

Identification of Chromosomal Abnormalities and its Diagnostic Techniques

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DESCRIPTION

Chromosomal abnormalities refer to changes in the structure or number of chromosomes in an individual's cells. Chromosomes are thread-like structures that contain Deoxyribonucleic Acid (DNA) and carry genetic information. Normal human cells typically have 46 chromosomes organized into 23 pairs, with one set inherited from each parent. However, chromosomal abnormalities can occur during the formation of reproductive cells (sperm and eggs) or during early embryonic development, leading to deviations from the usual chromosomal makeup.

Importance of genetic counseling and prenatal testing during pregnancy

Assessing genetic risk: Genetic counselors gather and analyze relevant medical and family histories to assess the risk of genetic disorders in the pregnancy. They identify potential risk factors such as advanced maternal age, previous pregnancy complications, family history of genetic conditions and exposure to environmental factors.

Educating and informing: Genetic counselors provide expectant parents with information about genetic conditions, including the causes, inheritance patterns, recurrence risks and available diagnostic and screening tests. They explain complex genetic concepts in a clear and understandable manner, allowing parents to make informed decisions.

Facilitating informed decision-making: Genetic counselors present the options for prenatal testing and screening, discussing the benefits, limitations and potential risks associated with each procedure. They help parents weigh the pros and cons based on their personal values, beliefs, allowing them to make decisions that align with their individual circumstances.

Coordinating prenatal testing: Genetic counselors help facilitate the logistics of prenatal testing, including scheduling appointments, explaining the procedures and ensuring appropriate communication with healthcare providers involved in the process.

Interpreting test results: Genetic counselors play a crucial role in interpreting and explaining the results of prenatal tests, such

as ultrasound, maternal serum screening, Chorionic Villus Sampling (CVS), amniocentesis and Non-Invasive Prenatal Testing (NIPT). They help parents understand the implications of positive or abnormal findings and assist in making decisions about further diagnostic testing or additional medical interventions.

Providing emotional support: Genetic counselors offer emotional support and address the anxieties, concerns, and emotional impact that expectant parents may experience during the prenatal testing process. They create a safe and empathetic space for parents to express their feelings and provide counseling to help cope with the uncertainties.

Referring for additional support: Genetic counselors collaborate with other healthcare professionals, such as obstetricians, perinatologists and pediatric specialists to ensure comprehensive care for expectant parents.

Advancements in diagnostic techniques

Techniques used in the detection of abnormalities are:

Cytogenetic analysis: Cytogenetic analysis is a branch of genetics that focuses on the study of chromosomes and their abnormalities. It involves examining the structure, number and behavior of chromosomes within cells to identify any genetic alterations or abnormalities. It typically starts with the collection of cells from an individual, such as blood, bone marrow or tissue samples. The collected cells are treated with specific stains, such as Giemsa or fluorescent dyes to enhance the visibility of chromosomes under a microscope. The stained cells are spread onto slides and examined under a microscope. Cytogeneticists analyze the size, shape and banding patterns of chromosomes to identify any abnormalities or variations. Chromosomes are typically arranged in pairs and organized into a karyotype, which provides a visual representation of an individual's chromosome complement. Cytogeneticists analyze the karyotype to detect various cytogenetic changes, including structural abnormalities such as deletions, duplications, inversions and translocations, as well as numerical abnormalities such as aneuploidy

Fluorescence *in situ* hybridization: Fluorescence *In Situ* Hybridization (FISH) is a molecular cytogenetic technique that

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uses fluorescently labeled DNA probes to detect and visualize specific DNA sequences within cells. By using specific DNA probes, FISH can identify microdeletions, microduplications, translocations, inversions and other subtle chromosomal rearrangements. DNA probes can be designed to target specific chromosomal regions or genes associated with particular disorders or genetic abnormalities.

Array comparative genomic hybridization: Array Comparative Genomic Hybridization (aCGH) is a molecular cytogenetic technique used to detect Copy Number Variations (CNVs) across the genome. It is based on the principle of hybridizing DNA from a patient (test sample) and a reference DNA sample to a microarray containing thousands or millions of DNA probes representing specific genomic regions. Advantages include such as genome-wide coverage and higher resolution.

Next-generation sequencing: Next-Generation Sequencing (NGS) can identify structural variants, which are alterations in the structure of the genome, such as deletions, duplications,

inversions, and translocations. By sequencing the entire genome or specific regions using techniques like Whole-Genome Sequencing (WGS) or targeted sequencing, NGS can detect structural variations at a high resolution. Techniques like targeted gene panel sequencing, exome sequencing, and whole-genome sequencing can capture and analyze specific genes or the entire coding region of the genome. NGS allows for the detection of point mutations, small insertions or deletions (indels) and other variants within genes associated with specific disorders. This enables rapid and comprehensive genetic testing, facilitating the diagnosis and understanding of single-gene disorders.

Non-invasive prenatal testing: Non-Invasive Prenatal Testing (NIPT) utilizes the analysis of cell-free fetal DNA in maternal blood to detect common chromosomal abnormalities. This section discusses the advantages of NIPT, such as its non-invasive nature and high detection rates for conditions like trisomy 21, 18 and 13.