Patient Information Sheet
Atrioventricular (AV) Canal Defect

You have recently learned that your baby has a congenital heart defect called an atrioventricular (AV) canal defect. Sometimes this type of heart defect is also called an endocardial cushion defect or atrioventriculoseptal defect (AVSD). We expect that you may have questions about what this could mean for your developing baby. Hopefully, this information 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 an AV canal defect?

To understand an AV canal defect, it can be helpful to understand how the heart works. The heart has two sides, right and the left, that are separated by an inner wall of tissue called the septum. The two sides are separated into upper chambers (atria) and the lower chambers (ventricles). Normally, the right side of the heart collects oxygen-poor blood in the right atrium. The blood is then moved through a valve (which keeps blood flowing in one direction) into the right ventricle, which is the pumping chamber. The ventricle then pumps the blood out of the heart to the lungs to receive oxygen. When this happens, oxygen rich-blood is returned to the left side of the heart and collects in the left atrium. The blood then goes through another valve into the left ventricle, which pumps the oxygen-rich blood to the rest of the body.

An AV canal defect is a heart defect in which there are holes in the septum that divides the left and the right side of the heart, and the valves that control the flow of blood between the chambers may not be formed correctly. This can cause the blood to have lower amounts of oxygen as well as not flow in the correct direction. The heart and lungs then have to work harder to supply the rest of the body with oxygen-rich blood. If not corrected this can lead to various medical concerns, including heart failure.

There are two general types of AV canal defects, depending on which structures are not formed correctly. In a *complete* AV canal defect there is a large hole in the septum that separates the left and right sides of the heart. The hole is in the center of the heart, where the atria and the ventricles meet. Also, instead of having two valves directing blood flow, there is only one (common valve). In comparison, a *partial* or *incomplete* AV canal defect is considered less severe as the hole does not extend between the ventricles and the valves are better formed. A small number of babies with a *complete* AV canal defect may also have what is called an "unbalanced" AV canal defect where one of the pumping ventricles becomes small and weak and does not pump blood well.

How many babies are found with an AV canal defect?

AV canal defects occur on average in 1-4 per 10,000 (0.1-0.4%) births.

What causes an AV canal defect?

AV canal defect can occur on its own (isolated), or can be seen along with other problems in the heart or the in the rest of the baby's body. More than half of the babies with this type of heart defect have an underlying chromosome disorder, such as Down syndrome.

In order to understand a possible chromosomal cause or genetic syndrome cause, 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. Down syndrome is caused by an extra copy of chromosome 21. Some genetic conditions are caused by changes in one or more genes in our genetic make-up that result in a particular set of characteristics or features (also called a syndrome). A genetic imbalance or genetic syndrome can cause birth defects and physical changes such as a heart defect, but also other organ defects and/or medical concerns.

If the AV canal is isolated and the baby does not have a syndrome, then it could have been caused by multifactorial (complex) inheritance, meaning that some environmental effects of unknown origin combined with a certain genetic predisposition caused the heart defect. Maternal health conditions, such as uncontrolled diabetes, have also been associated with AV canal defects.

What problems could be associated with an AV canal defect for my baby and how are they treated?

The prognosis and surgical repair will likely depend on the type of AV canal defect that your baby has and if the baby has any other problems in the heart or the rest of his/her body. *Complete* AV canal defects are considered more serious and, when not fixed, can cause serious medical problems in babies including heart failure and lung disease. Surgical repair is typically recommended between 4 to 6 months of age, though this may vary. *Incomplete* or *partial* AV canal defects are often asymptomatic until adulthood. However, surgical repair during the first few years of life is often recommended because the hole between the atria will not close on its own. If left uncorrected, these individuals are at risk for heart failure later in life.

There are several surgical methods used to correct an AV canal defect. Most commonly, a patch is used to close the hole between the ventricles and the atria and to separate the one valve into two. The operation is an open heart surgery performed under anesthesia and it is necessary to place the child on a 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. Most children do very well after surgery for AV canal defects. However, the left-sided heart valve can be prone to leaking even after surgery and some of the children may need another surgery to repair it.

Babies that have an "unbalanced" AV canal defect, however, are usually very sick at birth. Babies with an "unbalanced" AV canal defect may require at least 3 open heart surgeries, otherwise they do not survive. Because babies with an "unbalanced" AV canal defect have only a single working ventricle, the ventricle has to pump the blood from the heart both to the lungs and to the body. Babies that survive the surgeries usually do well, but there is a risk for complication with each surgery. Some parents may choose to end a pregnancy or choose comfort/palliative care for a baby that has been diagnosed with an AV canal defect.

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 an AV canal defect 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:

<u>Diagnostic Tests</u>

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.

<u>Screening Tests</u>

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 possible cause of the heart defect, 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 AV canal defects 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 an AV canal defect are often worried about the risk for it to happen in another pregnancy. If the AV canal defect is isolated and not part of a genetic condition and there is no other family history, the chance to have another baby with the same or different heart problem is approximately 2.5-3%.

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 an AV canal defect. 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 the AV canal defect, 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 AV canal 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.