Patient Information Sheet
Hypoplastic Left Heart Syndrome

You have recently learned that your baby has hypoplastic left heart syndrome (HLHS). We expect that you may have questions about what this could mean for your pregnancy. Hopefully, this information sheet will address some of your concerns; however, we encourage you to speak to your genetic counselor or healthcare provider for more information. It is important to remember that anyone can have a baby with a birth defect. Birth defects occur in 2-5% of all births and are rarely caused by something that the parents did or did not do before or during pregnancy. Currently, there are no tests available to detect all health problems.

What is hypoplastic left heart syndrome?

Hypoplastic left heart syndrome (HLHS) is a serious heart defect where the left side of the heart does not fully develop. Normally the left side of the heart receives the oxygen-rich blood from the lungs and pumps it to the rest of the body. In a baby with HLHS, the structures on the left side of the heart, including the left ventricle (pumping chamber), are too small and not well developed and cannot pump oxygen-rich blood to the body. Baby needs oxygen in the body after it is born, and HLHS causes the baby to have trouble breathing, a weak pulse, pounding heart and a blue tint to the skin. If not fixed with surgery, babies with HLHS cannot survive outside of mother's womb.

How many babies are found with hypoplastic left heart syndrome?

Hhypoplastic left heart syndrome is estimated to occur in 2-3 per 10,000 pregnancies (0.2%-0.3%).

What causes hypoplastic left heart syndrome?

The majority of HLHS cases are thought to be multifactorial, meaning that they are caused by a combination of both environmental and genetic factors. Maternal health factors such as poorly controlled diabetes, as well as the use of certain medications, can sometimes increase the risk for a pregnancy to be affected with a heart defect. A family history of congenital heart defects also increases the risk for a pregnancy to be have a congenital heart defect, including HLHS. Therefore, it is important to share information about your health as well as the health of your family members with your genetic counselor or physician.

Some cases of hypoplastic left heart syndrome are due to a genetic cause such as a chromosome abnormality or other syndrome. In order to understand chromosomal and other genetic causes, it is helpful to know a little about chromosomes. Chromosomes are packages of genetic information that carry the instructions (genes) necessary for our growth and development. Typically, there are 23 pairs of chromosomes in each cell of our bodies. One copy of each pair is from our mother and one copy of each is from our father. The first 22 pairs of chromosomes are the same in males and females. The last pair is known as the sex chromosomes and they determine our gender. Females typically have two X chromosomes and males usually have one X and one Y. It is important to have the correct amount of genetic material to have typical development. Sometimes there is an imbalance of genetic material that may cause birth defects. The imbalance may be caused by an extra or missing whole chromosome or sometimes only by extra or missing pieces of chromosomes. Some genetic conditions are caused by changes in one or more genes that result in a particular set of characteristics or features (also called a syndrome). A chromosome imbalance or genetic syndrome can produce birth defects and physical changes such as HLHS.

What problems could be associated with HLHS for my baby and how are they treated?

Hypoplastic left heart syndrome is one of the most complex types of heart defects. Without surgery most babies with HLHS pass away shortly after birth. Three separate open heart surgeries are generally needed for babies with HLHS. For each surgery baby is placed under anesthesia and on cardiopulmonary bypass, also known as a heart-lung machine, to provide blood flow to the rest of the body while the surgical team repairs the heart. The three surgeries are performed in stages and are usually completed by early childhood: Norwood, Glenn, and Fontan.

After the surgeries are completed the baby will have only a single working ventricle and this ventricle has to pump the blood from the heart both to the lungs and to the body. Unfortunately, these surgeries do have a risk of complications and not all babies survive all three surgeries. Survival into early adulthood and beyond is still not well known. A consultation with a cardiologist or cardiovascular surgeon during pregnancy is often helpful to go over the specific defect(s) and expected course for your baby after delivery, as well as the proposed surgical intervention and timeline. Some parents may choose to end a pregnancy or choose comfort/palliative care for a baby that has been diagnosed with a HLHS.

What further testing may be offered and what will it tell me?

Depending on how far along you are in pregnancy, a number of additional tests may be offered to you. If HLHS is suspected during a routine ultrasound, a more detailed ultrasound will usually be performed to look carefully at the baby's body. A fetal echocardiogram (a specialized ultrasound of the baby's heart) may also be recommended. You may be referred to a high risk maternal fetal medicine (MFM) clinic or a fetal center for further evaluation.

You may also be offered different types of genetic tests. There are two types of prenatal tests, diagnostic and screening:

Diagnostic Tests

Depending on how far along you are in pregnancy an amniocentesis procedure may be offered. The amniocentesis is a diagnostic test in which a small sample of amniotic fluid is obtained to examine the baby's chromosomes. This fluid is obtained by inserting a thin needle through your belly and into the amniotic sac. Because the procedure is considered an invasive procedure, there is a risk, likely less than 1%, for complications that can lead to miscarriage.

If you choose to pursue an amniocentesis, you may be offered an additional test called a chromosome microarray (CMA). It is used to identify small missing or extra pieces of the baby's chromosomes that may be associated with genetic conditions. CMA cannot detect all genetic conditions and may detect variations in the chromosomes that have uncertain clinical significance. Parental blood samples may help to clarify the meaning of a variation, but effects of these changes may not be known until after the baby is born. CMA may also detect information such as non-paternity and close relationships between parents. In addition to CMA, you may also be offered testing for specific genetic syndromes to look for changes in one or more genes.

Screening Tests

There are multiple screening tests to determine the chances for a pregnancy to have a chromosome condition. These screening tests cannot diagnose or rule out the presence of a chromosome condition but may be used to help you decide whether or not you would like to pursue a diagnostic test.

Cell-free DNA (cfDNA), also referred to as Non-Invasive Prenatal Testing (NIPT) or Non-Invasive Prenatal Screening (NIPS), is a screening test for certain chromosome conditions. During pregnancy, some of your baby's placenta's chromosome material is in your blood, along with your own chromosome material. Cell-free DNA screen is a blood test that measures the amount of chromosome material in your blood to determine if your baby could have an extra chromosome. This test screens for Down syndrome, trisomy 18, trisomy 13 and may also screen for some less severe conditions that are caused by different numbers of the X or Y chromosome. Cell-free DNA screen may also screen for several conditions caused by small missing pieces of chromosomes. While cell-free DNA screen is a good screen it is not a diagnostic test, like the amniocentesis. The detection rate for these chromosome conditions is typically between 90-99%. Other screening blood tests (often called a triple, quad, penta, or first trimester screen) may also be offered.

The risks, benefits, and limitations of screening and diagnostic testing options should be discussed with you by your genetic counselor or other health care provider. Follow-up counseling and referrals for support can be made if a chromosome condition is detected prenatally. As with all situations in which prenatal testing is discussed, it is your decision whether or not this test is done.

It is important to remember that prenatal screening and testing cannot detect or rule out all genetic conditions. After the baby is born, if additional birth defects or developmental delays are noted, an examination by a pediatric geneticist may be recommended. The geneticist may recommend that a small amount of blood be taken from the baby to evaluate for a chromosome or genetic problem. This examination and possible additional tests are important, since an accurate diagnosis will provide you with the best information regarding the prognosis for your baby, the cause of the HLHS, the risks to future children and that of other family members, as well as to determine appropriate tests to be offered to you in future pregnancies.

The decision to have one or more of these tests or to do no additional testing is a difficult and personal one. There is no one right decision. Some people will decide to have no further testing because they do not feel they need to know if their baby will have any of the chromosome conditions before delivery. Other people feel that they want more information as soon as possible. After gathering all of the information you need about HLHS and testing, you should make the decision that is right for you and your family.

What is the chance for this to happen again?

Parents of a baby with HLHS are often worried about the risk for it to happen in another pregnancy. If HLHS is isolated and not part of a genetic condition, and if there is no other family history, the chance to have another baby with the same or different heart defects is approximately 3%. After HLHS is diagnosed in a family it may be recommended that other family member get their hearts examined via an echocardiogram for changes in the heart because in some families changes in the heart can be familial.

What do I do now?

This information is only intended as an introduction to some of the terms and tests that you have already heard or will be hearing about from your genetic counselor and healthcare provider. We hope that this information sheet will be helpful as you begin to understand more about HLHS. We understand that any time something of concern is found through prenatal screening and testing, parents are going to be worried. Please don't hesitate to contact your genetic counselor with any questions or concerns you have. We are here to help you and your baby.

You may find it useful to have a consultation with a maternal-fetal medicine physician, a cardiologist, a cardiovascular surgeon, and a geneticist prior to delivery to discuss specifics about HLHS, prognosis, and available treatments. You may also find it useful to have a consultation and an evaluation performed at a fetal center experienced at taking care of babies with heart defects. However, depending on your specific case and what hospital you are seen at, your health care provider may recommend additional consultations and tests that are not discussed here. Support groups, either online or in person, can provide comfort for families facing this diagnosis. One-on-one therapy or family therapy may also be useful, both during and after the pregnancy.

Support groups:

The Heart House http://childrensheartinstitute.org/content/for-patients/heart-house.html
Little Hearts https://www.littlehearts.org
Linked by Heart http://linked-by-heart.org/
Mended Little Hearts http://www.mendedlittlehearts.org/