

# CHROMOSOMAL DISORDERS

**BY:**

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# गुणसूत्र विकार गुणसूत्रों की संख्या या संरचना में परिवर्तन के परिणामस्वरूप होता है।

Deoxyribonucleic acid (DNA) is a nucleic acid that contains the genetic instructions used in the development and functioning of all known living organisms and some viruses. The main role of DNA molecules is to store the long-term storage of information. DNA is often compared to a set of blueprints or a recipe, or a code, since it contains the instructions needed to construct other components of cells, such as proteins and RNA molecules. The DNA segments that carry this genetic information are called genes, but other DNA sequences have structural purposes, or are involved in regulating the use of this genetic information.

Chemically, DNA consists of two long polymers of simple units called nucleotides, with backbones made of sugars and phosphate groups joined by ester bonds. These two strands run in opposite directions to each other and are therefore anti-parallel. Attached to each sugar is one of four types of molecules called bases. It is the sequence of these four bases along the backbone that encodes genetic information. This information is read using the genetic code, which specifies the sequence of the amino acids within proteins. The code is read by copying stretches of DNA into the related nucleic acid RNA, in a process called transcription.

Within cells, DNA is organized into long structures called chromosomes. These chromosomes are duplicated before cells divide, in a process called DNA replication. Eukaryotic organisms (animals, plants, fungi, and protists) store most of their DNA inside the cell nucleus and some in mitochondria. In contrast, prokaryotes (bacteria and archaea) store their DNA only in the cytoplasm. Within the chromosomes, chromatin proteins such as histones compact and organize DNA. These compact structures provide the mitotic spindle between DNA and other proteins, helping control which parts of the DNA are transcribed.

DNA is a double helix structure. The two strands are made of nucleotides. Each nucleotide is composed of a phosphate group, a sugar, and a nitrogenous base. The phosphate group is attached to the sugar, and the sugar is attached to the nitrogenous base. The nitrogenous base is attached to the other strand, forming the rungs of the ladder. The two strands are twisted around each other, forming a helix. The distance between two rungs is 0.34 nm. The length of one full rotation is 3.4 nm. The diameter of the helix is 2 nm. The length of the human genome is approximately 2.2 billion base pairs long.

DNA exists in many possible conformations that include A-DNA, B-DNA, and Z-DNA. Forms so far thought only B-DNA and Z-DNA have been directly observed in nature. The conformation of DNA sequence depends on the hydration level, chemical environment, and direction of polymerization. Modifications at the base, the type and concentration of metal ions, as well as the presence of polyanions in solution [29].

The first published reports of A-DNA X-ray diffraction patterns—and also B-DNA—were based on the Patterson transform that provided only a limited amount of structural information. A detailed picture of DNA (B-DNA) in alternate analysis was then provided by Wilkins et al. in 1952, for the first time. X-ray diffraction/scattering patterns of highly hydrated DNA fibers in terms of squares of basal functions [32] in the same journal, Watson and Crick presented their molecular modeling analysis of the DNA. X-ray diffraction patterns to suggest that the structure was a double helix [2].

Although the B-DNA form is most common under the conditions found in cells, it is not a well-defined conformation but a family of related DNA conformations [33] that occur at the high hydration levels present in living cells. Their corresponding X-ray molecular scattering patterns are characteristic of disordered [35] [36].

Compared to B-DNA, the A-DNA form is a wider, right-handed spiral, with a shallow, wide minor groove and a narrower, deeper major groove. The A form is produced in highly concentrated solutions of DNA, while in the cell it may be produced in highly concentrated solutions of DNA and RNA strands, as well as in enzyme-DNA complexes [37] [38]. In naturally occurring DNA, most phosphates [37] [38], chemically modified by methylation may undergo a larger change in conformation and adopt the Z form.

random | plasmid

# Karyotype :-

गुणसूत्रों का एक सेट, जैसा कि एक माइक्रोस्कोप के नीचे देखा जाता है, एक करियोटाइप के रूप में जाना जाता है।

random][plasmid

Deoxyribonucleic acid (DNA) is a nucleic acid that contains the genetic instructions used in the development and functioning of all known living organisms and some viruses. The main role of DNA molecules is the long-term storage of information. DNA is often compared to a set of blueprints or a recipe, or a code, since it contains the instructions needed to construct other components of cells, such as proteins and RNA molecules. The DNA genes, but other DNA sequences have structural purposes, or are involved in regulating the use of this genetic information.

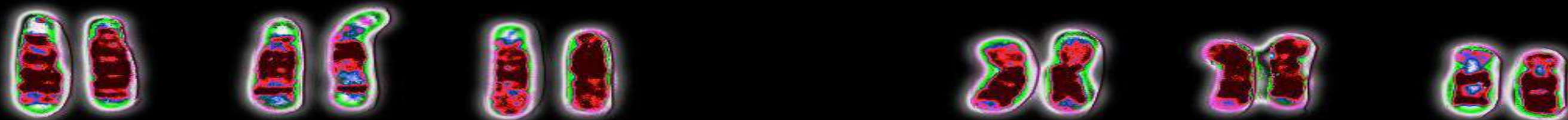
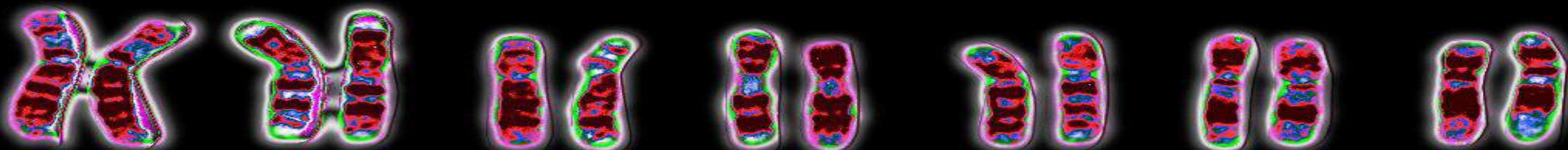
Chemically, DNA consists of two long polymers of simple units called nucleotides, with backbones made of sugars and phosphate groups joined by ester bonds. These two strands run in opposite directions to each other and are therefore anti-parallel. Attached to each sugar is one of four types of molecules called bases. It is the sequence of these four bases along the backbone that encodes information. This information is read using the genetic code, which specifies the sequence of the amino acids within proteins. The code is read by copying stretches of DNA into the related nucleic acid RNA, in a process called transcription.

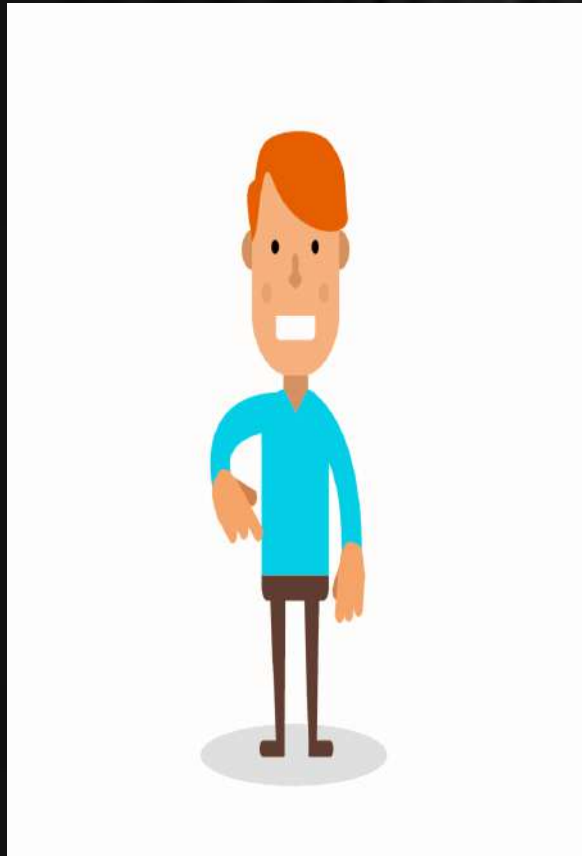
Within cells, DNA is organized into long structures called chromosomes. These chromosomes are duplicated before cells divide, in a process called DNA replication. Eukaryotic organisms (animals, plants, fungi, and protists) store most of their DNA inside the cell nucleus and some in organelles, such as mitochondria or chloroplasts. In contrast, as prokaryotes (bacteria and archaea) store their DNA only in the cytoplasm. Within the chromosomes, chromatin proteins such as histones compact and organize DNA. These compact structures guide the interactions between DNA and other proteins, helping control which parts of the DNA are transcribed.

DNA is a long, thin, double-stranded molecule. Each strand is made of a sugar-phosphate backbone with nitrogenous bases attached to the sugar. The two strands are twisted around each other, forming a double helix. The length of a DNA molecule is approximately 2.2 meters long, but it is only 2.5 nanometers wide. The DNA molecule is packaged into chromosomes, which are visible under a light microscope.

DNA exists in many possible conformational states. The most common is the B-DNA form, which is a right-handed helix. Other forms include A-DNA, Z-DNA, and H-DNA. The conformation of DNA is determined by the sequence of its bases and the presence of various proteins and other molecules. The B-DNA form is the most common and is found in most living cells. It is a well-defined conformation that is stable under physiological conditions. The A-DNA form is a compact, wide, shallow, wide minor groove. The Z-DNA form is a narrow, deep, narrow groove. The H-DNA form is a four-stranded structure. The conformation of DNA is also affected by environmental factors such as temperature, pH, and the presence of various ions and molecules.

Although the 'B-DNA form' is most common, conditions found in cells, such as high salt concentrations, can induce DNA to adopt other conformations. The A-DNA form is a compact, wide, shallow, wide minor groove. The Z-DNA form is a narrow, deep, narrow groove. The H-DNA form is a four-stranded structure. The conformation of DNA is also affected by environmental factors such as temperature, pH, and the presence of various ions and molecules. The B-DNA form is the most common and is found in most living cells. It is a well-defined conformation that is stable under physiological conditions. The A-DNA form is a compact, wide, shallow, wide minor groove. The Z-DNA form is a narrow, deep, narrow groove. The H-DNA form is a four-stranded structure. The conformation of DNA is also affected by environmental factors such as temperature, pH, and the presence of various ions and molecules.





**46  
Chromosomes  
or 23 in pairs**



**Somatic  
cells**



**Homologous  
chromosomes  
(2n) / 44 in no.**



**Germ  
cells**



**Sex  
chromosomes  
(n) / 2 in no.**

# **Numerical abnormalities**

(संख्यात्मक असामान्यताएं)

<b>Syndrome</b>	<b>Abnormality</b>
<b>Klinefelter's</b>	<b>XXY</b>
<b>Turner</b>	<b>Monosomy X</b>
<b>XYY</b>	<b>XYY</b>
<b>XXX</b>	<b>XXX</b>
<b>Down's</b>	<b>Trisomy 21</b>
<b>Edwards'</b>	<b>Trisomy 18</b>
<b>Patau's</b>	<b>Trisomy 13</b>

# Klinefelter syndrome (44+XXY = 47)

Additional X chromosome leading to male hypogonadism



- In males
- Reduced muscle mass
- Less facial and body hair
- Broad hips
- Enlarged breasts
- Increased belly fat

- नर में होता है
- मांसपेशियों में कमी
- चेहरे और शरीर में कम बाल
- चौड़े नितम्ब
- बड़े हुए स्तन
- पेट की बढ़ी चर्बी

## Turner's syndrome ( $44 + x0 = 45$ )



- एक गुणसूत्र विकार जिसमें एक महिला केवल एक एक्स गुणसूत्र के साथ पैदा होती है।

- **In female with limited secondary characters**

- **Sterile**

- **Webbed neck**

- **Short stature**

- **Swollen hands and feet.**

- सीमित द्वितीयक शारीरिक वृद्धि वाली महिला

- बाँझ

- झिल्लीदार गर्दन

- छोटा कद

- हाथ-पैर फूल हुए

# Jacob's / XYY / Criminal syndrome ( 44 + XYY = 47 )

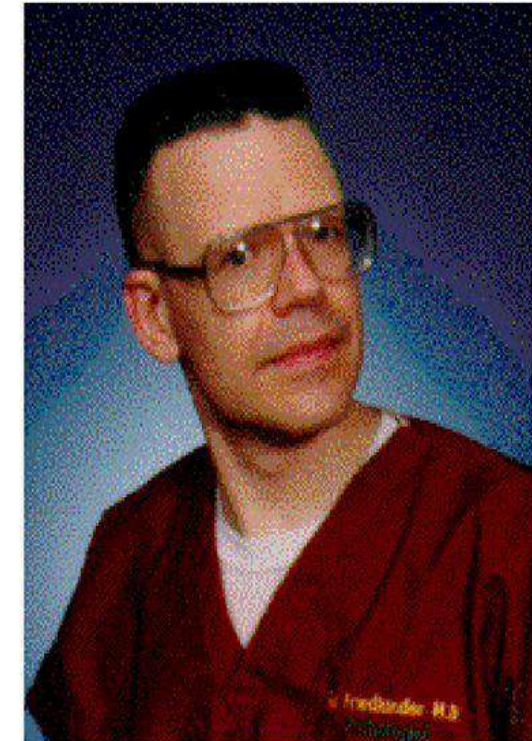
## Jacob's Syndrome

- ✿ Normal physically
- ✿ Acne
- ✿ Increased testosterone; can be more aggressive
- ✿ Learning delays



## Super Male Syndrome

- Nondisjunction of sex chromosomes ending up with XYY
- Characteristics
  - More aggressive
  - Tall
  - Acne



- टेस्टोस्टेरोन हॉर्मोन ज्यादा स्रावित होने के कारण ज्यादा आक्रामक
- सामान्य से ज्यादा लम्बा कद
- मुहासे
- नई विधा सीखने में ज्यादा समय लगना

## Super female / XXX / Multiple X Syndrome

( 44 + XXX = 47, 48, 49 )

- **Tall** (लम्बी)
- **thin** (पतली)
- **mentally retarded** (मानसिक रूप से मंद)
- **often sterile** (अक्सर बाँझ)

## Down syndrome ( trisomy 21 )



- **A genetic chromosome 21 disorder causing developmental and intellectual delays.**
- एक आनुवंशिक गुणसूत्र 21 विकार जिसके कारण विकासात्मक और बौद्धिक देरी होती है।

- **Developmental: delayed development, learning disability, short stature, or speech delay in a child.**
- विकासात्मक: विलंबित विकास, सीखने की विकलांगता, छोटे कद, या बच्चे में भाषण में देरी .
- **This is also known as “Mangolism”.**
- इसे "मंगोलिस्म" के रूप में भी जाना जाता है।

## Edwards' syndrome (trisomy 18)

**Edwards' syndrome, also known as trisomy 18, is a rare but serious condition.**

एडवर्ड्स सिंड्रोम, जिसे ट्राइसॉमी 18 भी कहा जाता है, एक दुर्लभ लेकिन गंभीर स्थिति है।

**Edwards' syndrome affects how long a baby may survive. Sadly, most babies with Edwards' syndrome will die before or shortly after being born.**

एडवर्ड्स का सिंड्रोम प्रभावित करता है कि बच्चा कितनी देर तक जीवित रह सकता है। अफसोस की बात है कि एडवर्ड्स सिंड्रोम वाले अधिकांश बच्चे जन्म लेने के कुछ समय पहले या बाद में मर जाएंगे।



# Patau syndrome ( Trisomy 13 )

- **A condition in which a person has an extra chromosome 13.**
- ऐसी स्थिति जिसमें एक व्यक्ति में एक अतिरिक्त गुणसूत्र 13 होता है।
- **Prenatal testing can detect Patau syndrome during pregnancy.**
- प्रसवपूर्व परीक्षण गर्भावस्था के दौरान पटौ सिंड्रोम का पता लगा सकता है।
- **Patau syndrome causes severe intellectual disability and physical defects.**
- पटौ सिंड्रोम गंभीर बौद्धिक विकलांगता और शारीरिक दोष का कारण बनता है।



## □ Symptoms include:

□ **Cleft lip or palate** (फांक होंठ या तालु)

□ **Clenched hands (with outer fingers on top of the inner fingers).**

□ भीतरी उंगलियों के शीर्ष पर बाहरी उंगलियों के साथ

□ **Close-set eyes** (आंखें निकट होना )

□ **Decreased muscle tone.** ( मांसपेशियों की टोन में कमी )

□ **Extra fingers or toes (polydactyly)**  
(पॉलीडेक्टली)

उंगलियां या पैर में अतिरिक्त उंगलियां

□ **Hernias** ( हर्निया )

□ **Low-set ears** ( कान का छोटा होना )