

# Chromosomal Disorders

# Inheritance Variations

- Are there any other funny things that affect inheritance?
- Are there any ways that meiosis can be “messed up?”

## Errors in Meiosis

- Nondisjunction: failure of homologous chromosomes to separate in Meiosis I or failure of sister chromatids to separate in Meiosis II
- Why can this be problematic?

# Problems?

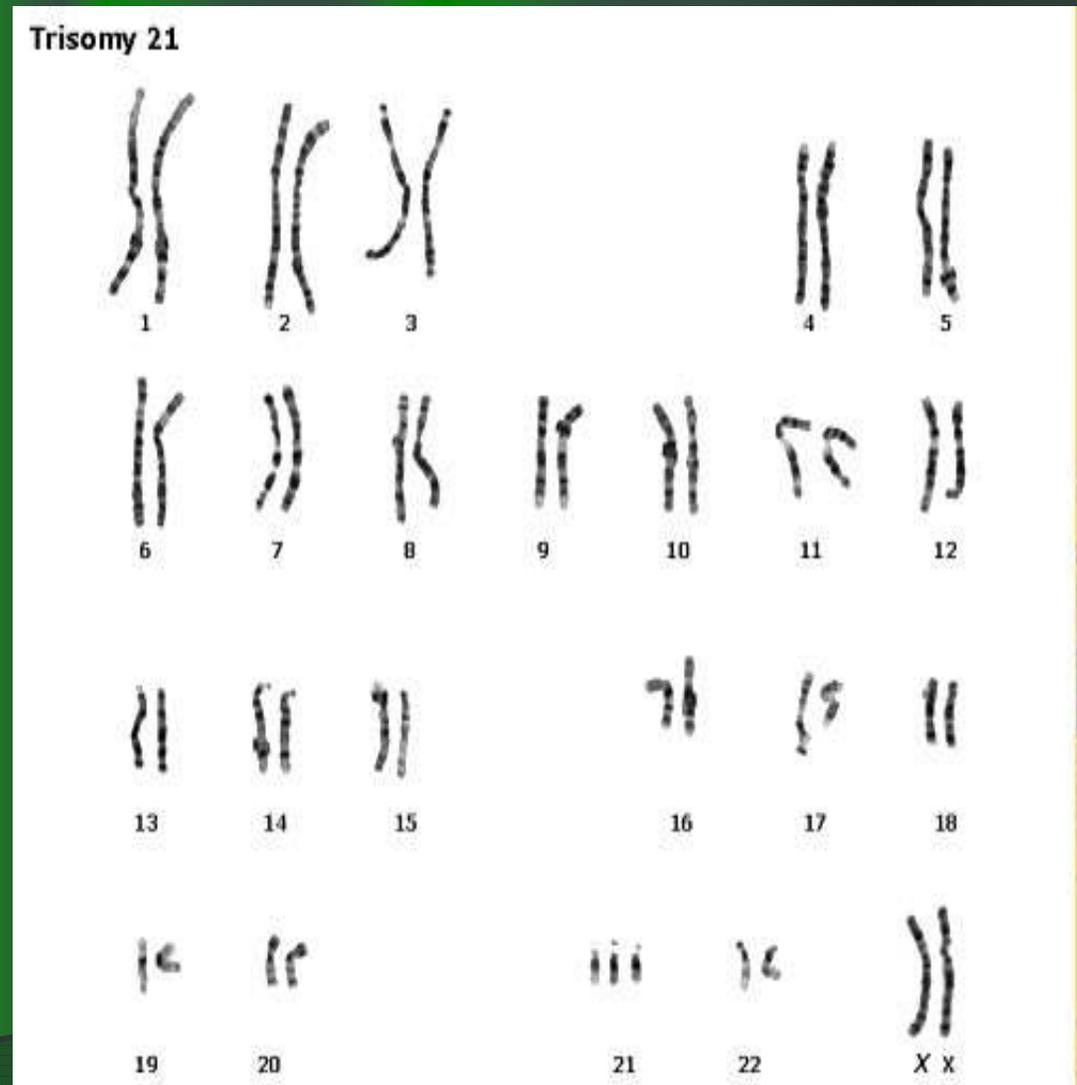
- If you are missing a chromosome, then you are missing important genes
  - Without the information, cells lose the ability to function correctly
  - Development also gets affected

# Issues with chromosome number

- Aneuploidy: Fertilization results in an individual with an abnormal number of chromosomes
- Examples:
  - **Trisomy** three copies of a particular chromosome ( $2n + 1$ )
  - **Monosomy** only one copy of a particular chromosome ( $2n - 1$ )

# Chromosome Disorders

- Most aneuploid zygotes do not survive to birth
- Down syndrome (trisomy 21)
  - 3 copies of chromosome 21
  - Frequency increases with age of the mother



# Common aneuploid chromosome disorders

- Down syndrome
- Klinefelter syndrome
- Trisomy X
- Turner syndrome

# Polyploidy

- Organisms with more than 2 complete sets of chromosomes
- Triploid:  $3n$
- Tetraploid:  $4n$
  
- Common in plants, rarer in animals



# How can polyploidy occur?

- Is this good, bad or neutral?

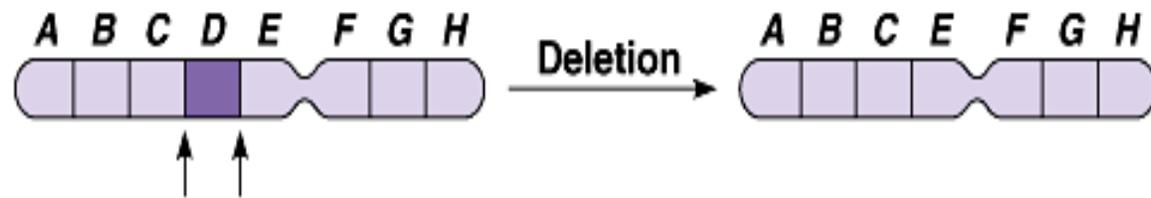
# Disorders can also be caused by changes in chromosome structure

- How can chromosome structure be altered?
- Results from problems with crossing over
- Nonreciprocal crossing over- some chromosomes receiving more than they give

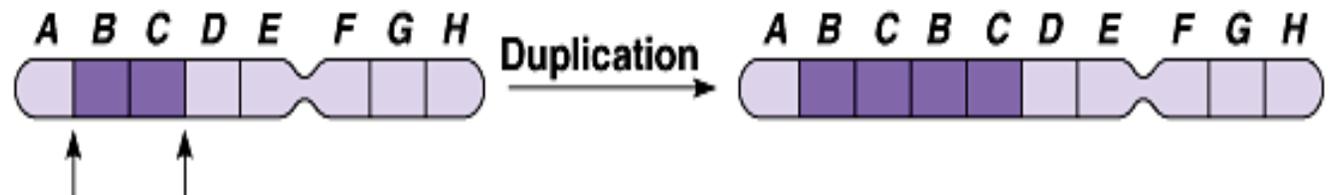
# Alterations in Chromosome Structure

- Deletion: fragment is lost
- Duplication: repeats in a segment
  - (deleted portion attaches)

(a) A **deletion** removes a chromosomal segment.



(b) A **duplication** repeats a segment.



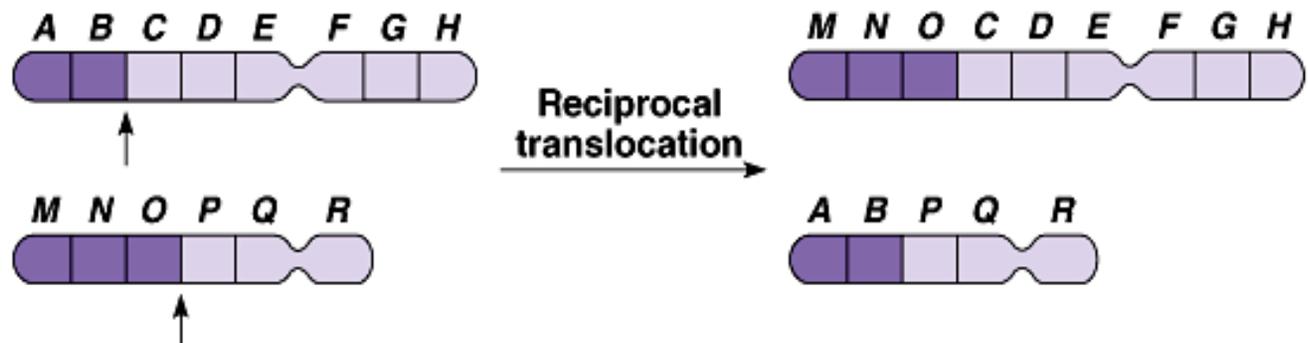
# Alterations in Chromosome structure

- Inversion: fragment binds in reverse order
- Translocation: fragment binds to nonhomologous chromosome

(c) An **inversion** reverses a segment within a chromosome.



(d) A **translocation** moves a segment from one chromosome to another, non-homologous one.



# Why do these matter?

- Missing genes = lost info and function
- Genes placed in the wrong place are not able to be used normally

# Structural Disorders

- Things like deletions and translocations can cause disorders
  - Cri du chat
  - Williams Syndrome
  - Chronic Myelogenous Leukemia
    - Cancer
    - Check out the book on page 288 if you want to learn more
      - What is the Philadelphia chromosome?