DIAGNOSTIC TESTING

Genetics & IVF Institute

DIAGNOSTIC TESTS provide accurate, definitive information about the chromosomal or genetic status of a pregnancy. They can determine if a fetus is affected or unaffected with a specific medical problem. Diagnostic tests are more comprehensive than screening options in pregnancy. To obtain diagnostic results, either chorionic villus sampling (CVS) or amniocentesis is required.

WHO SHOULD CONSIDER DIAGNOSTIC TESTING?

Diagnostic testing is available to any pregnant patient who wants to obtain the most genetic information as is possible about their current pregnancy. However, since the procedures to obtain this information carry a small (approximately 1 in 1,000) risk of complications, many people consider diagnostic testing only if a specific genetic risk is identified in the pregnancy.

DIAGNOSTIC TESTING OFFERS EVERY PREGNANT PATIENT:

- Definitive test results instead of probabilities
- The ability to rule out potential abnormalities
- Knowledge of the chromosome status of the pregnancy prior to delivery
- Additional information to guide follow-up evaluations
- Testing for more conditions than is available by screening tests

WHAT IS CHORIONIC VILLUS SAMPLING (CVS)?

CVS is a procedure to remove a small amount of tissue called chorionic villi from the placenta. This tissue is within the uterus, but outside of the gestational sac where the fetus is located. The tissue provides information about the chromosome or other genetic disease status of the developing fetus.

HOW IS CVS PERFORMED?

CVS is performed either by guiding a thin needle through the lower abdomen into the uterus, or by guiding a thin flexible catheter through the cervical opening after inserting a speculum in the vagina. Ultrasound is used for guidance, and the location of the placenta on the day of the procedure will determine which approach is used.

CVS OR AMNIOCENTESIS?

For the most part, similar tests can be ordered following either procedure. Only in rare circumstances would both procedures be indicated. However, sometimes one procedure is recommended over the other based on the patient's gestational age and prior test results. At Genetics & IVF Institute, your care will include a consultation with one of our genetics specialists who will provide guidance regarding your current pregnancy and testing recommendations.

WHAT IS AMNIOCENTESIS?

Amniocentesis is a procedure to remove a small amount of amniotic fluid from the uterus. The fluid contains cells that have been shed by the fetus. The sample is used for chromosome or other specific genetic tests, and to screen for open neural tube defects.

HOW IS AMNIOCENTESIS PERFORMED?

Amniocentesis is performed by guiding a thin needle through the lower abdomen and into the amniotic sac. The procedure is done using ultrasound guidance.

WHAT OCCURS AFTER A CVS OR AMNIOCENTESIS?

Most patients experience no medical complications after either procedure. It is recommended that they limit their activities for 48 hours afterwards.

Test results are communicated directly to patients directly by our genetic specialists. Our team specializes in result interpretation within a guided and supportive environment.

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HOW DO I SCHEDULE MY DIAGNOSTIC TEST?

Patients should call **703-698-7355** to request a CVS or amniocentesis appointment. Consultation appointments are scheduled prior to the procedure and are also available for those that are unsure if they will proceed with a diagnostic test or are unsure which procedure would be most appropriate for them personally. At the time of scheduling, please inform the staff if you are currently taking blood thinning medications. We look forward to supporting you.