Prenatal Genetic Counseling: Patient Information Sheet
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The goal of this clinic is to provide you with information about the health of your children. Some couples we see are concerned about risks for a future pregnancy. Others are pregnant and may want to learn about prenatal screening or testing. A number of birth defects can be detected after 10 weeks of pregnancy. For many women, test results are normal and we are able to provide reassurance about the pregnancy. If a problem is identified, we can provide additional information and arrange for appropriate consultations. You should know birth defects are found in 2 to 3% of all babies and that some birth defects exist that we are unable to test for. Thus, prenatal screening and testing cannot guarantee a healthy baby, but the chance for a birth defect is reduced in pregnancies with normal test results.

Our clinical program includes a number of steps. A genetic counselor will take your family and medical histories, discuss specific risks to your baby's health for which you were referred and go over the various screening and testing options available. After this discussion you may decide to proceed with further evaluation. If you are pregnant, this counseling session can help you decide if you wish to have screening or testing.

Your prenatal testing options may include ultrasound, blood tests, amniocentesis and chorionic villus sampling (CVS). Ultrasound is the study of your fetus, placenta and womb, using a technique in which ultrasound waves are reflected off your tissues. From this procedure we learn where your placenta is located, how well your fetus is growing and how far along you are in pregnancy. A sonogram at 18 to 22 weeks of pregnancy will provide you with information about the fetal anatomy. Certain major birth defects are often detectable by ultrasound.

There are several forms of screening available during pregnancy that can provide you with an adjusted calculation of the chance for the fetus to have certain chromosome abnormalities, including Down syndrome. These screening tests are available in the first or second trimester. They involve a blood test and may also involve an ultrasound. Screening tests cannot provide a fetal diagnosis, so CVS or amniocentesis may still be available after these screens.

Chorionic villus sampling (CVS) is a procedure involving the retrieval of a small sample of the developing placenta by way of the vagina or through your abdomen. This procedure is performed in the first trimester of pregnancy at 10 to 13 weeks. If the vaginal route is advised, a thin catheter will be advanced by the obstetrician through the cervix to the placenta. If the placenta is more accessible through an abdominal approach, the procedure will be similar to an amniocentesis.

Since chromosomes contain the genetic material of your fetus, an abnormality would often result in the baby having physical and mental disabilities. For some patients, studies

to rule out other inherited conditions will be indicated. The placental villi will be grown in the laboratory and appropriate chromosome and/or other genetic tests can be performed. Results from CVS will be available within about two weeks and we will notify you as soon as possible.

Amniocentesis is done beginning at 16 to 17 weeks and involves placing a needle into your womb to collect about two tablespoons of amniotic fluid, which is the fluid that surrounds the fetus. Amniotic fluid contains cells that have been shed by the fetus which can be grown and analyzed in the laboratory. As with the placental villi, the chromosomes of these amniotic fluid cells can be studied to make sure that their number and structure are normal.

A small portion of the amniotic fluid sample is also sent to measure a substance called alpha-fetoprotein (AFP). AFP is a protein made by the fetus, and an elevated value can occur when the fetus is affected with certain conditions, particularly structural abnormalities involving the spinal column, skull or the abdomen.

Since the amniotic fluid cells take time to grow, it usually takes two weeks to complete the tests after an amniocentesis, and we will notify you as soon as the results are available.

CVS and amniocentesis are generally considered safe diagnostic tests, but as with any medical procedure there are risks to your pregnancy. The possible risks may be balanced by the benefits to you, all of which will be discussed with you in the genetic counseling session.

If your insurance company requires pre-authorization for these services, you will need to obtain this prior to the day of your scheduled appointment. Co-pays should be paid when you arrive at our center. Some insurance companies will cover a portion of the total bill, and patients will be responsible for any balance.