

# Mendel

- What was Mendel's contribution to our understanding of Heredity?
- What is the Law of Segregation?
- What is the Law of Independent Assortment?
- What is a punnett square and how is it used to illustrate the principles of inheritance?

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# Human Mendelian Traits

- OMIM estimates that about 20,000 genes are inherited in a simple Mendelian way.
- Many blood characteristics and diseases and disorders - some simple physical traits
- Examples: Sickle cell, tented eyebrows, hitchhiker's thumb
- Most human "traits" are more complicated, but particles of inheritance - genes - are passed on in a simple Mendelian way

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# Terminology

genotype  
phenotype  
gene  
allele  
locus

DNA  
chromosomes  
bases

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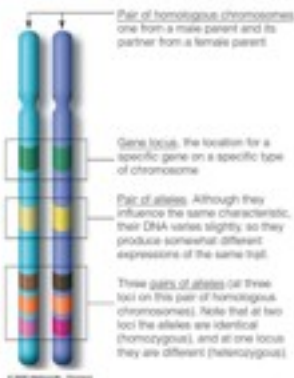
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locus= the location of a gene on a chromosome

Allele= alternative form of a locus

homozygous= having the same allele at the locus on both chromosomes

heterozygous= having different alleles at the locus on both chromosomes



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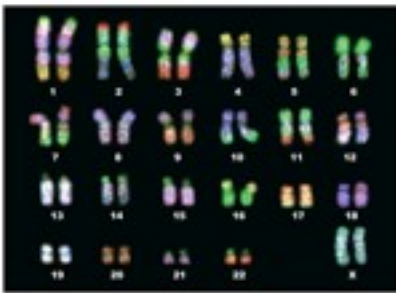
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## Heritability - a 3 Part question

- How come we resemble our parents? That is, **how is our heritable information passed from generation to generation?**
- **How does the genetic code create a characteristic?**
- Where does variation in the code come from?

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## Human Karyotype

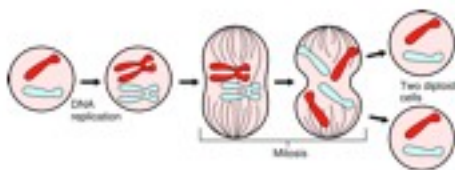


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- MITOSIS - somatic cell division
- MEIOSIS - gametic cell division

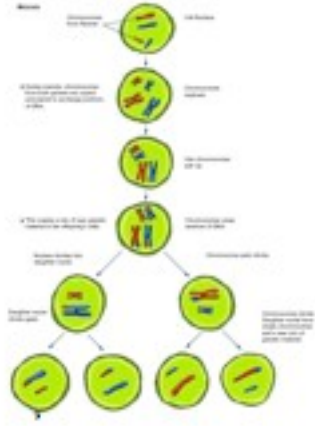
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## Mitosis



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# Meiosis



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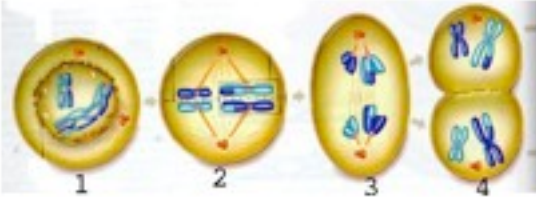
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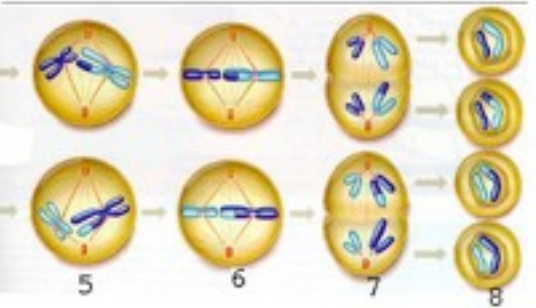
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## Variation comes from

- Recombination
- Crossing Over
- Mutation

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## Recombination

2 chromosomes  
x  
2 possibilities for each  
=  
4 possible combinations

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## Recombination

23 chromosomes  
x  
2 possibilities for each  
=  
2 to the 23rd power  
=

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## Recombination

23 chromosomes  
x  
2 possibilities for each  
=  
2 to the 23rd power  
=  
8,388,608

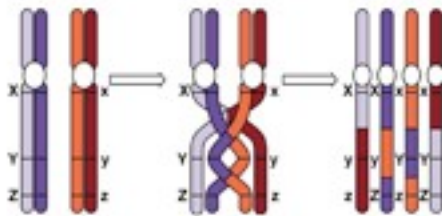
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## Variation comes from

- Recombination
- Crossing Over
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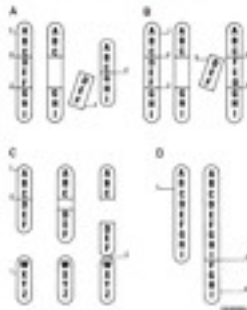
## Crossing over during meiosis



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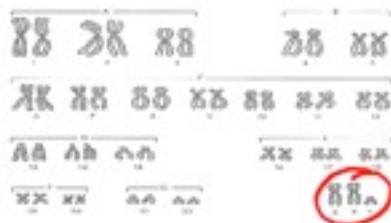
## Chromosomal mutations

- Down's syndrome -21
- Klinefelter's syndrome -Sex
- Turner's syndrome - sex
- William's Syndrome - 7



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## Klinefelter's Karyotype



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## Down's Karyotype



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## Trisomy 13

Karyotype from a female with Patau syndrome (47,XX,+13)



- small head
- small eyes
- cleft lip
- ear shape
- palm differences
- extra fingers/toes
- heart defects
- kidney defects
- etc.

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## Variation comes from

- Recombination
- Crossing Over
- Mutation

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## Mutation

- Change in base sequence of DNA
- Occurs during replication stage of meiosis (or mitosis)
- MAY change the amino acid change and therefore the protein

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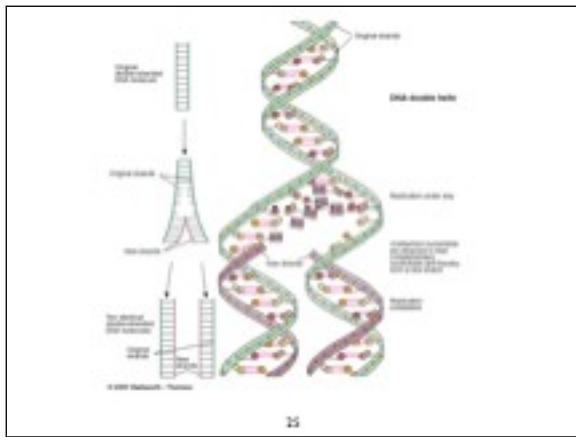
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## Kinds of Mutations

- **Substitution** - replace one base with another
- **Frame Shift** -
  - **Insertion**- an extra base gets pulled in
  - **Deletion**- a base gets omitted

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## How common is mutation?

- happens all the time
- assume a rate of one in a million per locus per gamete
- assume approximately 50,000 loci
- $(1 \times 10^{-6}) \times (5 \times 10^4) = 0.05$   
5% of gametes have a mutation
- an individual is combination of two gametes  
 $2 \times 0.05 = 0.1$  - 10%

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