

## Impacts & Effects of Chromosome Abnormalities

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### ABSTRACT

Chromosome abnormalities will occur by accidentally when egg or sperm is formed during early stage of fetus. Our body have 23 pairs of chromosomes for total of 46 chromosomes; half comes from mother and another half from father. Every chromosome has thousands of genes which help to make protein that directs the body growth, development and chemical reactions.

KEYWORDS: Chromosome abnormalities, Chromosomes, Down syndrome, Genes

### INTRODUCTION

In our body, we have total 23 pairs of chromosomes and total of 46 chromosomes. Chromosomes abnormalities are many types and categorized as either numerical or structural. Chromosomal abnormality will happen when an extra pair, missing pair, upside down happened to chromosome. Common type of chromosomal abnormality is known as aneuploidy, it will happen because of an missing or extra chromosome. Breakage and incorrect rejoining of chromosomal segments gives result of Structural chromosomal abnormalities. If the complete chromosomal set is still present, though rearranged, and unbalanced if information is additional or missing then it is defined as balanced structural rearrangement. Unbalanced rearrangements include deletions, duplications, or insertions of a chromosomal segment. When a chromosome undergoes two breaks and the broken ends fuse into a circular chromosome then ring chromosomes will form. When an arm of the chromosome is missing and the remaining arm duplicates then isochromosome will form.

Common chromosomal disruptions include:

**Aneuploidy:** Chromosome number that is not a multiple of 23.

**Inversion:** Reunion of separated portion.

**Deletion:** Loss of part of chromosome.

**Polyploidy:** Chromosome number that is 3 or 4 times the haploid number of 23.

**Translocation:** Exchange of chromosome parts between non-homologous chromosomes.

**Robertsonian translocation:** Joining of long arms of two acrocentric chromosomes with loss of short arms.

**Balanced translocation:** No genetic material lost; clinically asymptomatic.

**Cri du Chat Syndrome:** It is caused by partial deletion of chromosome 5p.

Patients have severe developmental delay and cognitive deficits and distinctive facial abnormalities (round face, low-set ears, microcephaly, and a hypoplastic nasal bridge) and also patients exhibit a high-pitched cat-like cry at birth, which is usually due to structural abnormalities in the larynx.

**Down Syndrome:** It caused by Trisomy 21, 95% of cases (usually because of meiotic nondisjunction); 1% of cases are caused by mosaicism, resulting from mitotic nondisjunction of chromosome 21 during embryogenesis; 4% of cases are caused by Robertsonian translocation of the long arm of chromosome 21 to another chromosome (usually chromosome 14 or 22).

**Edward Syndrome:** It caused by trisomy 18. It also caused by mosaicism, resulting from mitotic nondisjunction of chromosome 18 during embryogenesis. It includes many Characteristics like rocker-bottom feet; Severe mental retardation; specific facial features include prominent occiput, micrognathia (small jaw), congenital heart and renal defects; overlapping third and fourth fingers; low-set ears.

**Klinefelter syndrome:** This disorder caused by characterized by two or more X chromosomes with one or more Y chromosomes and other causes include mosaicism or paternal meiotic

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nondisjunction. It have characteristics like Male infertility, often resulting from reduced spermatogenesis. Occasionally associated with mild mental retardation and Small atrophic testes; tall stature; lack of secondary male characteristics and gynecomastia.

**Patau Syndrome:** It caused by trisomy 13 and it have Characteristics like polydactyly; congenital heart and renal defects; umbilical hernia; rocker-bottom feet.