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Opinion Article

Breif Note on Chromosome Abnormalities in Down Syndrome

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DESCRIPTION

Chromosomal abnormalities can either be structurally or numerically aberrant. Numerical disorders, which include any departure from the typical diploid number for a specific species, are much more prevalent than structural ones. This category can be further broken down into two groups: one where specific chromosomes may be lost duplicated, or both; and the other where entire haploid sets of chromosomes may be added or When it comes to structural problems, however, one or more chromosomes have their genetic order changed or an uneven chromosomal exchange in between enzyme during the process repair of two damaged chromosomes is the main causes of structural abnormalities. Examples of this situation include duplications, ring chromosomes, iso chromosomes, translocations, and inversions. According to their classification as structural or numerical anomalies, chromosomal anomalies Chromosomal abnormalities can be classified as either acquired or constitutional. Constitutive chromosomal abnormalities impact all or a significant fraction of an organism's cells and first appear during gametogenesis or early embryogenesis. More than a thousand of these anomalies have been observed in live-born patients, and between 20% and 50% of all human conceptions are thought to be affected by them. Acquired chromosomal abnormalities often appear in adults and impact only one cell clone with a specific location inside the body and it not be on the pathogenesis of the many neoplasms affected by these modifications.

Aneuploidy

The failure to separate chromosomes during cell division, meiosis I or II, is the main cause of aneuploidy. Non-disjunction, premature disjunction, and anaphase lag are examples of this result. The most common of the aforementioned phenomena, non-disjunction, happens when the replicated chromosomes fail to adequately segregate during one of the two meiotic divisions, most frequently during meiosis I of egg development as opposed to sperm. If this does occur to the one or more copies of a specific chromosome will either be extra or absent in the ensuing germ cells. In contrast, paternal meiotic non-disjunction is typically the cause of at least 50% of cases of sex chromosomal aneuploidies. Depending on which meiotic cycle is involved in non-disjunction, different outcomes happen. Meiosis I is when 24 chromosomes from the paternal and maternal homologs would be present in the gamete. The gamete would still contain 24 chromosomes, but only two copies would come from either the paternal or maternal homolog if non-disjunction occurred in meiosis II.

Cytogenetic

A chromosome is referred to as cytogenetic. Although the physical foundation of inheritance—chromosomes—has been understood for well over a century, the field of mammalian cytogenetic and diploid chromosomes in human cells was determined to be 46, and a method for producing chromosomes from leukocytes grown from peripheral blood had also been developed. Since then, scientists have been able to accurately describe any deviations from the usual number or structure of human chromosomes.

Turner syndrome: Turner syndrome often does not run in families rather, it develops during the production of a parent's reproductive cells the first cell division stage of development. There are no known environmental dangers, and the mother's age has no bearing. A chromosomal anomaly in which all or a portion of one of the X chromosomes is missing or mutated is the cause of Turner syndrome. People with TS typically have 45 chromosomes in part or all of their cells, compared to the average of 46It are referred to as TS with mosaicism when the chromosomal abnormality only affects a few cells. Typically, there are fewer symptoms in these situations, and occasionally none at all. Physical symptoms and genetic testing are used to make the diagnosis.

XYY syndrome: About 1 in 850 male infants have a sex chromosomal aneuploidy called XYY syndrome that raises the risk of neurodevelopmental issues however, the profile of neurodevelopmental abnormalities, which includes ASD signs and symptoms (ASD).

Klinefelter syndrome: when a child is born with an extra copy of the X chromosome, a genetic disorder known as Klinefelter

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syndrome develops. Males with Klinefelter syndrome have a hereditary disorder that is frequently not discovered until adulthood. Klinefelter syndrome may negatively impact testicular growth, resulting in testicles that are smaller than usual, which can limit testosterone output.