About CombiMatrix

CombiMatrix is a clinical genetics laboratory that specializes in prenatal diagnosis, miscarriage analysis, pediatric developmental disorders and pre-implantation genetic testing.

As one of the most experienced microarray laboratories in the country, we are dedicated to providing patients with exceptional service through quality of care. Our testing services allows patients to obtain precise data to help them make more informed healthcare decisions.

Additional information is also availabe at

- combimatrix.com
- facebook.com/combimatrixcorp
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Understanding Parental Karyotyping

Shared Understanding.

Better Care.

CombiMatrix

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High-Tech. High-Touch.

What Is A Karyotype?

A karyotype is a test that studies your chromosomes. Chromosomes are found inside our cells, and hold all of the genetic information that is required for the body's development. Typically, we have a total of 46 chromosomes in each cell, organized into 23 pairs. The first 22 pairs are the same in men and women, while the 23rd pair represents the sex chromosomes. Men typically have an X and a Y, while women typically have two X chromosomes. Changes to the number of chromosomes or the arrangement of the genetic information on the chromosomes can cause a chromosomal disorder, depending on the type of change(s) present.

When Is This Testing Typically Recommended?

Your doctor may recommend karyotype if:

- You and your partner have had ongoing difficulty conceiving a pregnancy
- You and your partner have experienced two or more miscarriages/pregnancy losses
- A male partner has very low levels of sperm or no sperm in his semen (oligospermia/azoospermia)
- A female partner has been diagnosed with primary ovarian insufficiency (POI/POF)

What Does The Karyotype Tell Us?

Infertility

Individuals with unexplained infertility, absent or low sperm count, or primary ovarian insufficiency sometimes have an atypical set of sex chromosomes. For example, a woman may have only one X chromosome instead of two, or a man may have two X chromosomes plus a Y instead of one X and one Y.

History of Miscarriages/Pregnancy Losses

Occasionally, the genetic information on the chromosomes gets rearranged. If this happens and no genetic information is lost or duplicated, it is known as a balanced chromosomal rearrangement. Carriers of balanced chromosomal rearrangements are generally not at risk for specific disorders themselves; however, the atypical arrangement of the genetic information increases the chance of passing along an unbalanced chromosome rearrangement to their offspring. An individual with an unbalanced chromosome rearrangement has missing and/or extra segments of genetic information, which may lead to abnormal fetal development or even pregnancy loss, depending on the size and location of the genetic segment(s) involved.

Why Is Genetic Counseling Important?

Before undergoing any genetic testing, it is important to speak to a genetic counselor or another qualified healthcare provider about the risks, benefits, and limitations of parental karyotyping. For example, one limitation of karyotyping is that it cannot detect disorders due to mutations in single genes, such as cystic fibrosis, Tay-Sachs disease, or sickle cell anemia. Genetic counseling is recommended both prior to testing (to ensure that you understand what karyotyping can and cannot tell you), as well as after the results are complete, in order to help answer any questions you may have.

What If The Results Are Abnormal?

Your genetic counselor or health care provider will discuss any abnormal results with you in detail. In some cases, couples may have the option to use in vitro fertilization along with pre-implantation genetic screening (PGS) or pre-implantation genetic diagnosis (PGD). PGD and PGS are tests that are used to screen your embryos for specific conditions of concern, and can help identify those embryos that are most suitable for transfer.

What If I Have Questions?

For more information, you can speak with your healthcare provider, or ask to be referred to a genetic counselor in your area. To find a genetic counselor near you, visit: www.nsgc.org.

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