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Perspective

An Overview on Chromosome Abnormalities

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DESCRIPTION

Chromosomes are thread-like structures found within the nucleus contain tightly packaged DNA. Chromosomes contain information for the growth and development of body. Genes are located on chromosomes, which are located in the nucleus of the cell human body has near about 20,000 different genes in each and every cell. Our body contains 23 pairs of chromosomes in every cell. In those 46 chromosomes half of the chromosomes come from our maternal side (egg), and the other half come from our Paternal Side (sperm). The 23rd pair consists of autosomes (the first 22 pairs) and two sex chromosomes X and Y. Females have two X chromosomes, and males have one X and one Y chromosome along with 22 pairs of autosomes in each and every cell.

A chromosomal abnormality is a morphological or numerical alteration in single or multiple chromosomes, affecting autosomes, sex chromosomes, or both.

Generally when the egg or the sperm is formed or the fetus is developing its early stages is the time when chromosomal abnormalities can occur. Meiosis is the start of the process of a baby. Meiosis, a type of cell division process, causes each parent to give 23 chromosomes to a pregnancy. When a sperm fertilizes with an egg, the union leads to form a baby (zygote in initial stage) with 46 chromosomes. A baby may have an extra chromosome (trisomy), or have a missing chromosome (monosomy) if meiosis cell division doesn't happen properly. These problems lead to pregnancy loss or several kinds of chromosome abnormalities in a child. The age of the pregnancy and some environmental factors also play a role to form genetic errors in babies.

There are various types of chromosome abnormalities, classified mainly in two basic categories numerical abnormalities and structural abnormalities. Numerical Abnormalities are the types of chromosome abnormalities when an individual is missing one of the chromosomes from a pair known as monosomy or an individual has more than two chromosomes instead of a pair known a trisomy. So in this case there may be 45 or 47 chromosomes instead of the typical 46 chromosomes in each cell of the body.

Turner syndrome is an example of monosomy. This is a genetic condition in which a female is missing an X chromosome. Signs and symptoms of Turner syndrome are a short and webbed neck, low-set ears, low hairline at the back of the neck, short stature, and swollen hands and feet. Affected are unable to have children without reproductive technology.

Cri du chat syndrome is a genetic disorder (monosomy) due to a partial deletion on chromosome. This syndrome gets its name because the characteristic cry of affected infants, which is similar to that of a meowing kitten, due to problems with the larynx and nervous system.

Down syndrome is an example of trisomy. It is also called trisomy 21. It includes certain birth defects, learning problems, heart defects and problems with vision and hearing. Severity of these problems are varies from child to child.

Trisomy 18 or Edward syndrome is the condition where the babies have three copies of chromosome 18 instead of two. Patients with Edward syndrome suffered from physical irregularity of the kidneys, ureters, heart, lungs and diaphragm, cleft lip or cleft palate, small skull, malformations of the sex organs. Generally Survival beyond the neonatal period is uncommon for babies with Edward syndrome.

Trisomy 13 or Patau syndrome disrupts normal development of patients and cause multiple and complex organ defects.

Like Down syndrome and Edwards's syndrome the risk of Patau syndrome in the offspring increases with maternal age at pregnancy.

Structural abnormalities are the types of chromosome abnormalities when a chromosome's structure can be altered in several ways though the total number of chromosomes is typically 46 per cell. Structural chromosome abnormalities occur when part of a chromosome is missing called deletions, a portion of the chromosome is duplicated named duplications which resulting in extra genetic material, translocations, in which portion of one chromosome is transferred to another chromosome. In some cases of structural chromosome abnormalities a portion of the chromosome has broken off, turned upside down otherwise called Inversions.

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Loss of genetic material from chromosome 11 cause Jacobsen syndrome. Jacobsen syndrome is also known as 11q terminal deletion disorder as the deletion occurs at the end (terminus) of the long (q) arm of chromosome 11. Due to this disorder the affected individuals have delayed development of speech and motor skills like sitting, standing, and walking and learning difficulties.

Charcot-Marie-Tooth disease type 1A is an example of duplications disorder caused by duplication of the gene encoding peripheral myelin protein 22. This disease affects the peripheral nerves, People with this disease experience weakness and wasting (atrophy) of the muscles of the lower legs beginning in adolescence, hand weakness and sensory loss. Translocations of chromosomes can causes Cancer or Infertility in both male and female.

Chromosome abnormalities can be prevented by avoiding late pregnancy. Chromosomal Testing also can help to prevent chromosome abnormalities providing information about the probability having a chromosomal abnormality in baby. But these tests cannot diagnose a chromosomal abnormality. These tests include ultrasound and blood tests, like panel of biomarkers or testing of circulating placental DNA.