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GENETIC CHARACTERISTICS AND CLINICAL SIGNIFICANCE OF X CHROMOSOME-LINKED DISORDERS

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Abstract

This article discusses the genetic basis of X-linked hereditary diseases, their mechanisms of inheritance, clinical manifestations and relevance. The study analyzes the structure of the X chromosome, the genes located on it and their functional significance in the human body. Also, the genetic mechanisms of X-linked diseases such as hemophilia, Daltonism, Duchenne muscular dystrophy are explained through clinical examples. The article covers modern trends in early detection, genetic counseling and diagnostic methods for X-linked hereditary diseases.

Keywords

X chromosome, hemophilia, color blindness, Duchenne muscular dystrophy, diagnostics, heredity, gene therapy, genetic counseling.

INTRODUCTION.

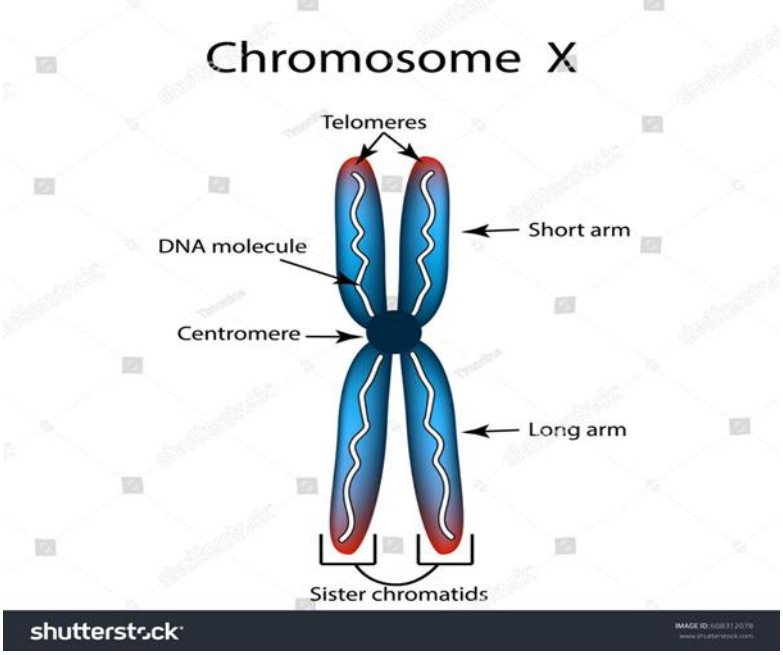
Human genetics has become one of the fastest-growing fields in medical science in recent years. In this process, chromosomal disorders, particularly X chromosome-linked hereditary syndromes, have been at the center of biomedical research. The X chromosome is a crucial part of the human genome, containing hundreds of genes that not only determine sex but also participate in numerous physiological and biochemical processes of the body. Mutations in these genes can lead to various hereditary diseases, including severe conditions such as hemophilia, Duchenne muscular dystrophy, color blindness (daltonism), and Fragile X syndrome. One of the most important branches of human genetics is the study of hereditary diseases associated with sex chromosomes. The human body contains 46 chromosomes, of which 44 are autosomes and 2 are sex chromosomes (X and Y). The X chromosome differs significantly from the Y chromosome in terms of size, gene content, and functional importance. Approximately one thousand genes are located on the X chromosome, playing critical roles in the functioning of various bodily systems. Mutations in these genes give rise to X-linked hereditary disorders. A distinctive feature of these diseases is that they are more commonly expressed in males, since males possess only a single X chromosome. In females, who have two X chromosomes, these mutations often remain in a carrier state due to the presence of a compensatory healthy gene on the other X chromosome.

Males have XY sex chromosomes, while females have XX. If a male carries a defective (mutated) gene on his single X chromosome, there is no corresponding healthy gene on the Y chromosome to compensate for the defect, causing the disease to manifest immediately. In females, the presence of two X chromosomes provides a compensatory mechanism: if one X chromosome carries a defective gene, the healthy gene on the other X chromosome can mask its effects. As a result, females usually become carriers of the disease, while clinical symptoms are often mild or absent. Currently, advances in genetic diagnostics, DNA analysis, and molecular marker technologies allow early detection of X chromosome-linked disorders. This enables the prevention of hereditary pathologies, improvement of genetic counseling services, and

maintenance of reproductive health. In Uzbekistan, early diagnosis of genetic disorders and strengthening preventive measures remain among the most pressing issues in medical practice.

MAIN PART.

The X chromosome is a large and gene-rich chromosome, with numerous genes located on its long arm. Its length is approximately 155 million base pairs, accounting for about 5% of the human genome. Among these genes are those responsible for diseases such as muscular dystrophy, hemophilia, intellectual disability, and enzyme deficiencies. In females, the presence of two X chromosomes leads to the inactivation of one chromosome during early embryonic development. This process is known as lyonization (Lyon hypothesis).



1.1 Figure

In Figure 1.1, the X chromosome is shown as an X-shaped condensed structure consisting of two chromatids joined at the centromere during cell division. The centromere divides the chromosome into a short arm (p arm) and a long arm (q arm), each exhibiting a distinct banding pattern. The X chromosome is composed of tightly coiled DNA wrapped around proteins called histones, containing genes that determine genetic traits.

• p arm (short arm) – primarily contains pseudoautosomal regions (PARs). These regions share homologous genes with the Y chromosome, allowing pairing during meiosis.

• q arm (long arm) – constitutes the largest portion of the chromosome, containing numerous unique genes.

X-linked disorders are inherited in two main patterns: dominant and recessive. The most common X-linked recessive disorders include:

1. Hemophilia A and B – disorders of blood coagulation caused by mutations in the F8 and F9 genes, respectively.
2. Duchenne Muscular Dystrophy (DMD) – characterized by progressive degeneration of muscle fibers due to mutations in the DMD gene.
3. Fragile X Syndrome – caused by mutations in the FMR1 gene, leading to intellectual disability and autistic features.
4. G6PD Deficiency – a condition resulting from deficiency of the enzyme glucose-6-phosphate dehydrogenase in erythrocytes, leading to hemolysis.
5. Rett Syndrome – a neurodegenerative disorder predominantly affecting girls, associated with mutations in the MECP2 gene.

The X chromosome is not only responsible for determining sex but also harbors genes that regulate numerous essential physiological processes. Disruption of its genetic balance can result in a variety of hereditary disorders.

Genetic Diagnostics, Research Methods, and Clinical Significance

Modern medicine employs a wide range of molecular genetic analyses for the detection of X-linked disorders, including DNA sequencing, polymerase chain reaction (PCR), genotyping, and karyotyping. Prenatal diagnostics allow for identification of these disorders during pregnancy. Research in gene therapy offers hope for future treatment and potential cures for these conditions. X-linked disorders affect a portion of the global population and are often severe, reducing both quality of life and life expectancy. Early diagnosis, genetic counseling, and potential gene therapy are critical for healthcare systems worldwide. In Uzbekistan, scientific research is actively conducted in this field, and medical-genetic counseling centers are operational.

CONCLUSION.

X-linked disorders represent a significant concern in human genetics and clinical medicine. Studying these conditions expands the possibilities for early diagnosis, treatment, and prevention of hereditary diseases. In the future, advances in gene therapy, CRISPR technology, and bioinformatics will play a crucial role in developing effective treatments for these disorders.

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