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Understanding Genes and Chromosomes

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Your body is formed from cells. within the middle of every cell may be a nucleus, and inside the nucleus are chromosomes. Chromosomes are important because they contain genes that determine your physical characteristics, your blood group , and even how susceptible you'll be to certain illnesses.Each cell within the body typically contains 23 pairs of chromosomes—46 chromosomes in total—each of which contains roughly 20,000 to 25,000 genes.

Chromosomal Abnormalities

Chromosomal abnormalities occur where there are missing or extra chromosomes, or pieces of chromosomes. mongolism , the foremost common chromosomal abnormality, is caused by an additional chromosome number 21. Chromosome abnormalities are often inherited from a parent or they will happen accidentally.

Numerical Abnormalities

Numerical refers to the very fact that there are a special number of chromosomes than expected; there might be more or less. this is often also called aneuploidy. Each scenario features a specific term:

• Monosomy: A chromosome is missing from a pair.
• Trisomy: There are three chromosomes rather than two.

Structural Abnormalities

When the makeup of the chromosome presents a problem , this is often referred to as a structural abnormality. There are several ways in which structural abnormalities may occur.

• Translocation: One piece of a chromosome is transferred to a different . (This might be a Robertsonian translocation, where one chromosome attaches itself to a different , or a reciprocal translocation, where two chromosomes are traded.)
• Deletion: there's a deleted or missing a part of a chromosome.
• Duplication: A chromosome is copied, leading to extra genetic material.
• Ring: A ring/circle forms as a results of some of a chromosome tearing off.
• Inversion: a bit of chromosome breaks off and turns the wrong way up , then reattaches itself to the first structure.

Chromosomal Disorders

There are differing types of disorders that would arise from abnormal chromosomes. the subsequent list isn't exhaustive, but rather it includes the disorders that a fetus has the simplest chance of surviving to birth and beyond.

Down Syndrome

One of the foremost well-known chromosomal disorders is mongolism (also referred to as trisomy 21), which is caused by an additional copy of chromosome 21. a number of the common traits of mongolism are developmental delay, small stature, an upward slant to the eyes, low muscular tonus , and a deep crease across the center of the palm.

Turner Syndrome

Turner syndrome (also referred to as gonadal dysgenesis), which only affects females, results when one among the X chromosomes is missing or partially missing. It could make them infertile or shorter than average. Other features of the disorder could also be swelling of feet/hands, extra neck skin, kidney and heart issues, also as skeletal abnormalities. Turner syndrome also can end in miscarriage or stillbirth.

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Klinefelter Syndrome

Klinefelter syndrome, also referred to as XXY syndrome, is that the results of an additional X chromosome in males. it's related to high rates of sterility and sexual dysfunction. It typically goes unnoticed until puberty when it's characterized by weak musculature, tall stature, little hair , and little genitalia.

Trisomy 13

Trisomy 13 or Patau syndrome, is caused by an additional copy of chromosome 13. The syndrome can cause severe intellectual disability also as heart defects, underdeveloped eyes, extra fingers or toes, a harelip, and brain or medulla spinalis abnormalities. Patau syndrome occurs in one among every 16,000 births, with infants usually dying within the primary days or weeks of life.

Trisomy 18

Trisomy 18, sometimes called Edwards syndrome, is caused by an additional copy of chromosome 18. The syndrome occurs in one among every 2,500 pregnancies and about one among every 6,000 births within the us . Edwards syndrome is characterized by low birth weight, a small, abnormally shaped head, and other life-threatening organ defects. Edwards syndrome has no treatment and is typically fatal before birth or within the primary year of life..

Chromosomal Testing

Fetal chromosomal testing is out there to expecting parents who choose it. This testing includes both screening and diagnostic tests.

Toward the top of your trimester , you'll elect to possess a screening test which will provide you and your doctor with information about the probability of your baby having a chromosomal abnormality. These tests cannot diagnose a chromosomal abnormality. Screening tests include ultrasound and blood tests, like a panel of biomarkers or testing of circulating placental DNA.

While screening tests cannot diagnose a chromosomal abnormality, there are tests which will . Diagnostic tests like villus sampling and amniocentesis are more invasive but can diagnose abnormalities that previous screening tests may have flagged.

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