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Commentary

Characterisation of Various Chromosomal Abnormalities and Its Balanced Rearrangements

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DESCRIPTION

Chromosomal abnormalities can occur accidentally during egg or spermatogenesis, or during the early stages of fetal development. Maternal age and certain environmental factors can play a role in the development of genetic defects. Prenatal screening and testing can be done to examine the fetal chromosomes and detect some, but not all, types of chromosomal abnormalities. Chromosomal abnormalities usually occur when there is a misconception in cell division. There are two types of cell division, mitosis and meiosis.

- Mitosis results in two cells that are replicas of the original cell. A cell with 46 chromosomes divides into two cells, each with 46 chromosomes. This type of cell division occurs systemically except for the reproductive organs. This is how most of the cells that are made and replaced.
- Meiosis results in 23 cells, which is half the number of chromosomes, instead of the usual 46. This is the type of cell division that occurs in the reproductive organs and results in eggs and sperm.

During mitotic and meiotic cell division of mammalian gametogenesis, DNA repair is effective in removing DNA damage. However, in spermatogenesis, monopoly sperm cells undergo major nuclear chromatin remodeling into highly condensed sperm nuclei, significantly reducing their ability to repair DNA damage later in the process.

Types of chromosomal abnormalities

Chromosomal abnormalities can be numerical or structural, some of them are:

Numerical abnormalities: Numerical abnormalities are a type of chromosomal abnormality. These types of birth defects occur when the cells of the body have a different number of chromosomes than are normally found. Instead of the typical 46 chromosomes in each somatic cell, there may be 45 or 47 chromosomes. Too many or too few chromosomes can cause health problems and birth defects.

Structural abnormalities: Structural abnormalities are present when a single chromosome is missing, unwanted, replaced by another chromosome, or upside down. Chromosomal abnormalities can occur accidentally during egg or spermatogenesis, or in the early stages of fetal development. Maternal age and certain environmental factors can play a role in the development of genetic defects. Structural rearrangement is defined as balanced if a complete set of chromosomes is still present, but balanced if it is rearranged and there is additional or missing information.

Disproportionate rearrangements include deletion, duplication, or insertion of chromosomal segments. Prenatal screening and testing can be done to examine the fetal chromosomes and detect some, but not all, types of chromosomal abnormalities.

Other structural chromosomal abnormalities do not result in the acquisition or loss of genetic material. These rearrangements occur due to inversions caused by two break events and end-to-end inversions of intervening chromosomal segments, translocations resulting from the exchange of chromosomal segments between two or more chromosomes, and Includes insertion. A segment of a chromosome is translocated and inserted into a new region of the same chromosome, another homologous chromosome, or a non-homologous chromosome. These rearrangements may be pathogenic if the gene is disrupted by a rearrangement breakpoint, a new fusion gene product is formed, or a positional effect is exerted on a gene adjacent to the rearrangement.

Balanced rearrangements include inverted or translocated chromosomal regions. A balanced chromosomal rearrangement may not be detected as it may not cause disease, as all DNA material is still there. As a result of balanced rearrangement, chromosomal breaks occur in one gene and the protein is missing or non-functional, or fusion of chromosomal segments results in a hybrid of the two genes, resulting in a new protein. In some cases, the disease can occur. A product whose function is damaging to the cell.

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