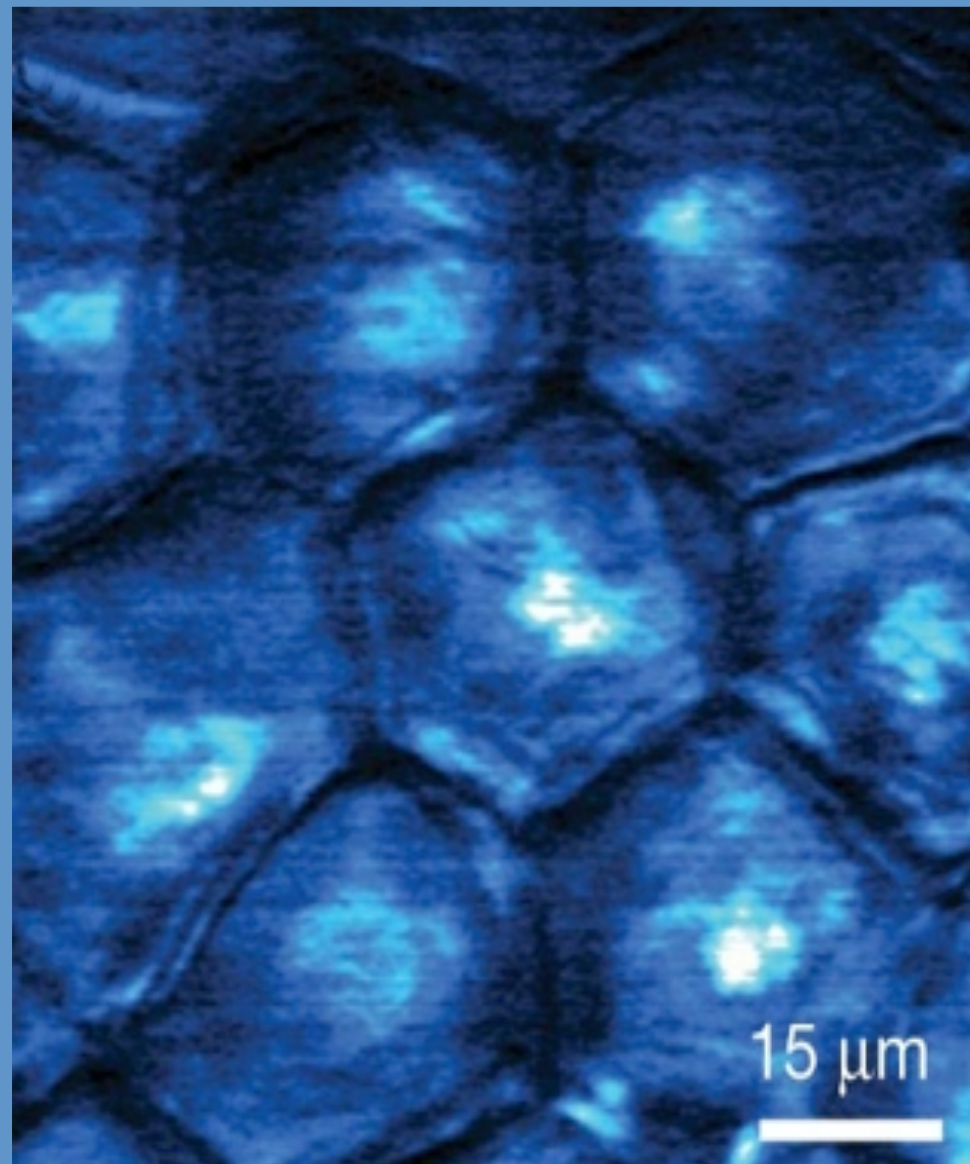


# GENES AND CHROMOSOMES II

## Lecture 4

BIOL 266/2

2014-15



2

# GENE AND THE GENOME



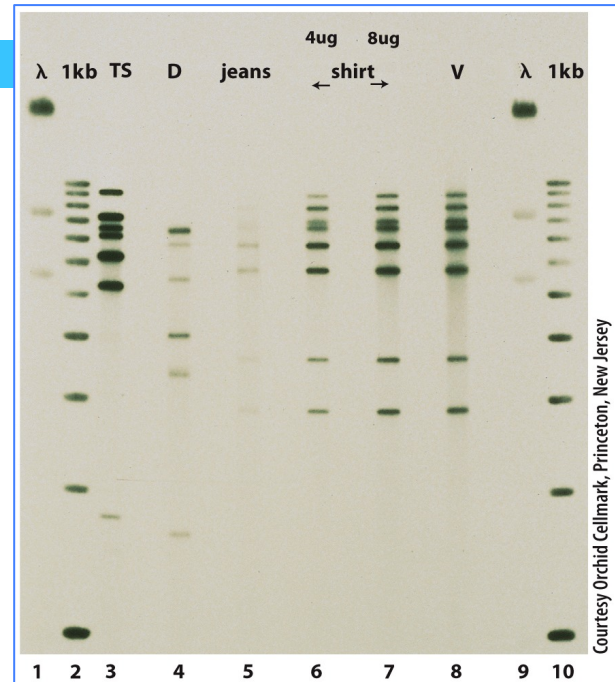
Science and Society/SuperStock

# The Structure of the Genome

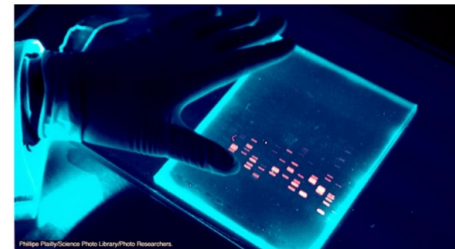
## DNA fingerprinting

3

- **DNA fingerprinting:** DNA-based identification system that relies on genetic differences among individuals or organisms.
- DNA is digested by *restriction endonucleases* and DNA fragments are size separated by gel electrophoresis
- Applications of DNA fingerprinting include forensic cases, paternity disputes, anthropology and wild life management etc.



DNA FINGERPRINTING AND FORENSIC APPLICATIONS



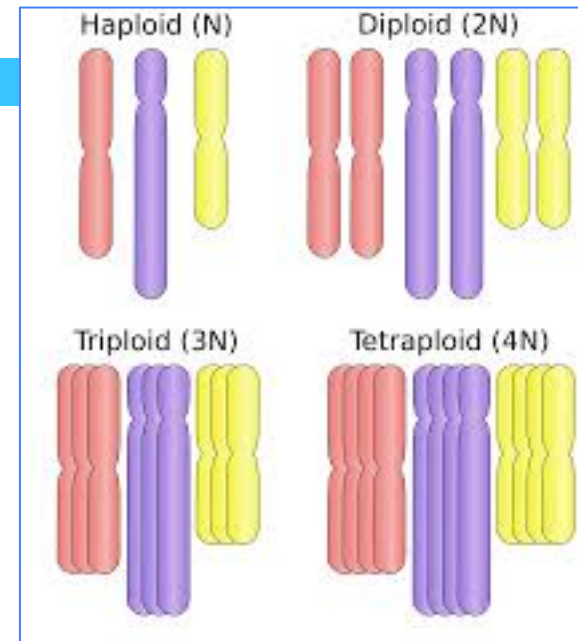
# The Stability of the Genome

## Duplication and Modification of DNA Sequences

4

### 1. Whole Genome Duplication (Polyploidization)

- Occurs when an offspring receive more than two sets of chromosomes from their parents.
- Could result from duplicate chromosomes not properly separated in embryonic cells.
- Found in agricultural crops to multiply their characters e.g., quantity of juice and size etc.



A sample of agricultural crops that are polyploid

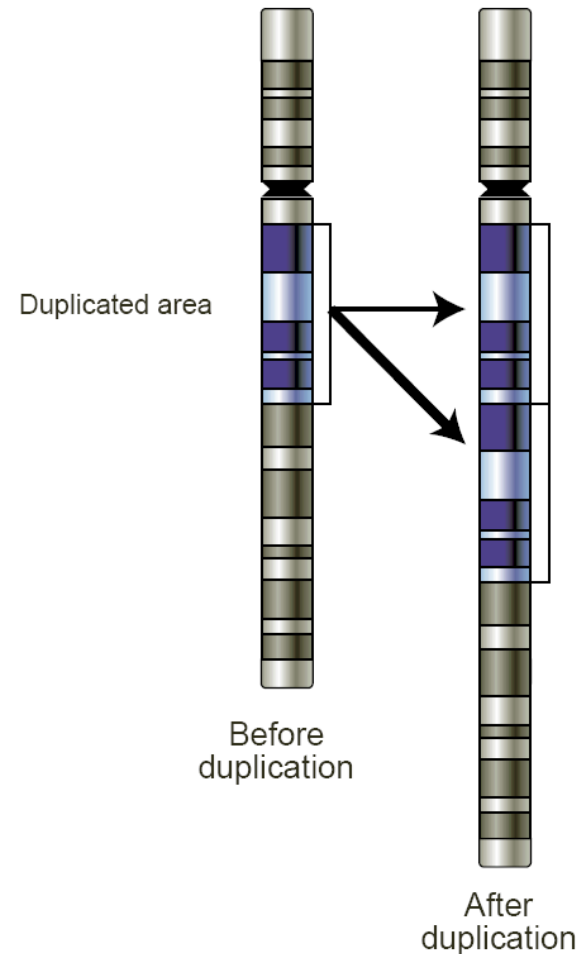
# The Stability of the Genome

## Duplication and Modification of DNA Sequences

5

### 2. Gene Duplication

- occurs within a small portion of a single chromosome.
- occurs by unequal crossing over between misaligned homologous chromosomes.
- has played a major role in the evolution of multigene families.



# The Stability of the Genome

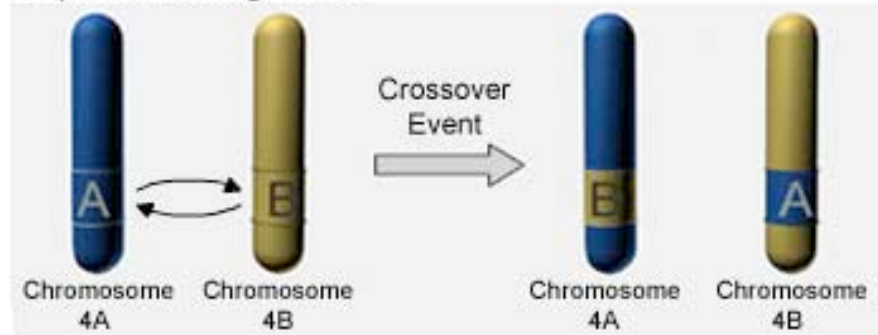
## Duplication and Modification of DNA Sequences

6

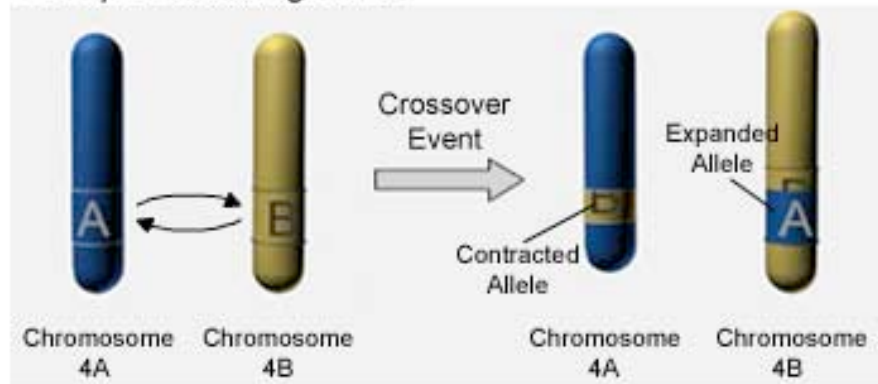
Unequal crossing over between duplicated genes provides a mechanism for generating changes in gene number

Figure Q-3: The Unequal Crossing Over Model

*Equal Crossing Over:*

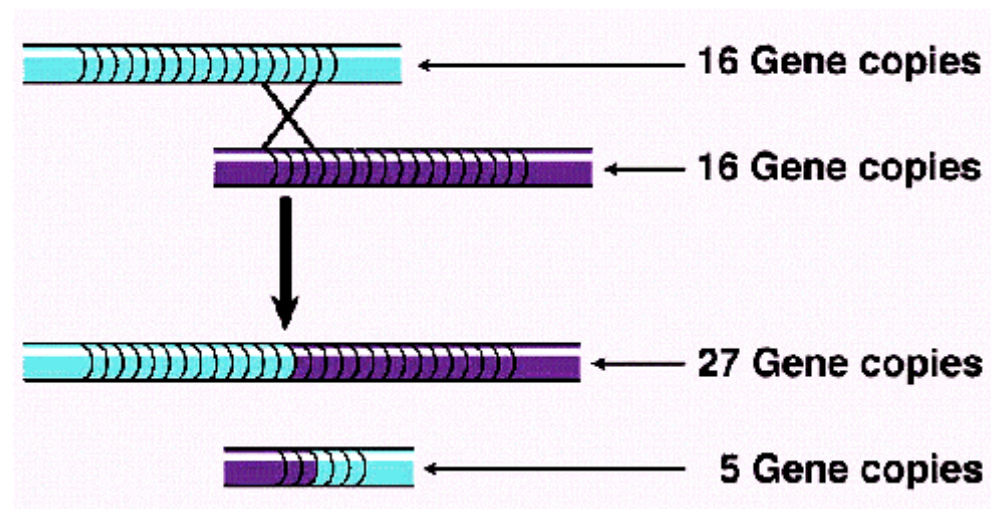


*Unequal Crossing Over:*



In equal crossing over, the entire segment of allele A switches places with the entire segment of allele B. But in unequal crossing over, only part of B switches places, leaving the rest behind to add to the length of B. The result is a shorter (contracted) segment B on chromosome 4A and a larger (expanded) segment comprised of all of A and part of B on chromosome 4B.

## Amplification of copy number by unequal crossing-over



Unequal crossing-over becomes more likely with increased copy number

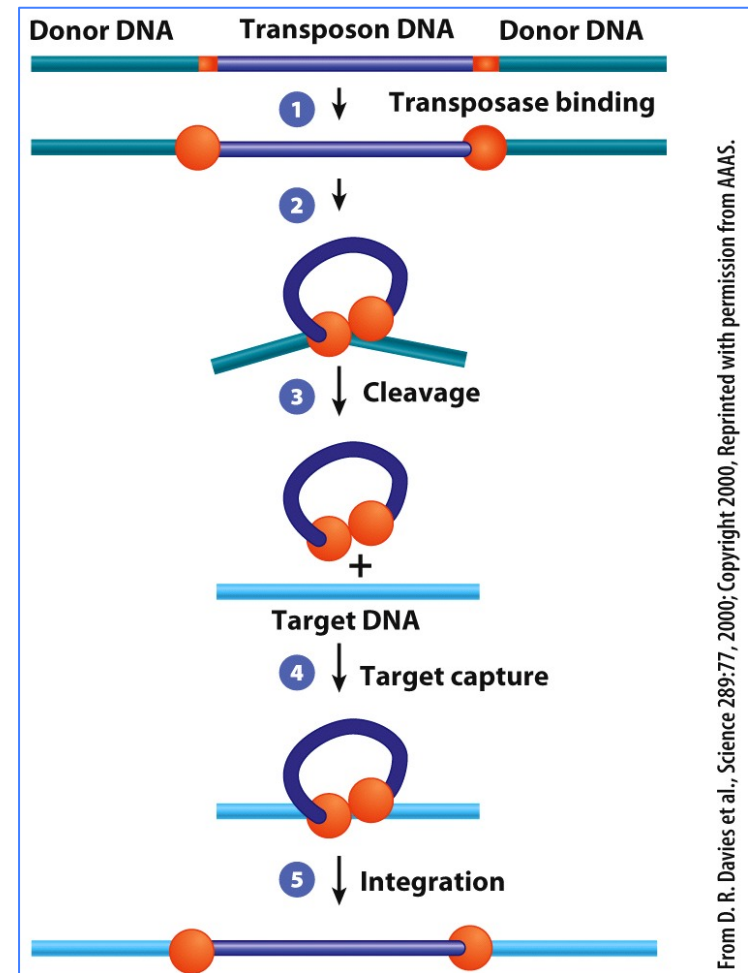
# The Stability of the Genome

## Transposition

8

### “Jumping Genes” and the Dynamic Nature of the Genome

- Genetic elements are capable of moving within a chromosome (transposition)
- Those mobile elements are called transposable elements.
- Some moderately repeated sequences in human DNA (*Alu* and *L1*) are transposable elements.



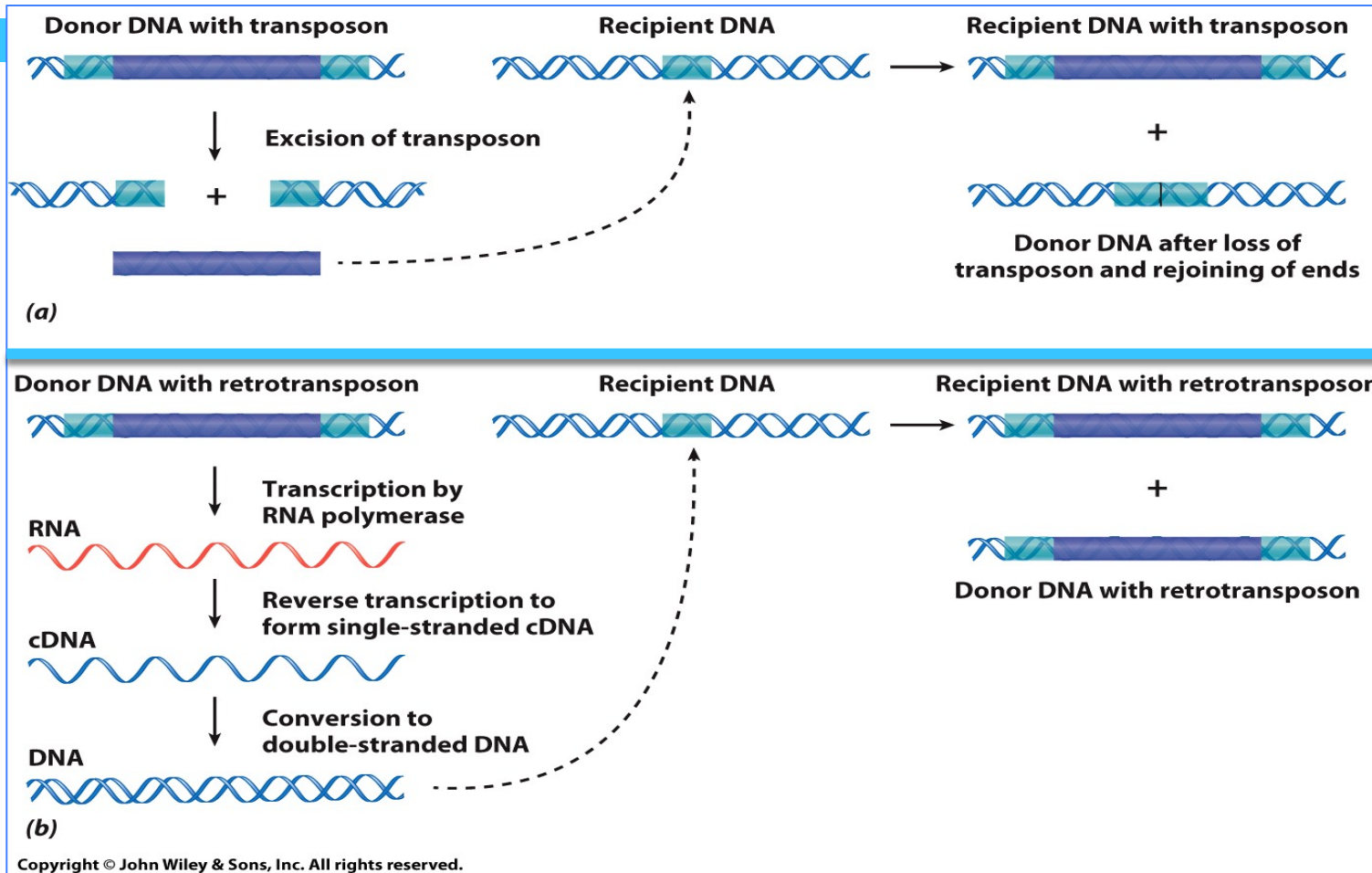
Transposition of a bacterial transposon by a “cut-and-paste” mechanism



# The Stability of the Genome

## Pathways in the movement of transposable elements

9



**Retrotransposons** use an RNA intermediate which produces a complementary DNA via reverse transcriptase

# DNA Sequence Variation

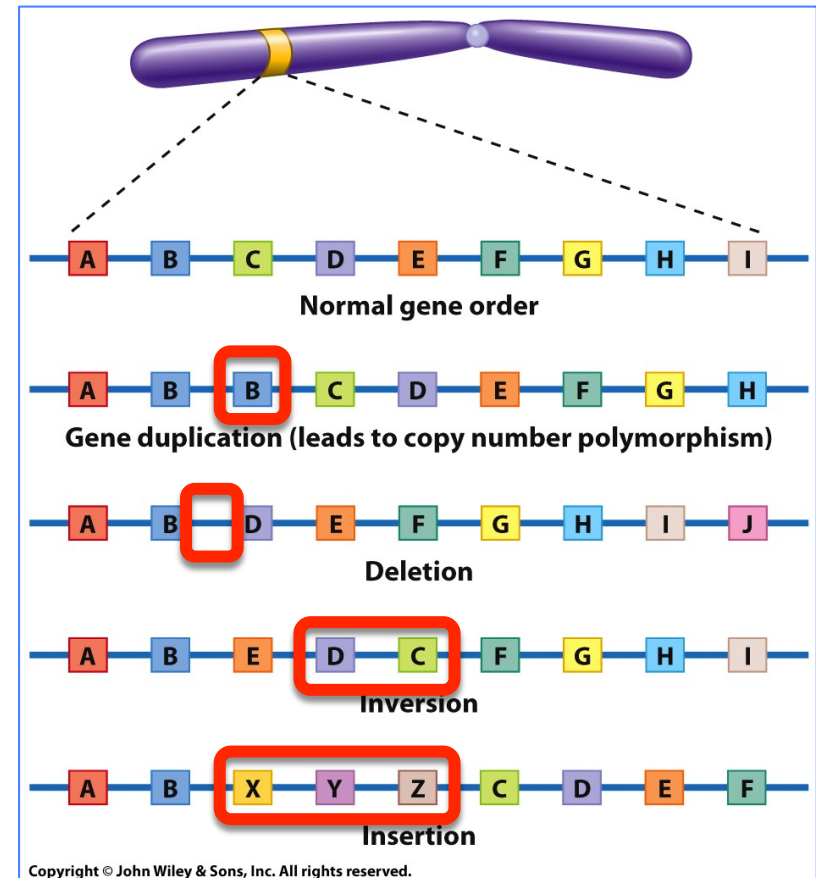
## Genetic Variation within the Human Species Population

10

Genome varies among different individuals due to genetic polymorphisms.

### 1. DNA Sequence Variation

- Most common variability among humans is at the single nucleotide difference *known as single nucleotide polymorphism (SNP)*



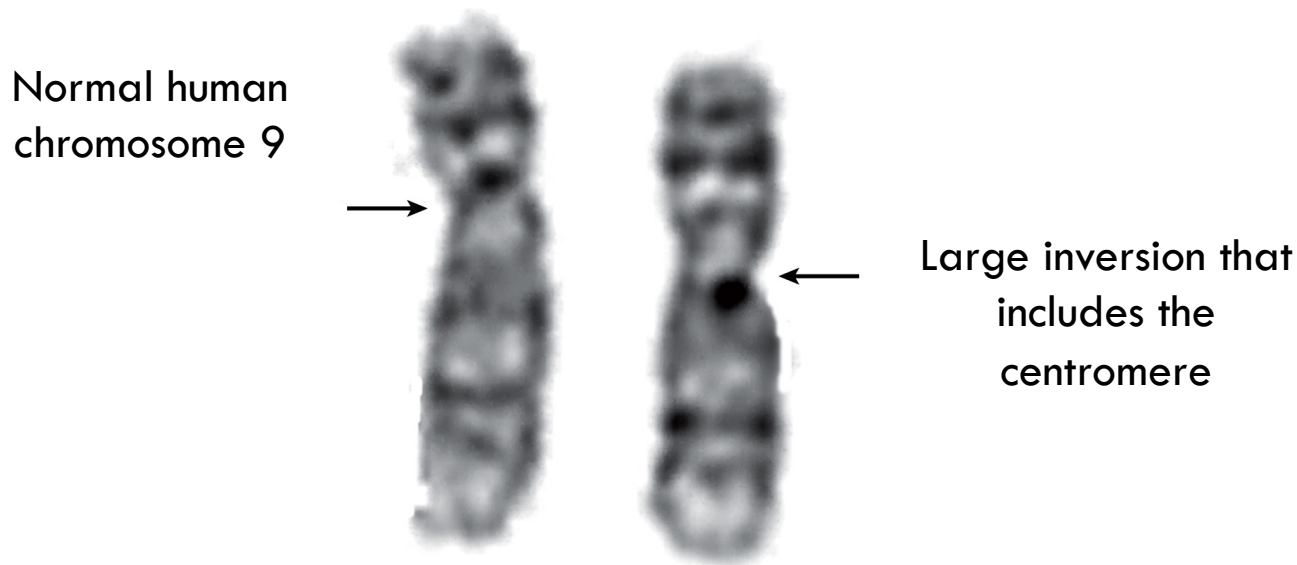
# DNA Sequence Variation

## Genetic Variation within the Human Species Population

11

### 2. *Structural Variation*

- Large segments of the DNA (structural variants) can change.

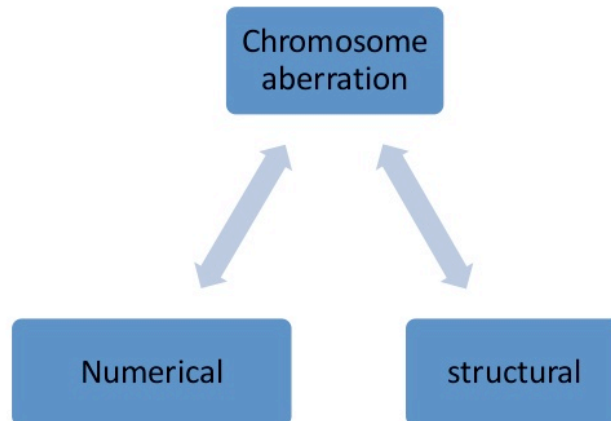


From Charles Lee, *Nature Genetics* 37:661, 2005.  
Reprinted by permission from Macmillan Publishers Ltd.

# Chromosomal Aberrations: Structural Variations

12

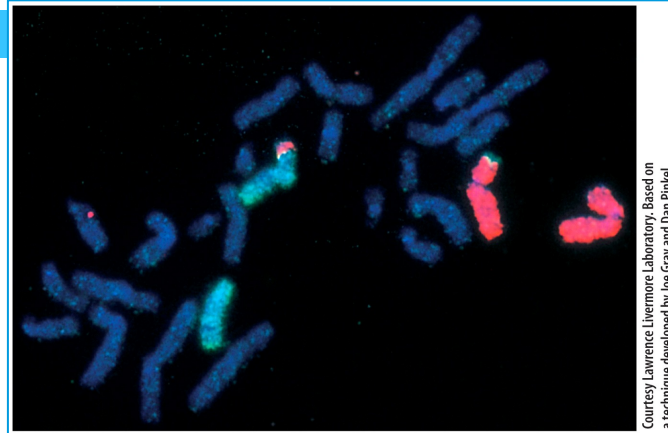
- A chromosomal aberration is loss or exchange of a segment between different chromosomes,
- Can be caused by exposure to DNA-damaging agents.
- Chromosomal aberrations have different consequences depending on whether they are in somatic or germ cells.



Variation in chromosome structure or number is called chromosomal aberration

# Chromosomal Aberrations: Structural Variations

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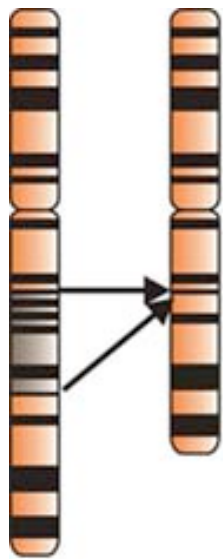
Courtesy Lawrence Livermore Laboratory. Based on a technique developed by Joe Gray and Dan Pinkel

**Translocation.** Exchange between **chr12** (bright blue) and **chr7** (red) in human cells

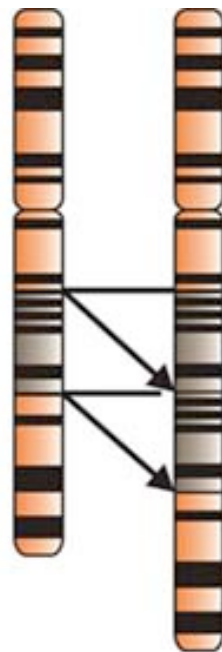
1. **Deletions** result when there is loss of a portion of a chromosome.
2. **Duplications** occur when a portion of a chromosome is repeated.
3. **Inversions** involve the breakage of a chromosome and resealing of the segment in a reverse order.
4. **Translocations** are the result of the attachment of all or one piece of one chromosome to another chromosome.

# Chromosomal Aberrations: Structural Variations

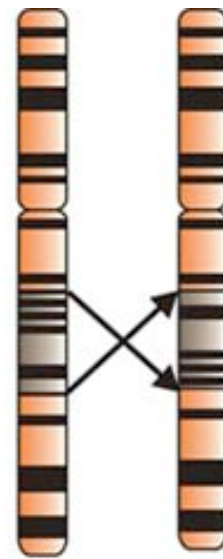
14



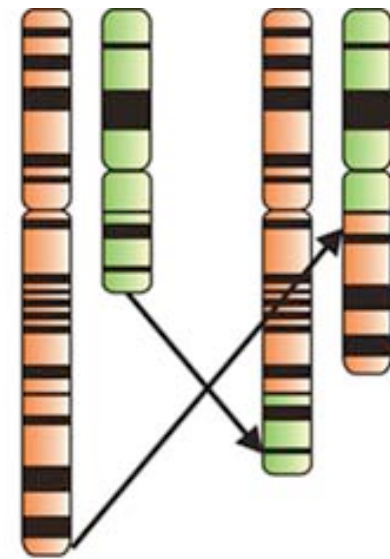
Deletion



Duplication



Inversion



Translocation

# Some interesting links

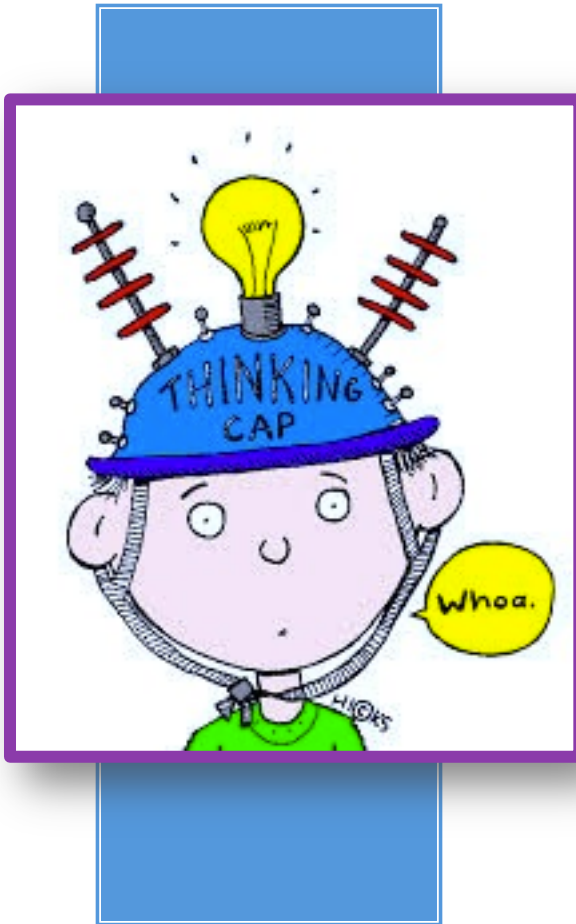
15

- <https://www.youtube.com/watch?v=xUrlreMaUrs>
- <https://www.youtube.com/watch?v=op7Z1Px8oO4>
- [https://www.youtube.com/watch?v=eig0p\\_tC\\_c4](https://www.youtube.com/watch?v=eig0p_tC_c4)



# Put your thinking cap on....

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1. If 30% of the bases on a single strand of a DNA is T, then 30% of the bases on that strand is A. True or False and why?
2. Given the sequence of one strand of a DNA helix (below), provide the sequence of the complementary strand and label the 5' and 3' ends.  
**5' -GCATTCGTGGGTAG-3'**
3. Transposase is an enzyme that \_\_\_\_\_.
  - a) degrades transposons
  - b) builds transposons
  - c) catalyzes transposon excision from a donor DNA site and its subsequent insertion at a target DNA site
  - d) degrades target DNA
  - e) rearranges transposon DNA