

Cytogenetic Analysis for the Examination of Chromosomes

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DESCRIPTION

Cytogenetics is a dynamic field of study that examines the number and structure of chromosomes in humans and animals. Changes in the number or structure of the chromosomes can cause issues with growth, development, and how the body functions. Chromosomal abnormalities can occur during the formation of egg and sperm cells, during early foetal development, or after birth in any cell in the body. Changes in chromosome structure can disrupt genes, resulting in over abundance, missing or faulty proteins produced by disrupted genes. Structure changes in chromosomes can cause birth defects, complex syndromes, or even cancer, depending on their size, location, and timing. Some chromosomal abnormalities may have no effect on a person's health.

A routine cytogenetic analysis includes assessing the structural integrity of each chromosome in the complement and evaluating 15 to 20 cells to determine their modal chromosome number. By counting chromosomes under a microscope, the modal chromosome number can be determined. A microscopic examination can identify an extra or missing chromosome, and the discovery is confirmed by the preparation of a set of karyotypes. A karyotype is a photographic or computer-generated representation of a cell's chromosomes arranged by size, centromeric position, and banding pattern. The microscope can also be used to assess the structural integrity of each chromosome in the complement, as well as detect gross structural chromosome aberrations. A set of two or more karyotypes must always be prepared readily.

Cells with well-prepared chromosomes have 350 to 400 bands in the complement are considered an essential component of any routine cytogenetic analysis. A standard chromosome analysis requires a chromosome count of at least 15 to 20 cells, the preparation and careful evaluation of two to three karyotypes consisting of well-banded chromosomes, and the exclusion of any structural chromosome aberrations. In this manner, homologous chromosomes can be accurately examined and compared for minute but clinically significant structural differences. The number of cells analyzed varies depending on the reason for referral, the initial cytogenetic findings, each

laboratory's protocol, and the chromosome morphology quality.

If an abnormal chromosomal complement is present in all of the cells in a specimen, a small number of cells like 10 to 12 may be sufficient for diagnosis. A single cell with a chromosome aberration is typically attributed to an error in cell division during tissue culture or a technical antiquity arising during harvesting. Most parents are informed that such a finding is not related to abnormal foetal development. The presence of an abnormal cell may necessitate analyzing 20 to 50 or more cells to differentiate between true mosaicism, which could cause foetal mal development, and pseudo mosaicism, which could be caused by a technical antiquity or an *in vitro* error.

True chromosome mosaicism occurs when cells with normal and abnormal chromosomes coexist. True mosaicism may be especially difficult to detect if it occurs infrequently. Most cytogenetic analyses are statistically based on a minimum of 15 to 20 cells to reduce the chance of true mosaicism not being detected to less than 5%. Confirmation of chromosome mosaicism may necessitate more invasive procedures, such as Percutaneous Umbilical Blood Sampling (PUBS), putting the patient at greater obstetric risk. Mosaicism for chromosome 5 in cultured amniotic fluid cells, followed by normal chromosome analysis of foetal blood obtained by PUBS, resulted in a child with developmental disabilities because the mosaicism was confined to the skin and not present in blood cells.

CONCLUSION

Cytogenetics analysis is important in patient care because it provides accurate testing results for disease diagnosis, differential diagnosis, and treatment evaluation. Every pregnancy carries a slight risk of a chromosomal or genetic condition. The basic code of prenatal screening is to provide a safe, accessible test to all pregnant women, thereby identifying those women who are at increased risk of having a baby with a chromosomal or genetic condition. Only an invasive test, such as amniocentesis or Chorionic Villus Sampling (CVS), can establish a genetic or chromosomal disorder in pregnancy. Cytogenetics has several advantages, including the ability to diagnose different types of cancer and determine the best treatment method.

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