What are the goals of this study?

1. Find out if whole exome sequencing improves diagnosis and treatment for patients with suspected genetic disorders.
2. Learn whether whole exome sequencing can be made available and affordable to a wide variety of patients.
3. Understand how genetic information affects families' lives and healthcare decisions.
4. Create new ways to communicate genetic information to patients, families, and doctors.

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Clinical Utility of Prenatal Whole Exome Sequencing

What is Whole Exome Sequencing?

Whole exome sequencing, or WES, is a new genetic test. It can be used to find the genetic cause of certain diseases. The test looks at the exome, which contains genetic information (DNA) used to control the functions of our body. WES examines thousands of genes at the same time. This information could lead to better treatments for some medical conditions and diseases.

What is PEGS?

PEGS is a research project to study how genetic information can improve clinical care. PEGS will provide genetic sequencing and counseling about the results. The *Clinical Utility of Prenatal Whole Exome Sequencing* project is a part of the larger PEGS program.

Who is Eligible?

The study will include pregnant women in whom a birth defect has been found in the fetus, and prenatal testing results done on amniocentesis or chorionic villus sampling (CVS) have not found the cause.

A doctor will decide if you are eligible. The study will analyze the DNA of the fetus and both parents. At least one biological parent must agree to have testing.

All testing will be paid for by the study.

What will happen if you decide to participate in this study?

- If your family is eligible for the study, a genetic counselor or doctor will explain the study to you and answer any questions you have.
- We will arrange to have DNA from your prenatal test (amniocentesis or CVS) sent to our lab. A blood or saliva sample will also be collected from both parents.
- You will meet with a genetic counselor or doctor a few weeks later to review the test results. These sessions will be recorded (with your permission). Some families will be given results using a special iPad App that will display the genetic results.
- You may be asked to participate in more detailed interviews about your experience with WES.
- You will be asked to return to the clinic about six months after you receive your results. This visit can be done by phone. You will be asked questions about your family's experience with WES.