**MODERN VIEW OF STUDY CHROMOSOME DISEASE**

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About 10% of all forms of pathological conditions are mono-factor diseases caused exclusively by genetic breakdowns of the genetic or chromosomal level. Chromosomal diseases are hereditary diseases caused by changes in the structure or number of chromosomes. These include mutations of genomes or structural changes of individual chromosomes. Chromosomal anomalies occur in the body as a result of mutations in the genital cells of one of the parents. Chromosomal anomalies often cause disturbances in the genetic balance, genes and regulation, and are correlated during the evolution of each species. The pathological effects of chromosomal and genomic mutations occur at each stage of ontogenesis, including at the gamete level, and also affect their formation (especially in men). Although the clinic and cytogenesis of chromosomal diseases are well studied, their pathogenesis is, even in general terms, unclear. A general scheme of complex pathological processes due to chromosomal anomalies and result in complex phenotypes of chromosomal diseases has not been developed**.**

Today, the frequency of chromosomal anomalies is 6-8 per 1,000 newborns. In the total group of premature babies, chromosomal pathology is about 3-5%. The causal factors of chromosomal diseases are all types of chromosomal mutations, namely deletion, duplication, inversion, translocation, etc. Factors contributing to chromosomal anomalies include ionizing radiation, exposure to certain chemicals, severe infections, and intoxication. One of the external factors is the age of the parents: mothers and older parents are more likely to have children with karyotic disorders. The balanced wear-and-tear of chromosomes plays an important role in chromosomal anomalies. Complete forms of chromosomal syndromes arise from harmful factors on sexual cells in meiosis, while in mosaic forms negative events occur during the fetal life in mitosis (Vorsanova S. etc. 1999 ). For prognosis of progeny, it is important to find out the type of mutation, achieved by studying the cariotype of the patient and his parents.

The clinical picture and severity of chromosomal diseases depend on the nature of the restructuring, the size of the fragments involved and their functional significance.

Trisomy 13, called Patau syndrome, is a chromosomal condition associated with severe intellectual deficiency and physical anomalies in many parts of the body. Persons with trisomy often have heart defects, brain or spinal cord disorders, very small or poorly developed eyes (microthalmus), extra fingers or fingers, opening in the lip (split lip), with or without opening on the roof of the mouth (cracked sky) and weak muscle tone (hypotonia). Due to several life-threatening diseases, many children with trisomy 13 die during the first days or weeks of life. Only 5 per cent to 10 per cent of children with this condition live in the past their first year. The trisomy of the 13 chromosomes occurs in about 1 in 16,000 newborns. Although women of any age may have a child with Patau syndrome, the chance of having a child with this condition increases when the woman is older.

The trisomy of chromosome 18, also called Edwards syndrome, is a chromosomal state associated with anomalies in many parts of the body. Individuals with Edwards syndrome often have slow growth before birth (intrauterine stunting) and low birth weight. Affected people may have heart defects and other organ abnormalities, develop before birth. Other features of chromosome 18 trisomy include small, irregular head shape; small jaw and mouth; and clenched fists with overlapping fingers. Due to several life-threatening diseases, many of the trisomy 18 chromosomes die before birth or within the first month. Five to ten per cent of children with this condition live in the past, and these children often suffer from severe intellectual deficiency. A trisomy of 18 chromosomes occurs in about 1 in 5,000 congenital children; this diagnosis is more common during pregnancy, but many fetuses do not survive until delivery.

The syndrome «cat screaming» is a genetic condition. Chromosome Short Shoulder Syndrome 5 is a rare stage found in only 1 in 20,000 to 1 in 50,000 newborns, according to Genetics Home. But this is one of the most common syndromes caused by chromosomal defects. Babies with this syndrome have a distinctive scream, sounds like meow cats. The syndrome is more pronounced in childhood, but becomes difficult to diagnose after 2 or more years. Most fatal complications occur before the baby’s first birthday. Children who have reached the age of 1 will usually have a normal life span. But the child is likely to have physical or educational complications for the rest of his life. In most cases, chromosome deletion occurs when the parent biometherial or egg is still developing. That means the child develops a syndrome when insemination occurs. According to the Orphanet Journal Of Rare Diseases, chromosome deficiency comes from the material of the father (80% of cases). About 90% are considered to be random mutations.

Various methods are used to study the inheritance of normal and pathological human traits: genealogical, cytogenetic, biochemical, and twin. The most universal is the genealogical, that is, the pedigree method. The genealogical method seems at first glance simple, but to avoid errors requires a detailed probe and a careful definition of the type of inheritance. Most diseases are heterogeneous and may have different types of inheritance, so the prognosis of offspring may vary with the same clinical manifestation. This method is available to every practical physician. At the same time, it can provide much useful information that will facilitate the correct diagnosis and the provision of adequate treatment. It should be borne in mind that the medical doctor of any specialty (particularly relevant for family doctors) should be not only the patient but also his or her family members.