

A chromosome analysis, or karyotype, is the traditional testing that can been done on cells from chorionic villus sampling (CVS) and amniocentesis procedures, or from a blood sample. A karyotype involves the lab examining the cells and determining the number of chromosomes.

How the lab determines the number of chromosomes is a complicated process. Chromosomes all have bands on them, which kind of look like stripes. The pattern of the bands can help the lab determine which chromosome they're looking at. For example, chromosome 21 has a very different banding pattern than chromosome 22.

These bands aren't easily visible so the lab uses staining, much like you would stain a glass window, to show the pattern of bands on the chromosome. The lab can then use that pattern to figure out what number chromosome they are looking at. They then use a computer to put the chromosomes in order. Most people have two of each chromosome, so the lab would use the computer to move the chromosome number 1s together, and next the chromosome number 2s together, and so on.

Once all 23 sets of chromosomes are laid out we get a picture, which is the karyotype. This allows us to see if there are any extra (called a trisomy) or missing (called a monosomy) chromosomes. A karyotype can also identify if large pieces of chromosomes:

- Have broken off and switched spots (called <u>chromosome translocations</u>),
- Have broken off and turned upside down (called chromosome inversions),
- That are missing (called chromosome <u>deletions</u>), or
- That are extra (called chromosome duplications).

If there is a trisomy, monosomy, <u>translocation</u>, inversion, <u>deletion</u>, or <u>duplication</u>, it is important to determine what chromosome(s) are involved. Each chromosome has different information on it, so if there is extra or missing chromosome information, what is extra or missing can help us determine if it can cause health concerns.

Karyotype results are usually back within 2-3 weeks after the lab receives them. It is important to remember that a karyotype can be diagnostic for large chromosome differences, but cannot identify all genetic or hereditary conditions.

Click here to learn more about scheduling a genetic counseling appointment for pregnancyrelated questions.



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How to Decide

Whether or not to pursue any genetic testing is a very personal decision. In some cases, moving forward with genetic testing may help to provide an answer to a health question that is running in someone's family, or may help to provide a name to a medical condition that someone...