

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

How the lab determines the number of chromosomes is a lengthy and complicated process. Basically, there are different staining techniques that can be used (think about staining a glass window). The staining process can show a pattern of bands that are specific to each chromosome. When the lab looks at the stained chromosomes under the microscope, they can recognize the unique banding patterns and determine what number chromosome it is.

Through this process, they number all of the chromosome, 1 through 22, as well as the sex chromosomes (X and Y). This forms the picture we call a karyotype. The lab is able to use this picture to count the number of chromosomes to determine if there are any extra (called a trisomy) or missing (called a monosomy).

If the karyotype finds that there is an extra or missing chromosome, then it is important to determine which chromosome is affected. Each chromosome has different information on it, so if we have an extra or missing chromosome, what information is extra or missing will determine what the related health concerns may be.

A karyotype is also able to show if there are any large missing or extra pieces of the chromosome, called deletions or duplications. Large deletions and duplications may cause health concerns, once again depending on what chromosome it is found in.

Traditional chromosome analysis is also able to show chromosome translocations, or when pieces of two different chromosome break off and switch spots with each other. This testing can also find inversions, or parts of chromosomes that have broken off and then turned upside down and reattached themselves. Chromosome translocations and inversions are overall rare, and can have varying implications for a pregnancy, so it is important to talk more with a genetics specialist about any specific questions you may have.

Karyotype results are generally back within 2-3 weeks after the lab receives them.

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Chorionic villus sampling, or CVS, is a procedure that can be done during the first trimester of pregnancy (usually between 11 and 14 weeks). CVS involves getting some

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