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CHROMOSOMAL DISEASES, IT'S CAUSES AND CONSEQUENCES

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ABSTRACT

Over the next 50 years, the deterioration of the ecological situation not only affects plants and animals, but also poses a great threat to human life. Due to environmental pollution, the number of genetic diseases, including chromosomal diseases, is increasing year by year. Due to the increase of these genetic diseases, the young generation around the world is struggling with the disease.

Chromasome diseases are one of the topics that are becoming a biologically global problem and causing a wide discussion among scientists. The birth of a healthy child means that he will grow up as a person who will protect him and be responsible for tomorrow's future for this country.

Keywords: Chromosome, karyotype, genotype, Down syndrome, Patau syndrome, Edwards syndrome, aneuploidy, Klinefelter syndrome, Turner syndrome.

INTRODUCTION

The birth of a healthy child depends, on the one hand, on the genetic factors of the parents, and on the other hand, on the factors of the external environment. In humans, chromosomal disease is caused by changes in the number of chromosomes or their structure. Changes in the number of chromosomes are usually caused by the uneven distribution of chromosomes to the poles during cell division. The main cause of this situation is anthropogenic influences. It is important to protect the environment, especially to prevent its contamination with radioactive substances. In addition, the main reason for the origin of this disease in newborns is the age of the parents, that is, if the age of the mother is over 35, the number of babies born with various chromosomal diseases increases. Children with the same disease are born when the father's age is over 42. The results of research show that 20% of the birth of sick children depends on the age of the father. As the age increases, the mitotic division process in

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germ cells is caused by a change in chromosomes 8,23,27. People with chromosomal disorders usually die in childhood (short-lived) or do not leave offspring. That is why chramasoma is not always transmitted from generation to generation and appears anew in each generation. Only 3-5% of it is a real hereditary disease and is passed from generation to generation.

LITERATURE ANALYSIS AND METHODOLOGY

Chromosomal diseases are hereditary diseases characterized by changes in the number or structure of chromosomes. Changes in chromosomes are a type of mutation. If chromosomal mutations occur in the first stages of the division of germ cells or fertilized egg cells, they are transmitted to many cells of the developing organism, resulting in a number of developmental defects. Embryos with abnormal chromosomal changes die before birth, 6% of stillborn babies have chromosomal abnormalities.

DISCUSSION AND RESULTS

1.0. Basic aspects of human genetics.

Man, being a part of the biosphere and a product of its development, is subject to the laws of heredity and change, like all organisms. The branch of genetics that studies human heredity and variability is called human genetics or anthropogenetics. Human genetics studies all the genetic traits found in people. Anthropogenetics studies the normal characteristics of the human body, while medical genetics studies its pathology, i.e. the heredity of hereditary defects.

Various genetic studies can be carried out on plants, animals and microorganisms. However, the methods used in plants, animals, and microorganisms cannot be used in humans. The main reason for this is not only a person's biological development, but also a product of social factors. Therefore, there are several difficulties in studying heredity and variability in humans.

1.1. Human karyotype.

The concept of karyotype can be defined in a narrow and broad sense. In a narrow sense, a karyotype is a diploid set of chromosomes of a single cell with a definite number, shape, and length. In a broad sense, a diploid set of organisms with a definite karyotype-number, shape, and length.

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Karyotype is constant for each species and is one of the main characteristics of this species. Each karyotype is distinguished by the following characteristics:

1. With the number of chromosomes,
2. With the shape and length of chromosomes,
3. With the number of secondary belts and the length of the umbilical cord,
4. With hetero and euchromatin threads.

Based on the study of human chromosomes, many chromosomal diseases have now been identified. For all people suspected of having a genetic disease, their karyotype will be determined. In the process of studying the human chromosome, scientists have created different systematics. This caused many difficulties in the study of human chromosomes. It was necessary to collect all the received information and develop a certain international systematics. For this purpose, in 1960, scientists of this field gathered in the city of Denver, USA and developed the international systematics of human chromosomes. Later, at the international conferences held in London (1963) and Paris (1973), clarifications were made to the Denver system, and today the system is used in practice.

1.2. Changes in chromosome structure.

According to the teachings of Thomas Morgan, each chromosome consists of a certain number of genes linked together. These genes are located in a certain order on the chromosome. According to the arrangement of genes in the chromosome, each chromosome has its own unique structure. An increase or decrease in the number of only certain chromosomes in the genome is called aneuploidy. K. Bridges was the first to identify the phenomenon of aneuploidy in the Drosophila fly. Aneuploidy usually occurs as a result of a violation of the separation of some chromosomes into the poles during cell division. Such changes are also observed in somatic and reproductive cells. Aneuploidy occurs on all chromosomes in humans.

1.3. Chromosomal disorders in humans.

The main object of studying human heredity based on the cytogenetic method is the cell, in which the subtle external and internal structure, chemical composition, and functions of the cell, especially chromosomes, are studied in detail. Currently, there are about 500 types of hereditary diseases that occur as a result of changes in the number and structure of chromosomes. Hereditary chromosomal diseases can occur due to changes in the number and structure of

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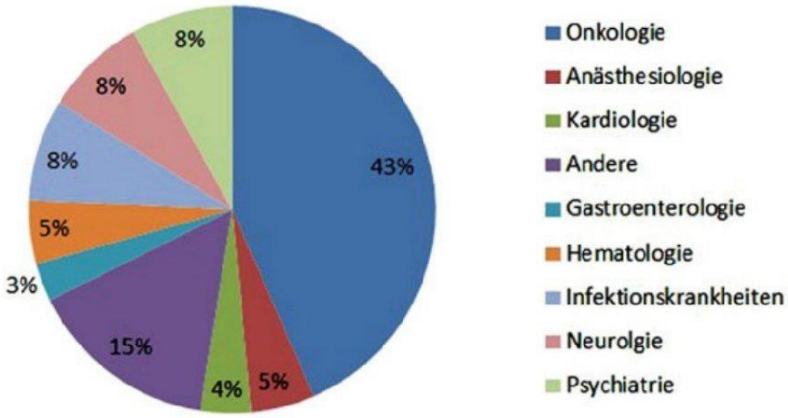
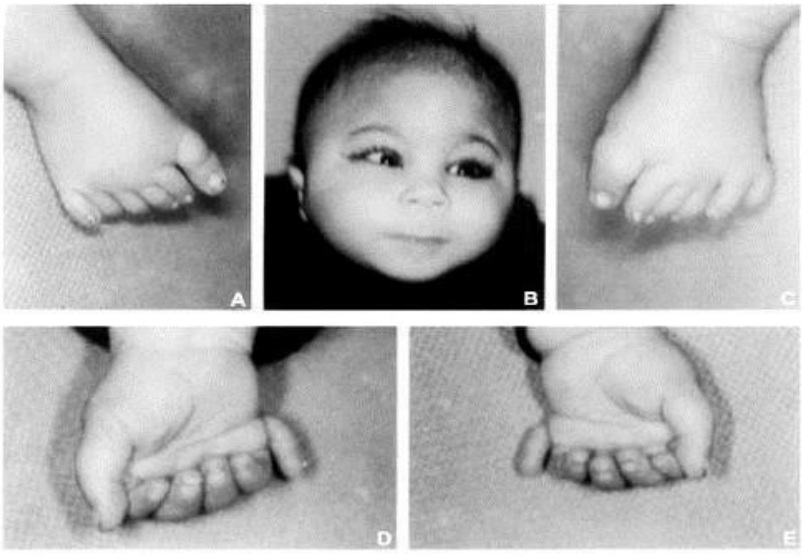
autosomes and sex chromosomes. Diseases related to autosomes: Down syndrome, Edwards syndrome, Patau syndrome are autosomal-chromosomal diseases.



Edwards syndrome was first identified in 1960 by D. Edwards, who found that one, i.e., the 18th chromosome is redundant. Children born with Edwards disease do not live long and die in the first months of life, while girls can live up to 2-3 years. Even if children with this disease are born fully formed, i.e. develop in the mother's womb for 9 months, their weight is very small. The following diseases are observed in them: bulging neck, elongated head, small jaws and oral cavity, high palate, ears located very low, circulatory system, eyesight and kidney structure are disturbed. the arms are very short. This disease is observed in one out of 4500, 6500 healthy children.



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Patau syndrome was first studied by K. Patau in 1961. The disease appears with an increase of one chromosome (46+1). Children born with this disease are born from healthy parents, and there is one sick child for every 3500, 4000 healthy children. Characteristics of the disease: the child's weight and height are small, most of the children suffering from this disease are born prematurely. There will be cracks in the upper lip and palate, and there may be no eyes. The brain is not well developed. The number of fingers will be more than usual, there will be changes in the kidneys, intestines, heart, and spleen.

Down's syndrome was identified by J. Down in 1866. The main reason for the origin of this disease is the result of an increase in the 21st pair of chromosomal chromosomes. The characteristic signs of benors suffering from this disease are as follows: the patient's head is very small, his face and forehead are wide, his eyes are small and close to each other, his mouth is half open, his mind is weak and barren. One in 800-1000 children with Down's disease will have this disease.

Симптомы синдрома Дауна

1.4. Diseases related to sex chromosomes.

Klanfelter's syndrome, a disease found in men, was identified by K. Klinefelter in 1942. In this disease, the X-chromosome is redundant. The ratio of children born with this disease to healthy children is 1:1000, which remains even in older people.

People suffering from Klinefelter's syndrome have the following symptoms: height, arms and legs are long, shoulders are narrow, pelvic bone and muscles are not well developed. In this disease, there will be no possibility of reproduction. Klinefelter's syndrome is the basis of 20% of male infertility.

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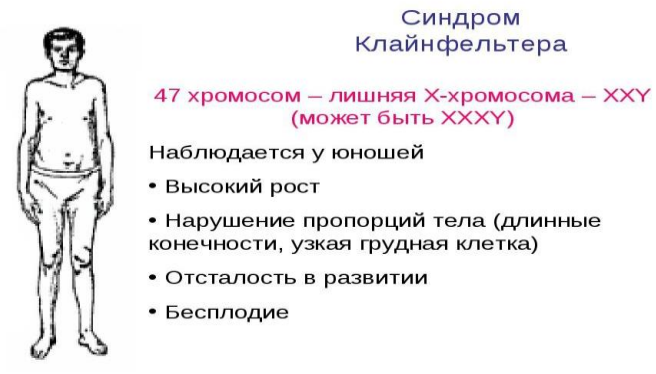

Shershevsky-turner syndrome was explained by N.A. Shershevsky in 1925 and Turner in 1939. Women with this disease have 45 chromosomes and one chromosome is missing. The main symptoms of the disease are as follows: short stature, light weight, very short and crooked neck, secondary signs are not well developed in the ovary, broad shoulders, short pelvis and legs, monthly cycle is not observed. His face looks old for his age. Changes occur in the blood circulation, nervous system and other internal organs. People suffering from this disease do not leave offspring. Treatment methods for this disease have not yet been found. In 80% of patients with the disease, sex chromatin was found to be absent.

CONCLUSION

1. In humans, chromosomal diseases are caused by changes in the number and structure of autosomes and sex chromosomes.
2. Changes in the number of chromosomes usually result from the uneven distribution of chromosomes to the poles during cell division.
3. Down syndrome is more common among urban residents than among rural residents. The main reason for this is marriage between close relatives, the age of the father and mother, and the mutagenic effects of the environment.

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