















- 1  **Chapter 9**  
Chromosome Variation
- 2  **Chromosomes**
  - ▶ Centromere location
  - ▶ karyotype
- 3  **Chromosome mutations**
  - ▶ Chromosome rearrangements
  - ▶ Aneuploids
  - ▶ polyploids
- 4  **Chromosome mutations**
  - ▶ Chromosome rearrangements
- 5  **Duplications**
  - ▶ AB\*CDEFG
  - ▶ sam the cat ate joe rat
    - ▶ Tandem
      - ▶ AB\*CDEF~~E~~FG
      - ▶ sam the cat cat ate joe rat
    - ▶ Displaced
      - ▶ AB\*CDEFGEF
      - ▶ sam the cat ate joe cat\_rat
    - ▶ Reverse
      - ▶ AB\*CDEFFEG
      - ▶ sam the cat tac ate joe rat
  - ▶ Problem during prophase I
    - ▶ incorrect protein amounts
- 6 
  - ▶ Deletions
  - ▶ AB\*CDG
  - ▶ sam the ate joe rat
    - ▶ Lethal
    - ▶ Pseudodominance
    - ▶ Haplo-insufficient gene
  - ▶ Problem
    - ▶ protein amounts/lack of
- 7 
  - ▶ Inversions
  - ▶ AB\*CDEFG
  - ▶ sam eoj eta tac eht rat
    - ▶ Paracentric
      - ▶ AB\*CFEDG
    - ▶ Pericentric
      - ▶ ADC\*BEFG
  - ▶ Problem = breakage, position, crossover problems - wrong # centromeres
- 8 
  - ▶ Translocations
  - ▶ AB\*CDEFG and MN\*OPQRS
    - ▶ Nonreciprocal
      - ▶ AB\*CDG and MN\*OPEFQRS
    - ▶ Reciprocal

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- ▶ AB\*CDQRG and MN\*OPEFS
- ▶ Robertsonian
- ▶ Acrocentric chromosomes
- ▶
- ▶ Problem = position & regulation, breakage, crossover problems
- ▶ Other effects
- ▶
- ▶
- 9  ▶ Fragile sites
  - ▶ Fragile-X syndrome
    - ▶ 1 in 1250 male births
- 10  **Chromosome number**
  - ▶ Aneuploidy
    - ▶ Nullisomy ( $2n-2$ )
    - ▶ Monosomy ( $2n-1$ )
    - ▶ Trisomy ( $2n+1$ )
    - ▶ Tetrasomy ( $2n+2$ )
  - ▶ Problem = gene dosage
    - ▶ X chromosome exception
  - ▶
  - ▶
- 11  **Downs syndrome**
  - ▶ Autosomal aneuploid
    - ▶ Trisomy 21
    - ▶ Primary
      - ▶ nondisjunction
    - ▶ Familial
      - ▶ translocation
- 12 
  - ▶ Worksheet
  - ▶ Chromosome Rearrangements & Aneuploidy
  - ▶
- 13  **polyploidy**
  - ▶ More than two chromosome sets
    - ▶ Triploids ( $3n$ )
    - ▶ Tetraploids ( $4n$ )
    - ▶ Pentaploids ( $5n$ )
    - ▶ More
  - ▶ Plants
    - ▶ 40% flowering plants
    - ▶ 70-80% grasses
  - ▶
- 14 
  - ▶ Autopolyploidy
    - ▶ Nondisjunction (single species)
  - ▶ Allopolyploidy
    - ▶ Hybrids (different species)
  - ▶ More DNA = larger cell size

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