

A Research Study

+ Blood samples are needed for important research on microdeletion/duplication syndromes

Who is eligible?

- Couples who have a child diagnosed with a microdeletion/duplication syndrome (a positive microarray test).

What is the purpose of the study?

The goal of this study is to develop a non-invasive prenatal blood test that can diagnose genetic disorders in the fetus by looking at fetal DNA (genetic material) found in the mother's bloodstream during pregnancy.

Specifically, the abnormalities in the fetus that we are looking for are microdeletions/microduplications (small missing or extra pieces of DNA that can cause problems) or specific genetic disorders. Parents of a child diagnosed with a genetic disorder (and their children) will be asked to participate.

If this study is successful, this will reduce the need for invasive procedures during pregnancy such as amniocentesis and chorionic villus sampling (CVS) but still enable women to find out accurate information regarding their baby's health early in the pregnancy.

+ What does participation require?

Both biological mother and biological father need to provide a blood sample. There will be 4 tubes collected from the mother (approximately 3 tablespoons) and 1 tube from the father (2 teaspoons). The child with a known genetic disorder will need to provide a blood sample (4 tubes) as well as his/her siblings who do not have a known genetic disorder (4 tubes). Study participants will need to provide medical records with genetic information to study sponsor. The samples will be collected at no cost to the family.

Neither subjects nor their doctor will receive any results from the study. Therefore, there is no direct benefit from participating. This study and the subjects who participate may help other women in the future if the study results in a new test available to pregnant women.

Each subject will receive \$100 for their participation. Parents will receive \$50 for each minor child who participates in the study.

Call Sallie McAdoo at 877-476-4743 X 322 or email at smcadoo@natera.com if interested

This study is sponsored and conducted by Natera, Inc. All patient samples and information will be treated in full compliance with HIPAA privacy laws.