



Chromosomal Abnormalities: Understanding Genetic Alterations and Their Clinical Impact

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DESCRIPTION

Chromosomal abnormalities are genetic alterations caused by changes in chromosome number, structure, or organization that affect normal development, growth and physiological functions. Chromosomes are complex structures composed of Deoxyribonucleic Acid (DNA) and proteins that carry genetic information required for cellular processes. Humans typically possess 46 chromosomes arranged in 23 pairs, with one chromosome from each pair inherited from each parent. Any deviation from this normal chromosomal arrangement can disrupt gene dosage, alter gene expression and contribute to a wide range of developmental, reproductive and medical conditions.

Aneuploidy is one of the most common types of numerical chromosomal abnormality and occurs when an individual has an extra or missing chromosome. Trisomy, in which an additional chromosome is present and monosomy, in which one chromosome is absent, represent major forms of aneuploidy. Down syndrome, caused by the presence of an additional copy of chromosome 21, is the most frequently recognized chromosomal disorder. The extra genetic material affects physical development, cognitive function and susceptibility to certain health conditions. Sex chromosome abnormalities involve variations affecting the X and Y chromosomes. Turner syndrome occurs when females have a single X chromosome instead of two complete sex chromosomes, resulting in features such as short stature, ovarian dysfunction and infertility. Klinefelter syndrome occurs in males with an additional X chromosome, commonly represented as 47, XXY and may cause reduced testosterone production, infertility and differences in physical development. Advances in molecular diagnosis have improved recognition and management of these conditions.

The development of advanced cytogenetic techniques has transformed the diagnosis of chromosomal abnormalities. Conventional karyotyping, which visualizes chromosomes under

a microscope, remains an important diagnostic method for identifying large-scale chromosomal changes. However, its ability to detect smaller genetic alterations is limited. Fluorescence In Situ Hybridization (FISH) technology uses fluorescent probes to identify specific chromosomal regions and detect targeted abnormalities with greater precision.

Chromosomal microarray analysis has become an important diagnostic tool for identifying submicroscopic chromosomal changes. This technique detects copy number variations, including small deletions and duplications that may not be visible through traditional karyotyping. More recently, Next-Generation Sequencing (NGS) and Whole Genome Sequencing (WGS) approaches have expanded the ability to identify complex chromosomal rearrangements and characterize genetic abnormalities at a higher resolution.

Chromosomal abnormalities play a significant role in reproductive health and pregnancy outcomes. Many chromosomal alterations are associated with infertility, recurrent pregnancy loss and congenital abnormalities. Preimplantation Genetic Testing for Aneuploidy (PGT-A) enables screening of embryos produced through assisted reproductive technologies to identify chromosomal abnormalities before implantation. Prenatal diagnostic approaches, including Non-Invasive Prenatal Testing (NIPT), analyze fetal DNA fragments present in maternal blood to estimate the risk of common chromosomal conditions.

Cancer research has revealed that chromosomal abnormalities also contribute to tumor development and progression. Chromosomal rearrangements can activate oncogenes or disrupt tumor suppressor genes, leading to uncontrolled cellular growth. The Philadelphia chromosome, resulting from a translocation between chromosomes 9 and 22, creates the *BCR-ABL1* fusion gene associated with chronic myeloid leukemia. Identification of such chromosomal changes has enabled the development of targeted therapies that specifically inhibit abnormal molecular pathways.

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In conclusion, chromosomal abnormalities represent important genetic changes that influence human development, reproduction and disease susceptibility. Advances in cytogenetics, sequencing technologies and computational biology have greatly improved the detection and understanding

of these conditions. Continued research into chromosome biology and genomic medicine will enhance diagnostic precision, therapeutic strategies and personalized healthcare approaches for individuals affected by chromosomal disorders.