogram (ECG) is a painless, non-invasive test that uses ultrasound to study the structure and function of the heart. There is no radiation involved in this test.

What to expect? An ECG is very similar to an ultrasound you experienced during pregnancy. Your child will be placed on a table in a regular doctor's examination room, not an operating room. Conductive gel will be placed on your child's chest, and then a transducer will be placed on top of the gel. The transducer is able to "broadcast" high frequency sound waves into your child's chest cavity. These sound waves bounce off of the heart and the results, or echoes, are picked up on the ECG machine, which makes pictures based on the sound waves. An echocardiogram usually takes between 30 minutes and an hour, in order for the doctor to perform a thorough examination of the heart's function.

Lumbar Puncture

A lumbar puncture (an LP) is the insertion of a needle into the fluid within the spinal canal. It is termed a "lumbar puncture" because the needle goes into the lumbar portion (the "small") of the back. An LP is most commonly done for diagnostic purposes, namely to obtain a sample of the fluid in the spinal canal (the cerebrospinal fluid) for examination.

MRI

MRI (magnetic resonance imaging) is a diagnostic procedure that uses a combination of a large magnet, radio frequencies, and a computer to produce detailed images of organs and structures within the body.

What to expect? Due to the need to remain quite still, your child may need to be sedated for this test and an IV may need to be started. Jewelry, glasses and objects containing metal must be removed prior to the procedure. The technicians need to be informed if your child has any metal implants or objects in his/her body (such as a cochlear implant).

Sleep Study

A sleep study or polysomnogram (PSG), is a non-invasive, painless test that records and measures various functions during various stages of sleep. Small electrodes are placed on numerous areas of the body (head, face, chest, legs) to monitor specific measurements of breathing, brain activity, muscle movements, eye movements and leg movements while your child is sleeping.

What to expect? You will typically bring your child to the study location in the evening, in order to spend the night at the facility. You will prepare your child for bed, and the technician will place numerous electrodes on the child's body. Then, the child needs to sleep a "typical night's sleep" for the study to be accurate. Similar in process to an EEG, all measurements are taken using sticky electrodes or probes, and belts around your child's chest. These will NOT shock your child. There is usually a camera in the room as well, so they record the movement and activity or your child throughout the night, to correlate with the electronic results. You should be able to sleep in the room with your child.

The results will not be given immediately, but will typically return in several weeks after your child's study has been read and analyzed. At that point, depending on the results, further action or studies may be recommended.

Most of these tests are fairly non-invasive, but please keep in mind, your child might require additional special treatment during a test due to his or her specific physical needs, personality or concerns

**We encourage
you to be
a strong advocate
for your child:
Speak Out
and
Speak Loud
especially
if you have any
questions,
concerns or
complaints**

Swallow Study

A videofluoroscopic swallow study (also commonly referred to as modified barium swallow study) is an objective assessment of swallow function. The test is performed with both a pediatric radiologist and speech language pathologist with specialized training in pediatric dysphagia. The purpose of the test is to assess for aspiration (food/liquid entering the trachea) during oral feeding.

What to expect? You may remain with your child during the testing. Your child will be positioned in a typical feeding position consistent with his/her age & development. Your child will be given a variety of food consistencies (thin liquid, thick liquid, puree, soft solid, hard solid) injected with barium. The study assesses all 3 phases of the swallow from the oral preparatory phase, oral initiation phase and the pharyngeal phase.

Upper GI Endoscopy

This is a procedure for looking inside the upper gastrointestinal tract which includes the esophagus, the stomach, and the duodenum, the beginning of the small intestine. Upper GI endoscopy is performed by a gastroenterologist, who uses an endoscope to diagnose and, in some cases, treat problems of the upper digestive system. An endoscope is a long, thin, flexible tube with a tiny video camera and light on the end. By adjusting the various controls on the endoscope, the gastroenterologist can safely guide the instrument to carefully examine the inside lining of the upper digestive system. The high quality picture from the endoscope is shown on a TV monitor giving a clear, detailed view of the upper GI.

What to expect? The physician may require that food and water be withheld in the hours prior to the procedure because anesthesia will be required for the procedure. An anesthesiologist should speak with you prior to the procedure to obtain medical history. You may be allowed to be present with your child during the procedure, and you may be asked to wear a surgical gown over your clothing. Most physicians explain each step of the procedure as they go along and will provide you with color photos of the various sections of your child's upper GI tract. The physician may use the endoscope to take tissue biopsies to test for bacteria or infection, and as a result, if your child has a g-tube, you may notice some brown blood particles in the stomach residual when you resume your child's feedings. Following the procedure, your child is taken to a recovery area for observation. Once your child is awake and responding, you will be allowed to leave.

Disclaimer

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