What are Chromosomes?

Chromosomes are structures made up of our genes. We have about 20,000 genes that need to fit inside every one of our cells. Our genes package themselves into chromosomes in order to fit inside our cells. You can think of this process as a string you hold on one end and twist on the other end, so it twists and condenses. If we unravel a chromosome, we get an extremely long piece or string of our genes.

Generally, we have a total of 46 chromosomes. Our chromosomes come in pairs. We inherit one chromosome of each pair from our mom/egg donor and the other from our dad/sperm donor. The pairs are labeled 1-22 based on size, and the 23rd pair determines our sex.

Each chromosome contains approximately 500-1000 genes. Changes in the number of chromosomes can result in missing or extra genes, leading to potential impacts on growth and development. Conditions such as developmental delays, birth defects, or autism may occur when there are chromosomal abnormalities.

Who We Are

Modern Reproduction's mission is to facilitate the discussion on the current technologies and genetic testing options available prior and during pregnancy by educating, empowering, and embracing modern reproduction. This handout does not serve as medical advice. Please refer to Modern Reproduction's Disclaimer and Terms and Service.

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Chromosomal Analysis

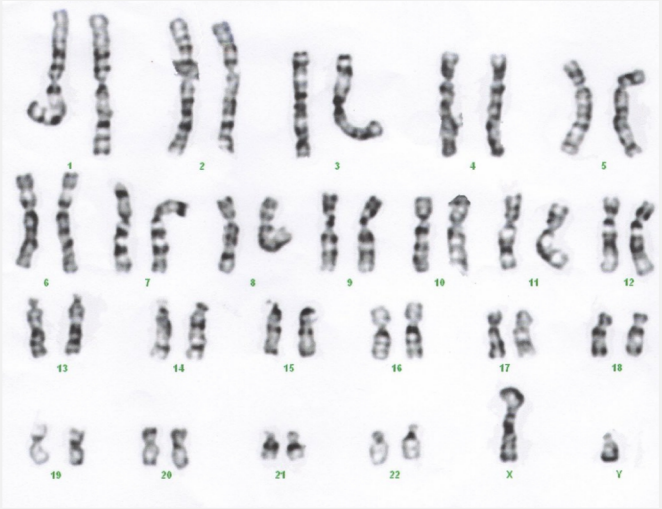
Why would someone have chromosome testing?

Chromosome testing, also known as chromosome analysis, can be done for several reasons. One reason is when a couple has a history of recurrent miscarriages. In some cases (3-5%), one member of the couple may have a chromosomal rearrangement where the chromosomes are arranged differently than usual. This can lead to an increased risk of passing on too many or too few chromosomes during conception, resulting in miscarriage (1).

Another reason for chromosome testing is unexplained infertility. Sometimes, individuals may have an undiagnosed chromosomal condition, such as Klinefelter syndrome, which is discovered when investigating the cause of infertility.

There are two main types of chromosome analysis: karyotype and microarray.

1. Hyde KJ, Schust DJ. Genetic considerations in recurrent pregnancy loss. Cold Spring Harb Perspect Med. 2015;5(3):a023119. Published 2015 Feb 6. doi:10.1101/cshperspect.a023119.



Karyotype

Karyotype is the above picture. It is a snapshot of the chromosomes which allows the ability to detect if there are any extra or missing copies of the chromosomes as well as if there is a rearrangement. This is the test that is most often ordered for adults undergoing assessment for recurrent miscarriage or infertility.

Microarray Analysis

Microarray analysis is a genetic test commonly ordered for individuals with a personal history of autism, developmental delays, or other health complications suggesting a genetic condition. It can also be performed in various other situations.

For example, a prenatal screen may suggest the pregnant person has a chromosomal condition like 22q11.2 deletion syndrome. Not all chromosomal conditions have obvious features, so there are times people never knew they had the condition.

Unlike karyotype, microarray analysis can detect small deletions, duplications, and whole chromosomal abnormalities. It is especially useful in identifying conditions like 22q11.2 deletion syndrome. However, it can also uncover genetic variations with unclear health implications known as "variants of uncertain significance".

Genetic counselors can provide guidance and interpretation of chromosomal analysis results.