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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 causes. Some congenital heart defects are caused by medication exposure during a pregnancy, while others are associated with a genetic disorder. Additionally, individuals with some chromosome abnormalities may also have an increased risk for a heart defect. One example is Down syndrome, in which approximately 40% of infants will have a congenital heart defect. Most isolated heart defects are inherited in a multifactorial pattern in families. In multifactorial inheritance, a combination of genetic and environmental factors cause a condition to occur and the risk for a similar condition in closely related family members is increased.

About 1 in 4000 individuals with a heart defect will have 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, as special blood test, called FISH, 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. Other possible problems may include missing or underdevelopment of the thymus and/or parathyroid glands, cleft palate, speech abnormalities, feeding difficulty, mental illness, learning disabilities as well as others. 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, 6% 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.

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.