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Congenital Heart Defects & 22q11.2 Microdeletion

Congenital heart defects are one of the most common birth defects, occurring in approximately 1% of the general population. Congenital heart defects may be isolated (occur alone) or associated with other problems. There are multiple causes for heart defects, some of which are outlined below.

Isolated Heart defects: Most individuals who have a heart defect will have it as an isolated birth defect. In individuals for which there are no other birth defects present and no known genetic reason for the heart defect, multifactorial inheritance is most likely. This means that there are both genetic and environmental factors that caused the heart defect. In these cases, the recurrence risk (chance that it will occur again in a future pregnancy) is approximately 3-5%, however this number varies with the specific type of heart defect, the number of affected individuals in the family, and the degree of relatedness between the pregnancy and the affected individual(s). Please refer to your physician or genetic counselor for a specific recurrence risk value.

Medication exposure and Heart defects: Some medications that a mother may take during her pregnancy pose a risk for heart defects. Examples of medications that may cause heart defects if a child is prenatally exposed include lithium and ACE inhibitors. In these cases, the recurrence risk for future children being affected with a heart defect is dependent upon the specific medication and the timing and extent of the exposure.

Maternal Medical Conditions and Heart Defects: Women who have certain medical conditions are at an increased risk for having a child with a heart defect. Examples of these conditions include uncontrolled diabetes and lupus. It is important that you speak specifically to your physician or genetic counselor regarding any medical conditions that you have and the risks that they may pose to your pregnancy.

Genetic Causes of Heart Defects: There are many genetic causes of heart defects, some of which are inherited from one or both parents and others that are caused by a de novo (new) change in an individual. In the majority of cases of heart defects with a genetic cause, there are other birth defects or other medical problems present. Examples of such conditions include Down syndrome, Williams syndrome and Marfan Syndrome. In these conditions, there is a change in an individual's DNA that causes them to have the condition. These conditions all require management by a geneticist, cardiologist and multiple other specialists to address the different features of these conditions.

Heart Defects and 22q11.2 Microdeletion:

About 1 in 2-4000 babies will be born with a microdeletion (very small missing piece) of chromosome 22. (This condition is sometimes called DiGeorge syndrome, Shprintzen syndrome or velo-cardio facial syndrome.) Individuals who have this microdeletion have a very small piece of the chromosome 22 missing at the 11.2 location. This is usually such a small deletion that it cannot be found by a standard chromosome test. In order to find this microdeletion, a special test, such as a FISH or microarray, must be used.

Individuals who have a chromosome 22q11.2 microdeletion may have a variety of symptoms and birth defects. Approximately 75% will have a heart defect, specifically a type called a conotruncal heart defect. Other possible problems may include missing or underdevelopment of the thymus and/or parathyroid glands causing immune problems or calcium problems (~75% of individuals), a range of palatal abnormalities (~70% of individuals), speech abnormalities, feeding difficulty, hearing loss, renal abnormalities, mental illness, and learning disabilities (70-90% of individuals). Not everyone with this microdeletion will have every symptom. In fact, some individuals will have only a few of the symptoms and the presence of this chromosome abnormality may go unnoticed by their doctors.

In most instances, if an individual has a 22q11.2 microdeletion, it occurs sporadically. However, 7% of individuals with this microdeletion inherited it from one of their parents. The parent may have very mild symptoms and may be undiagnosed. Therefore, if there is a family history of heart defects along with other symptoms such as learning disabilities, testing may be considered for this microdeletion. Many specialty medical centers now recommend any individual with a heart defect be tested for this chromosome 22 microdeletion, as approximately 1 in 68 individuals with a heart defect have a 22q11.2 microdeletion.

If you think you or a member of your family may have this condition, you may want to discuss testing with your physician or genetic counselor.