What is Chromosome Deletion?

Main Image: Chromosome deletion

Our body is made of millions of cells and each of these has a nucleus which houses the chromosomes. The chromosomes have genes which have the DNA. Genes hold the key to every individual instruction that keeps our body running smoothly. DNA helps in the development and propagation of the species. A chromosome deletion can be described as a chromosome break that results in the loss of genetic code. Deletion is also known as gene deletion or deletion mutation. This can occur anywhere along the chromosome and can occur with any chromosome. Any number of nucleotides can be deleted from a single base to an entire piece of chromosome. Nucleotides are molecules that form the structural unit of DNA and RNA. The deletion of chromosomes causes many genetic disorders and serious genetic diseases.

What Causes Chromosome Deletion?

Chromosome Deletion may occur due to the following causes:
1. Breaking of the chromosome without rejoining
2. Unequal crossing over
3. Chromosomal crossovers during meiosis
4. Loss from translocation

What Are the Types of Chromosome Deletion?

There are two types of chromosome deletions:
- **Terminal Deletion** - This is a type of deletion that occurs towards the end of the chromosome. Deletion of the terminal band may cause abnormal facial or skull structure, mental retardation, and growth delay.
- **Intercalary Deletion / Interstitial Deletion** - This is a kind of deletion that occurs from the interior of a chromosome. Interstitial deletion of chromosome cause rare conditions characterized by broad forehead, small chin, prominent eyes, downslanting palpebral fissures, and a downturned mouth.

What Are the Effects of Chromosome Deletion?

Chromosomal deletions have serious to no effects on the human body. In most cases, small deletion of the chromosomes remain unrecognizable. Large deletions are generally fatal. The disorders are recognizable in case of medium sized deletions. Deletion of a number of base pairs that is not evenly divisible by three will lead to a frameshift mutation that will cause all of the codons occurring after the deletion to be read incorrectly during translation, producing a severely altered and potentially nonfunctional protein. In contrast, a deletion that is evenly divisible by three is called "in-frame deletion". Chromosome deletions are also responsible for many more serious disorders such male infertility or frigidity.

Deletion of chromosome 5p results in a syndrome called "Cri Du Chat", a French word for "cry of the cat". The infants with this disorder have a distinctive cry, shortened lifespan and mental retardation. Prader-Willi syndrome is caused by the deletion of chromosome 15. Prader-Willi syndrome is a complex genetic disorder that causes low muscle tone, incomplete sexual development, short stature, cognitive disabilities, problem behaviors, and a chronic feeling of hunger that can lead to excessive eating and life-threatening obesity.

22q11.2 Deletion Syndrome is a chromosomal disorder caused by the deletion of a small piece of chromosome 22. The symptoms of 22q11.2 include congenital heart defects, palate defects, learning disabilities, and mild differences in facial features. People with 22q11.2 Deletion Syndrome often experience recurrent infections as their immune system is weak. It may also lead to kidney abnormalities, low levels of calcium in the blood (which can result in seizures), decrease in blood platelets (thrombocytopenia), significant feeding difficulties, and hearing loss.

Wolf-Hirschhorn Syndrome is also known as the chromosome deletion 4p Syndrome or Pitt-Rogers-Danks syndrome. It is caused by the deletion of chromosome 4. Distinct symptoms include mental retardation, seizures, distinctive facial features (including a broad, flat nasal bridge and a high forehead).

How are Chromosomal Abnormalities Detected?

The chromosomal abnormalities can be detected using the molecular techniques along with cytotgenetic methods. Microarray comparative genomic hybridization with BAC clones allows detection of DNA sequence copy number changes (deletions, additions, and amplifications) on a genome-wide scale in a single hybridization. The resolution of this could be as high as >
30,000 bands and the size of chromosome deletion could be as low as 5 – 25 Kb in length.

External References:
everydayhealth.com: chromosome deletion causes
netwellness.uc.edu: FAQ on chromosome deletion

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