Chromosomal Mutations

Changes in Chromosome Number or Structure

PRESENTED BY
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Chromosome Mutations

- A chromosome mutation is a change in the structure or arrangement of the chromosomes
- Mutations are caused by
- **Physical agents** e.g. X-rays and ultraviolet light
- **Chemical mutagens** such as nitrous acid or
- **By spontaneous** way by unequal crossing over.

Alterations in Chromosome

- **Structural changes**
  - DELETION
  - DUPLICATION
  - TRANSLOCATION
  - INVERSIONS
- **NUMERICAL CHANGES**
  - **Aneuploidy**

Excess or Deficiency in a **single** chromosome

- EUPLOIDY.

Excess or Deficiency complete one or more sets of chromosomes

TYPES OF Aneuploidy

- Monosomy (2n-1)
- Nullisomy
- Trisomy(2n+1)
- Tetrasomy (2n+2)

Human Chromosomal Aneuploids

Autosomal Aneuploids

Down Syndrome	Trisomy 21
Edward Syndrome	Trisomy 18
Patau Syndrome	Trisomy 13

Trisomy: three copies of one chromosome

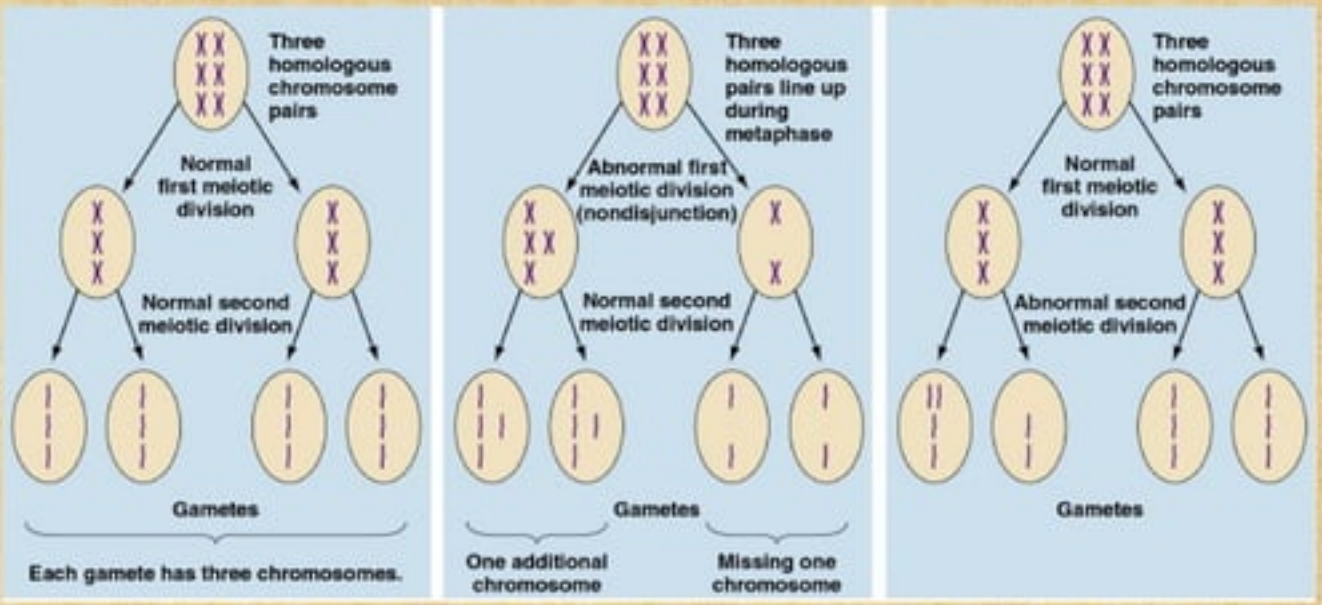
TABLE 8.1

Terminology for Variation in Chromosome Numbers

Term	Explanation
Aneuploidy	$2n \pm x$ chromosomes
Monosomy	$2n - 1$
Disomy	$2n$
Trisomy	$2n + 1$
Tetrasomy, pentasomy, etc.	$2n + 2$, $2n + 3$, etc.
Euploidy	Multiples of n
Diploidy	$2n$
Polyploidy	$3n$, $4n$, $5n$, . . .
Triploidy	$3n$
Tetraploidy, pentaploidy, etc.	$4n$, $5n$, etc.
Autopolyploidy	Multiples of the same genome
Allopolyploidy (Amphidiploidy)	Multiples of closely related genomes

Aneuploidy

- Arises by Non-disjunction
- Non-disjunction = failure of homologues or chromatids to separate during meiosis

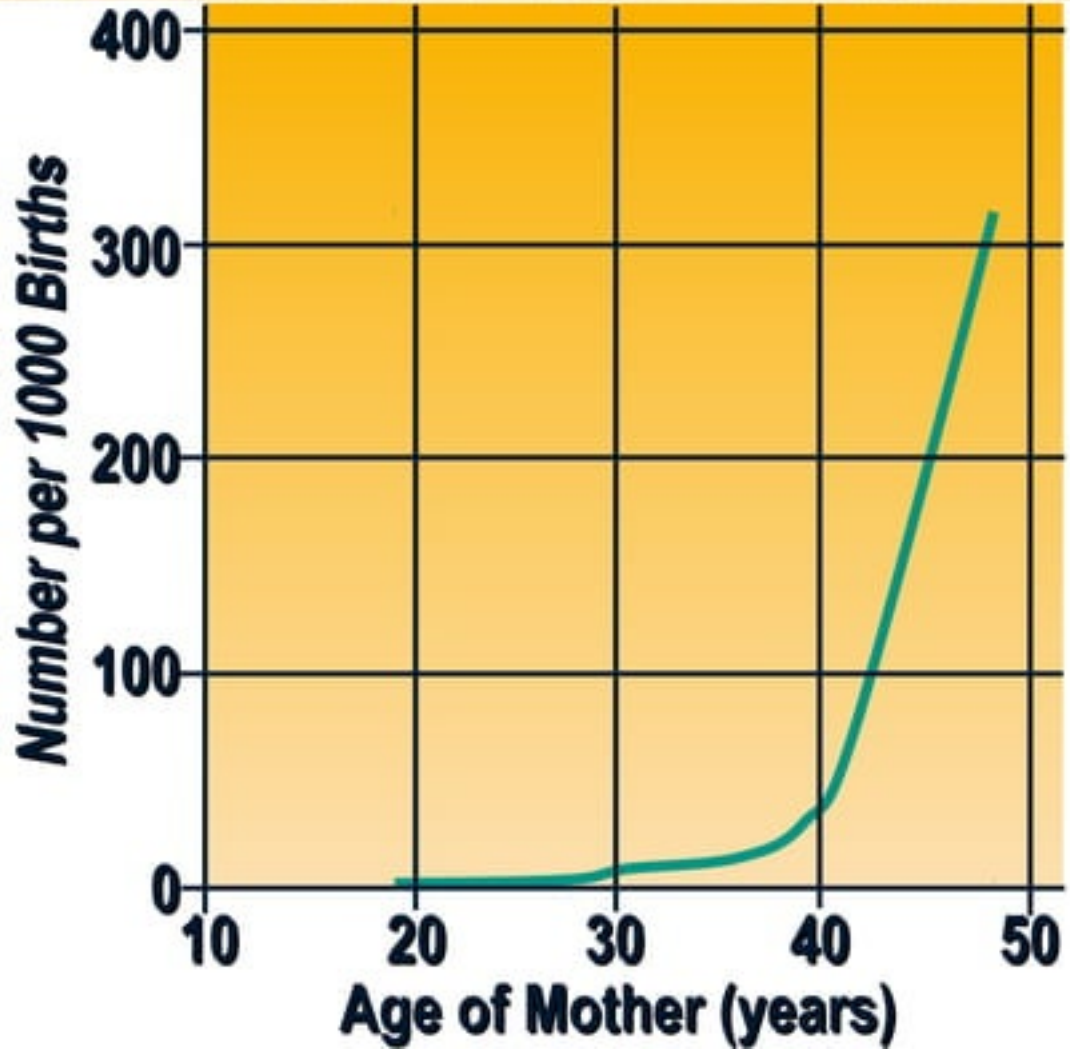


Normal Meiosis

Non-disjunction in Meiosis I

Non-disjunction in Meiosis II

Incidence of Down Syndrome Increases with Maternal Age



Human Chromosomal Aneuploids

Sex Chromosome Aneuploids

Turner Syndrome	45, XO	Sterile female
Triplo-X	47, XXX	Fertile female
Klinefelter Syndrome	47, XXY	Sterile male
XYY Syndrome	47, XYY	Fertile male

Applying Knowledge

Lets determine how many Barr bodies would be found in each cell of someone with:

Turner Syndrome	45, XO	0
Triplo-X	47, XXX	2
Klinefelter Syndrome	47, XXY	1
XYY Syndrome	47, XYY	0

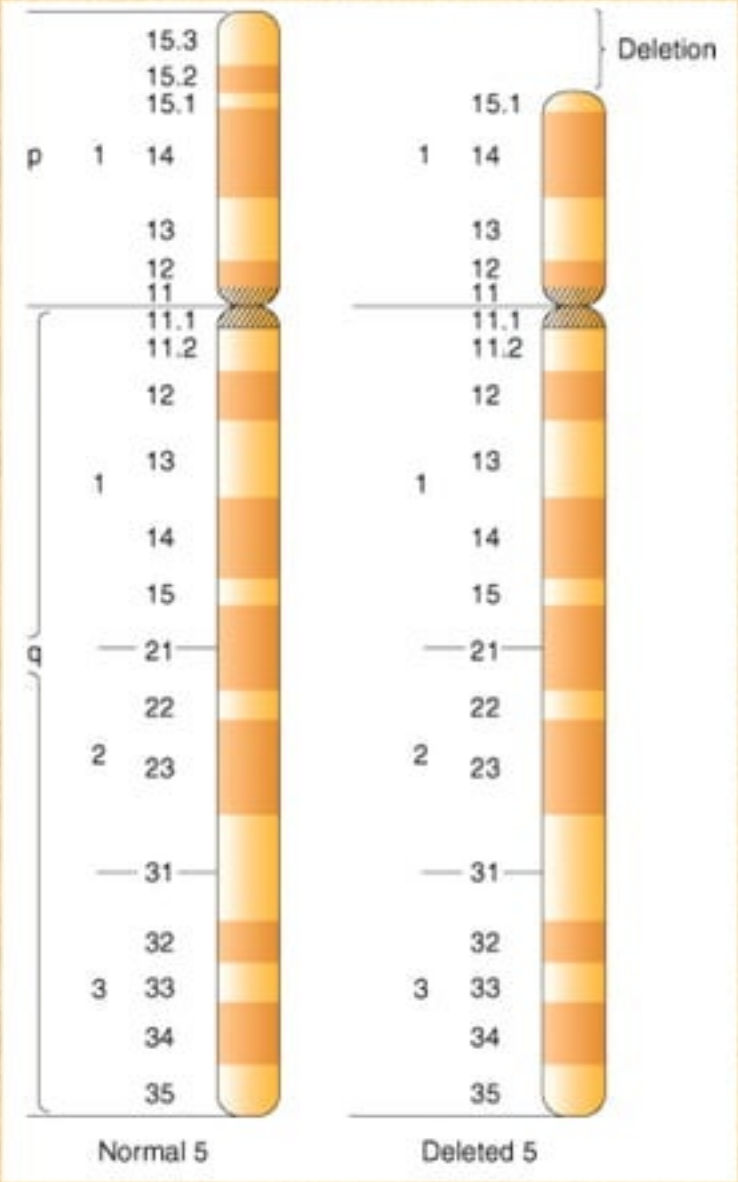
Euploidy

- Excess or Deficiency in the number of the **entire** chromomosomal complement

- Monoploid
- Diploid
- Triploid
- Tetraploid

Chromosome Structure Changes

Change	Description
Deletion	Loss of a chromosomal segment can occur terminally or internally
Duplication	Repeat of a chromosomal segment
Translocation	Movement of chromosomal segment to non-homologous chromosome or genes from one linkage group transferred to another
Inversion	Reversal of a chromosomal segment (rotated 180°)

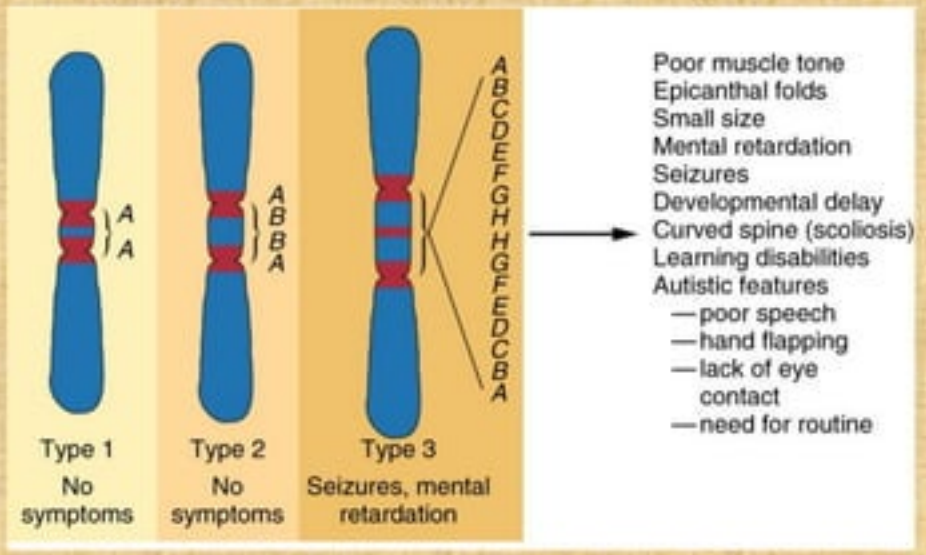


Chromosome Deletion in Humans

- Cri-du-chat syndrome is correlated with a deletion at the end of chromosome 5
- Deleterious effects, pseudosominance, absence of crossing over etc

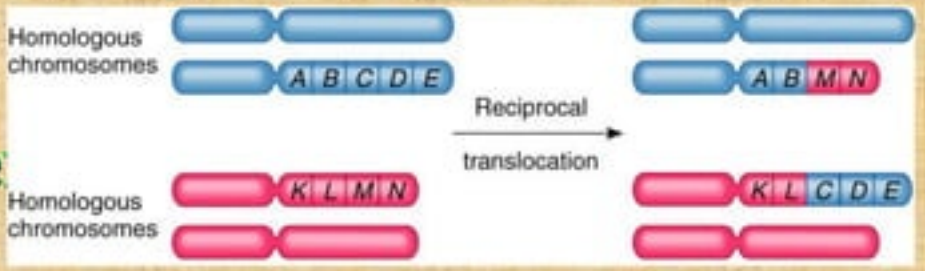
Chromosome Duplication in Humans

- Small duplications in chromosome 15 cause no symptoms and no deleterious effects
- Large duplication (with inversion) causes mental retardation

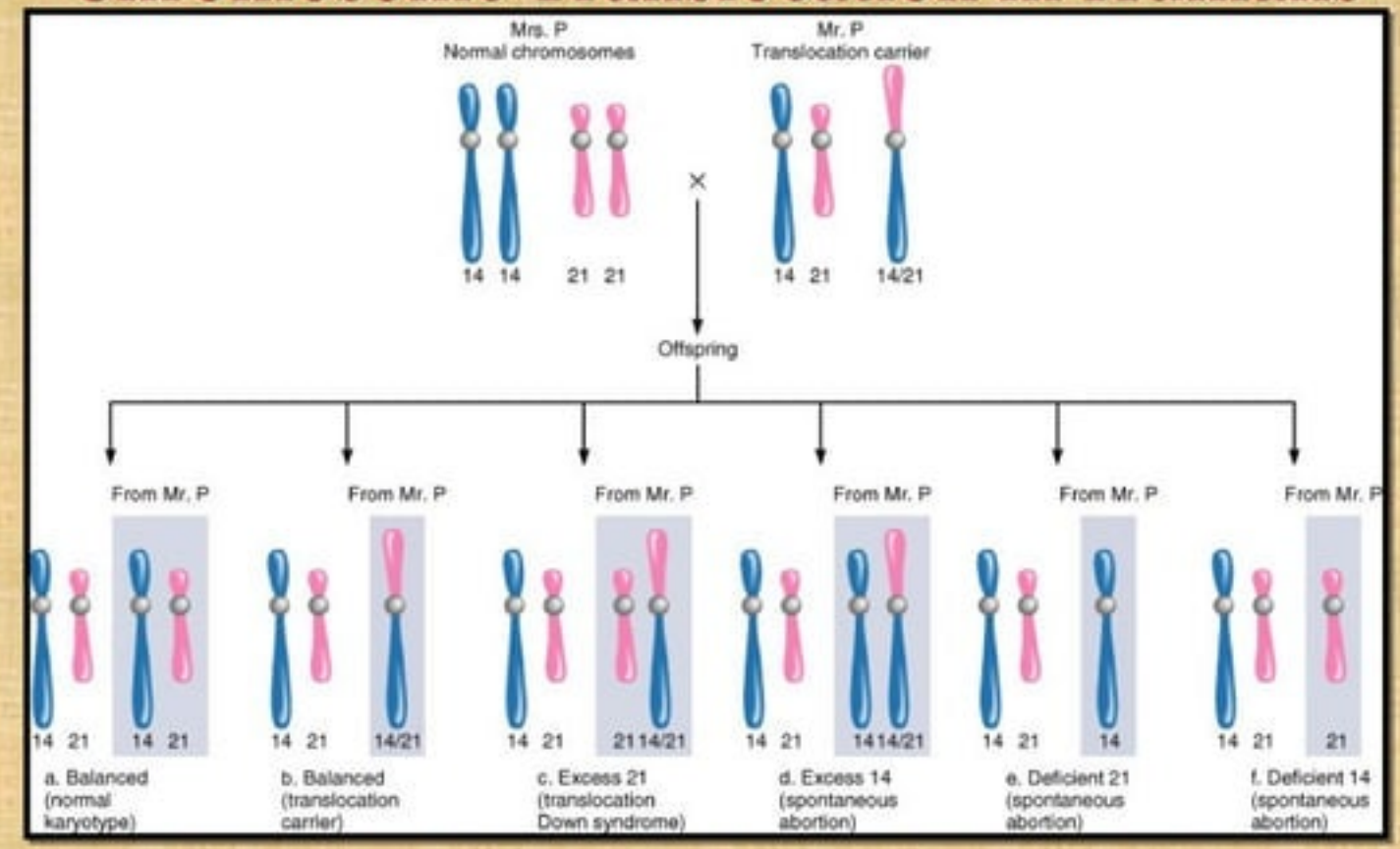


Chromosome Translocation in Humans

- Reciprocal Translocation involves exchange between two non-homologous chromosomes
- Reciprocal translocation betwee chromosomes 2 and 20 causes Alagille Syndrome
- Effects heart, liver, kidneys etc



Chromosome Translocation in Humans

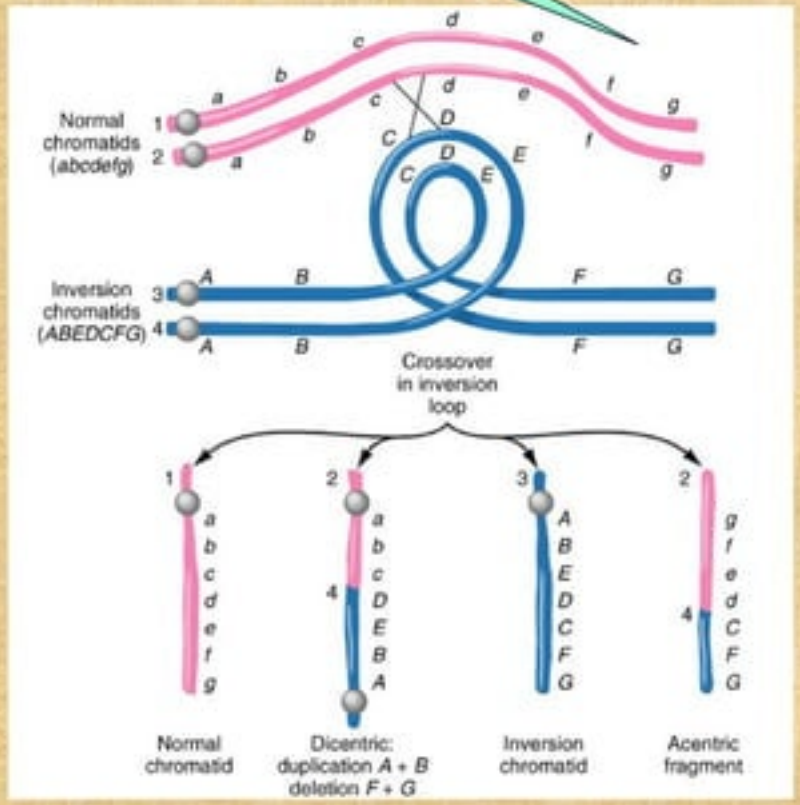


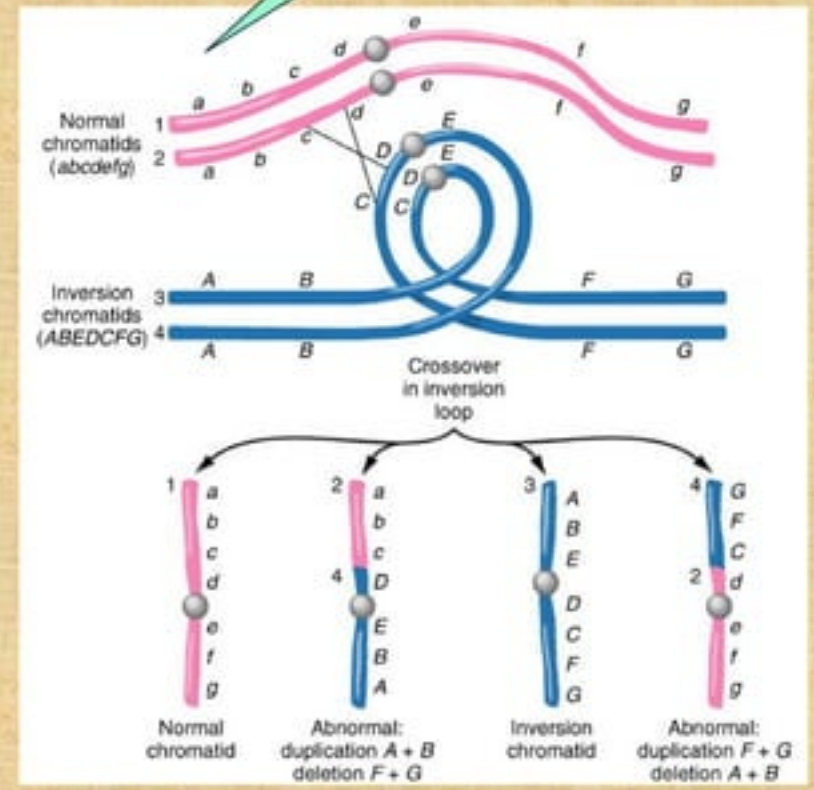
- Robertsonian Translocation involves a fusion of the long arms of two different chromosomes
- Translocation Down Syndrome involves a Robertsonian Translocation between chromosomes 14 and 21

Chromosome Inversions Lead to Unbalanced Meiotic Products

A paracentric inversion does not include the centromere

A pericentric inversion includes the centromere





SIGNIFICANCE OF INVERSION

- ORIGIN OF NEW SPECIES
- PROOF FOR THE OCCURANCE OF CROSSING OVER
- INVERSION IS CONSIDERED AS CROSSING OVER REPRESSORS

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THANK YOU