Basic Genetics (SQBS 2753)

# Introduction to Genetics

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# Genetics

- Classical Genetics
  - Mendelian genetics
    - Fundamental principles underlying transmission of genetic traits
  - Forward genetics
    - Identify or generate phenotypically mutant organisms
    - Determine mode of inheritance and interactions with other mutants
    - Identify mutated gene(s)
    - Correlate observed phenotype with biochemical mechanisms

# Genetics

- Molecular Genetics
  - Based on classical approaches
    - Refers to techniques applied to study genetic questions
    - Most modern genetic studies are molecular
  - Reverse genetics
    - Genes identified 1st
    - Gene mutated by molecular biology techniques
    - Mutant gene inserted into genome of organism and resulting phenotype studied

# History of Genetics

- Domestication of plants & animals suggests prehistoric peoples recognized concept of heredity
- Traits passed from parent to offspring
- Selection of desirable traits – artificial selection

# History of genetics

- Ancient Greeks
  - Mostly bunk
- Renaissance 1600-1800
  - Theories of epigenesis vs preformation
- 1800s – turn to a more reductionist approach
  - Dalton's Atomic theory
  - Schwann's cell theory

# History of Genetics

- Mid 19th century (1850)
  - Darwin & Wallace
    - Theories of evolution
  - Lamarck
    - Theories on acquisition of heritable traits
  - Mendel
    - Theories on transmission of traits

# Gregor Mendel (1822-1884)

- Systematically recorded results of crosses
- Theorized on nature of hereditary material
- Postulate mechanism of transfer of "Elementen" governing traits

# History of Genetics

- Pioneering work of Mendel was done in ignorance of cell division – particularly meiosis, and the nature of genetic material – DNA

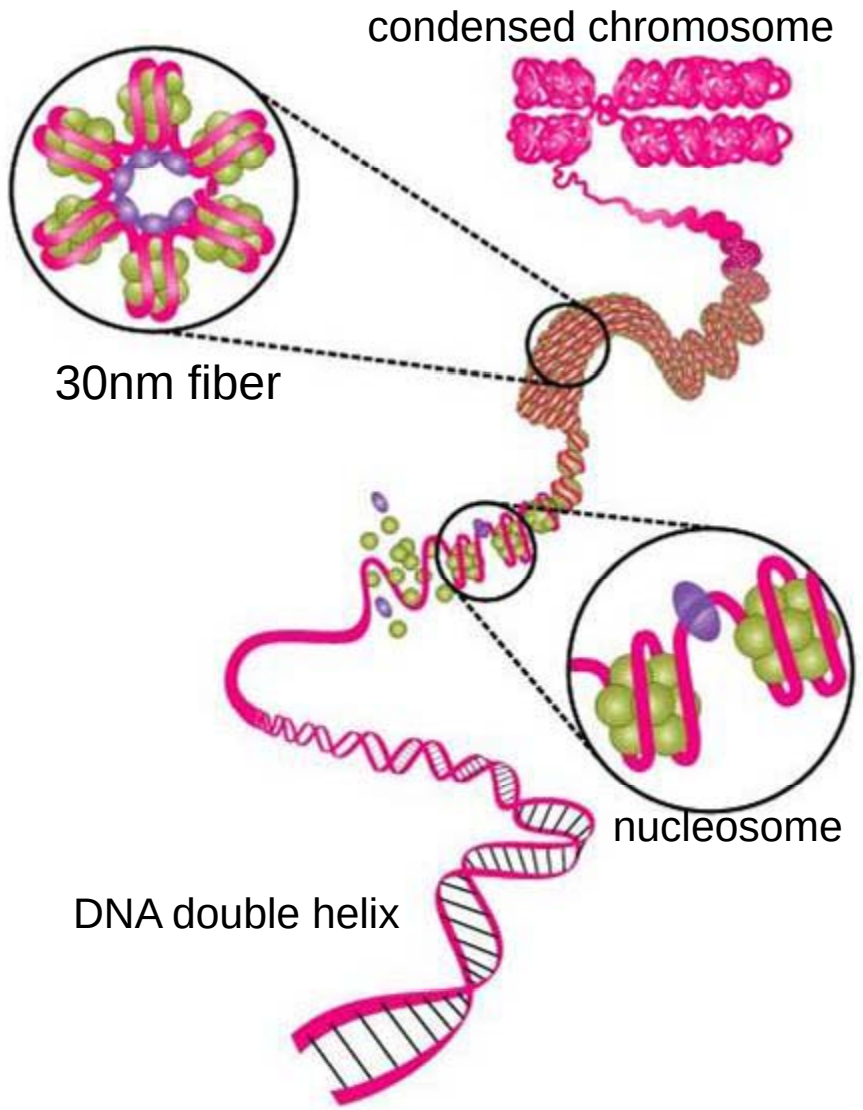
- 1869 - Friedrich Miescher identified DNA
- 1900-1913
  - Chromosomal theory of inheritance – Sutton & Boveri
  - Genes on chromosomes – TH Morgan
  - Genes linearly arranged on chromosomes & mapped – AH Sturtevant
- 1941 – George Beadle & Ed Tatum related "gene" to enzyme and biochemical processes
- 1944 – Oswald Avery demonstrated that DNA was genetic material

# DNA

- 1953 - James Watson, Francis Crick, Rosalind Franklin & Maurice Wilkins
- Lead to understanding of mutation and relationship between DNA and proteins at a molecular level
- 1959 – "Central Dogma"
  - DNA→RNA→protein

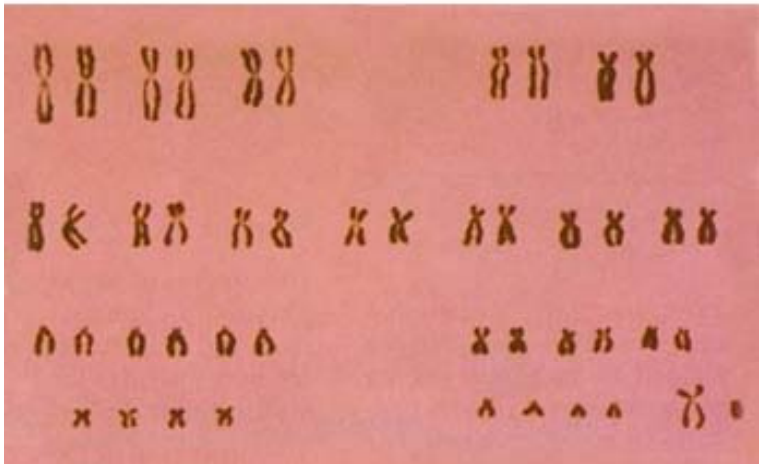
# Genetic Concepts

- Chromosome –
  - double stranded DNA molecule packaged by histone & scaffold proteins



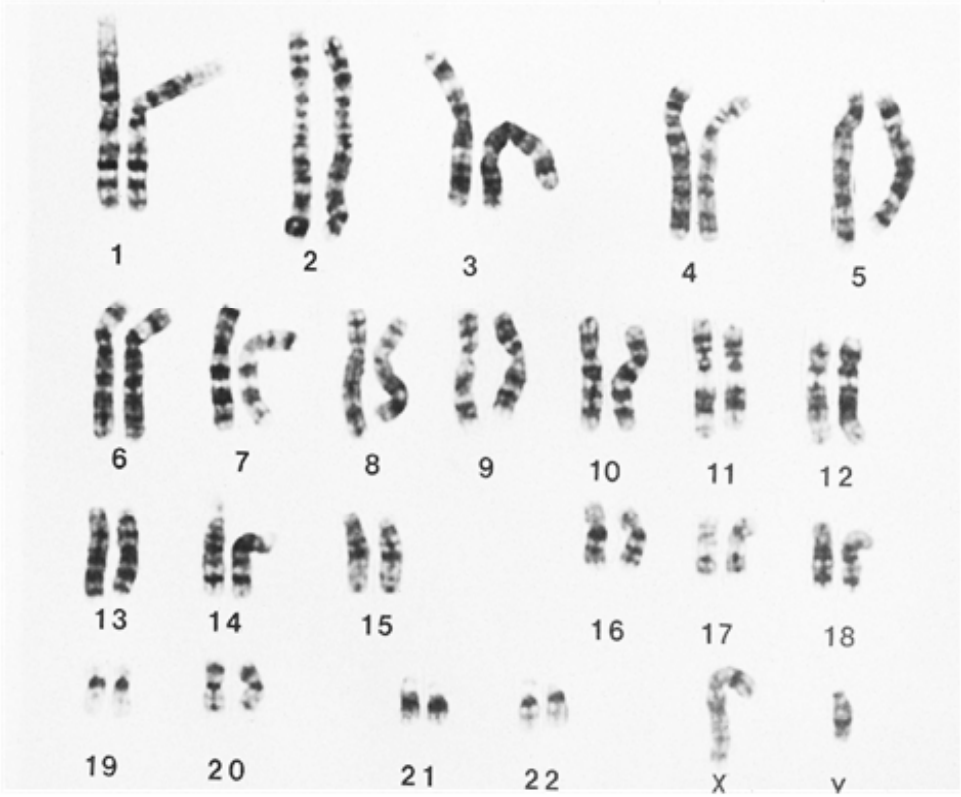
# Genetic Concepts

- Chromosome numbers
  - Constant for an organism
  - n - haploid number
  - 2n – diploid number
- Karyotype



# Genetic Concepts

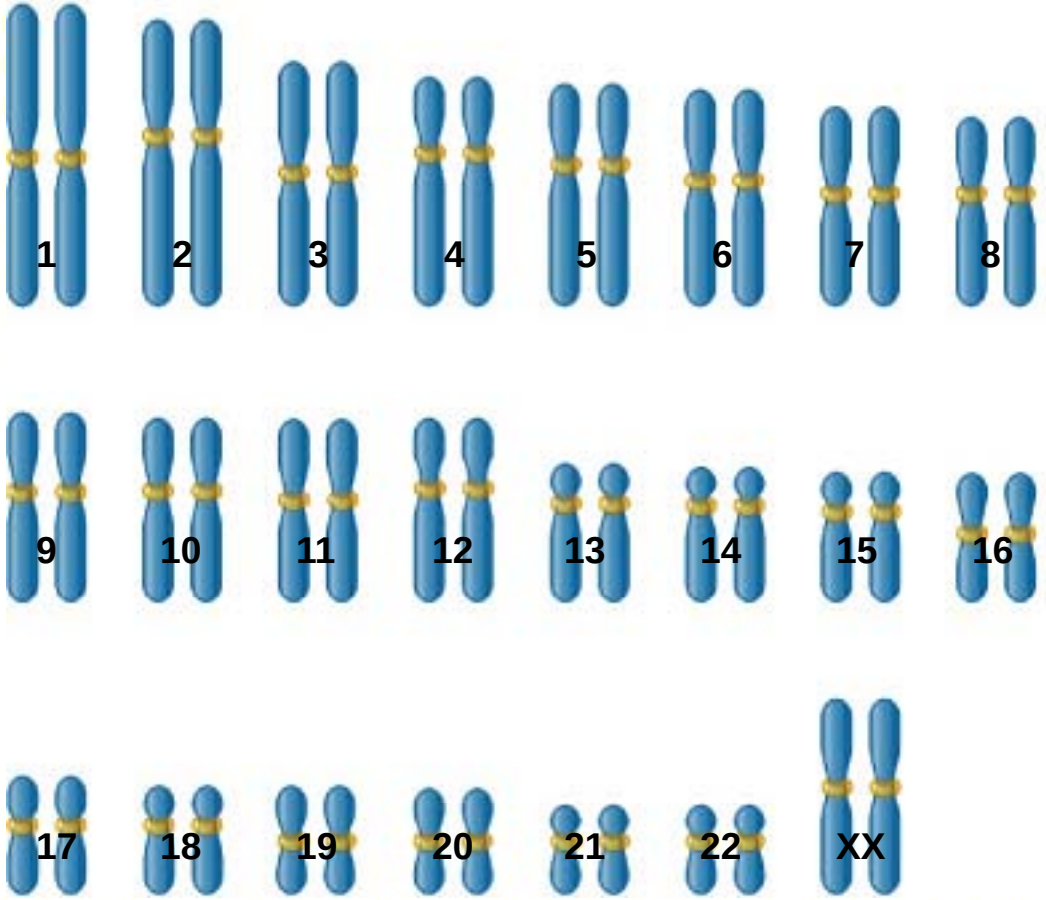
- Male karyotype



# Genetic Concepts

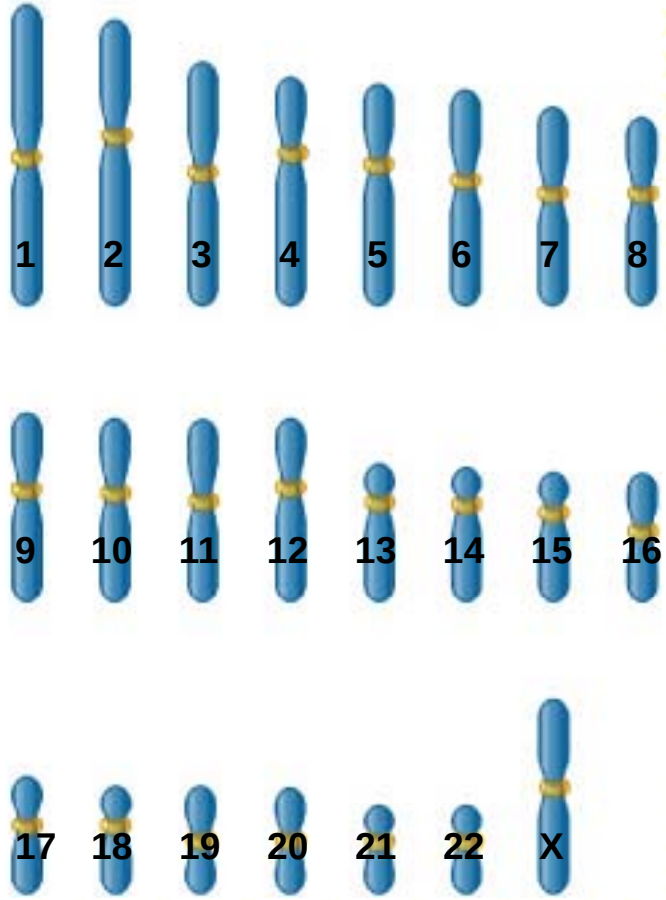
- Chromosome numbers
  - Each individual inherits *n* # of chromosomes from dad & *n* # from mom
  - Humans - 46 chromosomes = 2*n*
  - Humans 23 paternal, 23 maternal
  - Humans *n* = ____
  - Each maternal & paternal pair represent homologous chromosomes - called homologs

# Genetic Concepts




**(a) Chromosomal composition found in most female human cells (46 chromosomes)**

**Diploid**

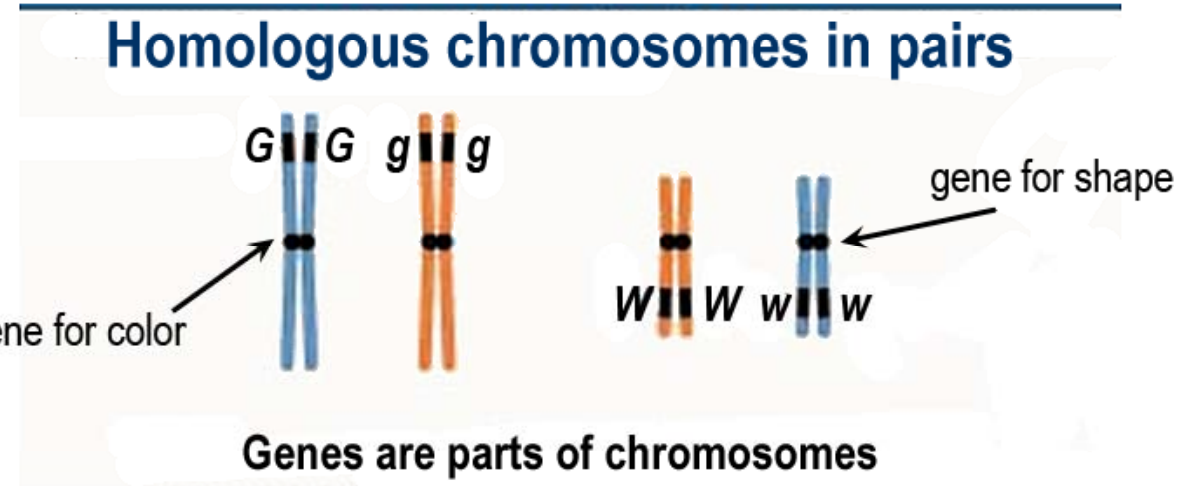



**(b) Chromosomal composition found in a human gamete (23 chromosomes)**

**Haploid**

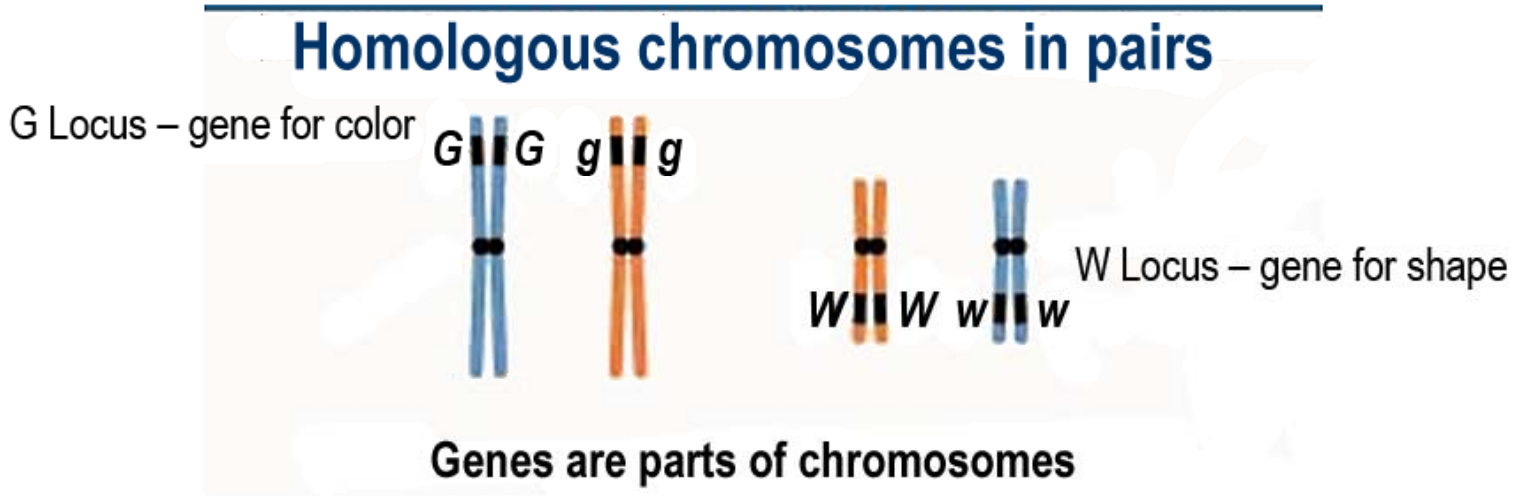
# Genetic Concepts

- Homologous Chromosomes
  - Share centromere position
  - Share overall size
  - Contain identical gene sets at matching positions (loci)



# Genetic Concepts

- Gene – sequence of DNA which is transcribed into RNA
  - rRNA, tRNA or mRNA
- Locus – the position on a chromosome of a particular DNA sequence (gene)

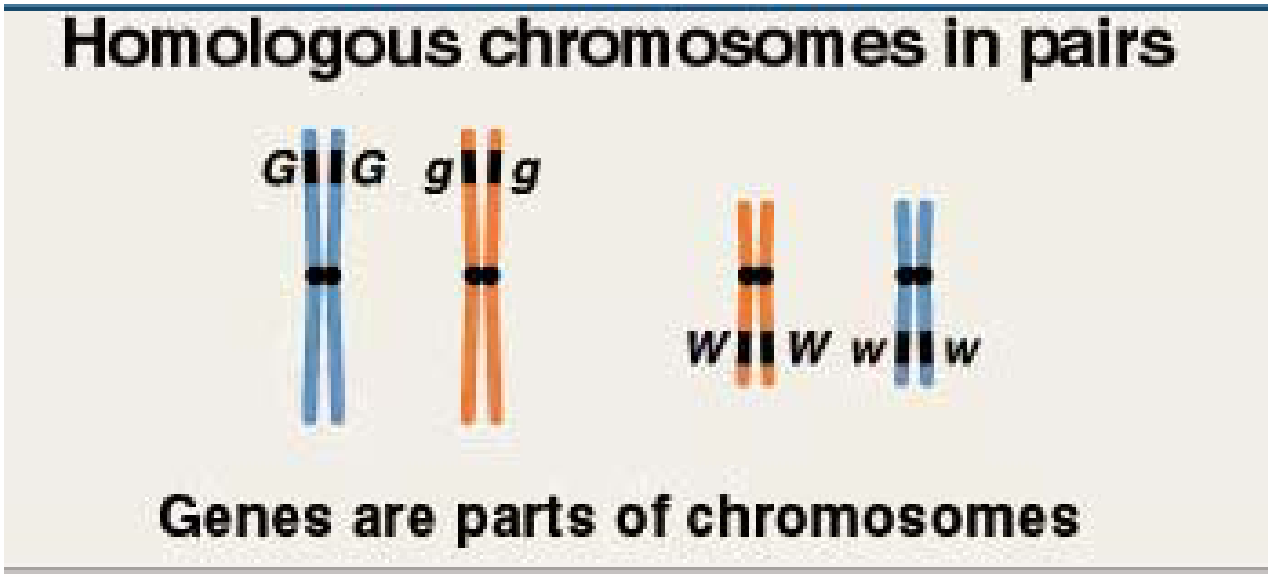


# Genetic Concepts

- DNA is mutable
- A variation in DNA sequence at a locus is called an allele
  - Diploid organisms contain 2 alleles of each locus (gene)
    - Alleles can be identical – homozygous
    - Alleles can be different – heterozygous
    - If only one allele is present – hemizygous
      - Case in males for genes on X and Y chromosomes

# Genetic Concepts

Allele – G vs g; W vs w

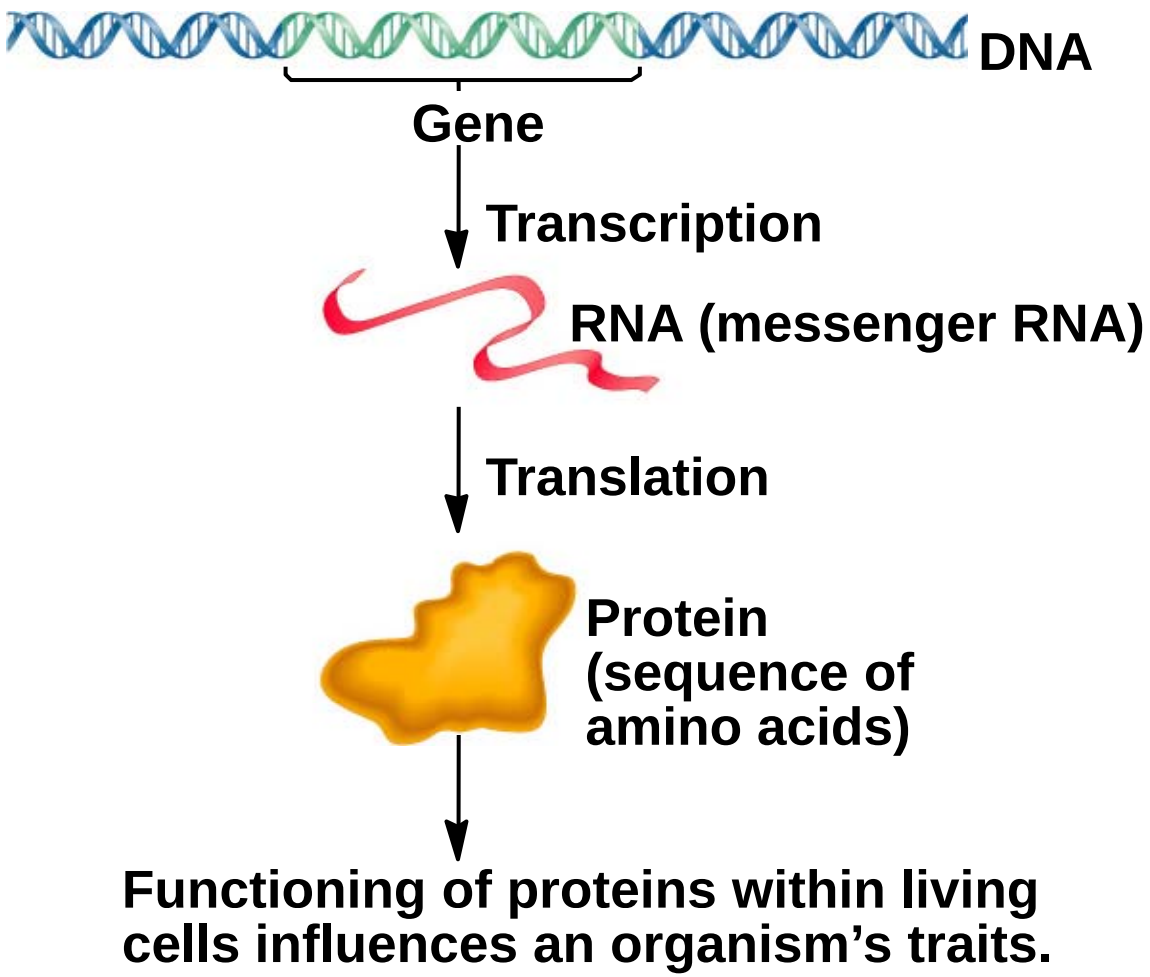



At the G locus either the G or g allele may be present on a given homologue of a homologous pair of chromosomes

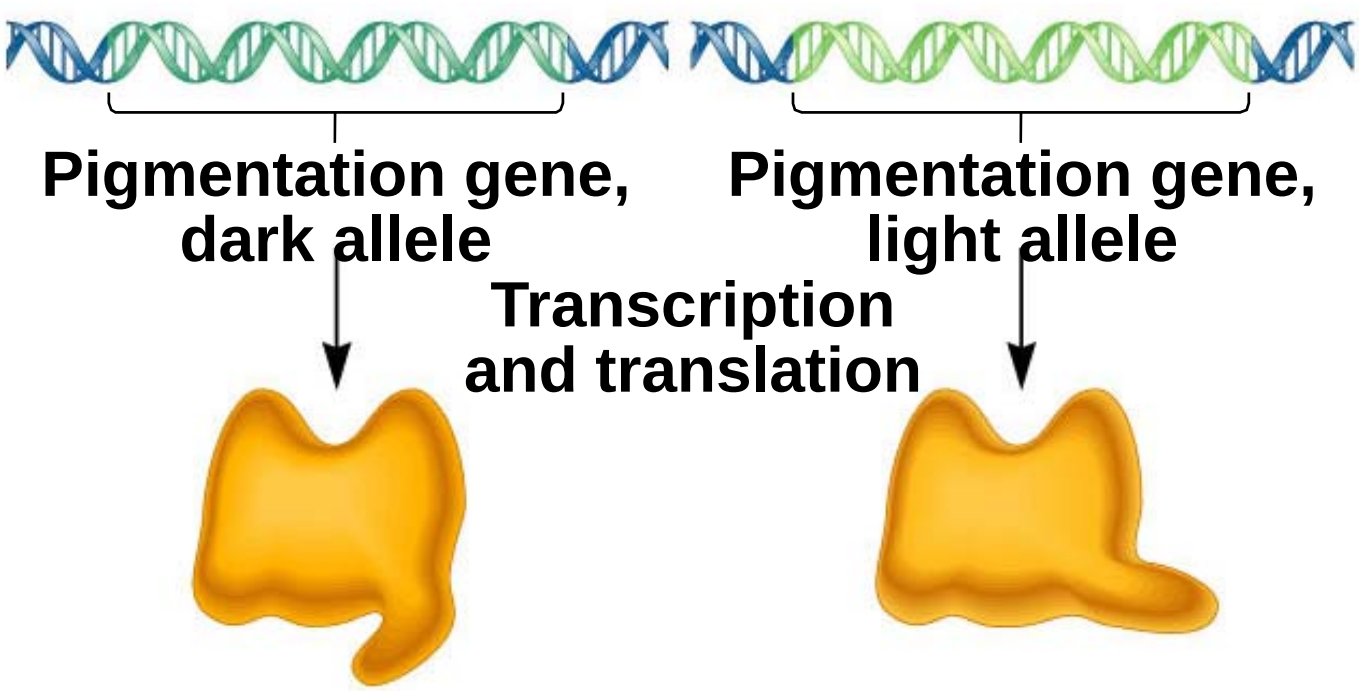
# Genetic Concepts

- Genome
  - Collection of all genetic material of organism
- Genotype
  - Set of alleles present in the genome of an organism
- Phenotype
  - Result of Gene Expression
  - Genes (DNA) are transcribed into RNA
  - mRNA is translated into protein, tRNA & rRNA work in translation process
  - Biochemical properties of proteins, tRNAs & rRNAs determine physical characteristics of organism

# Gene Expression



# Mutation & Phenotypic Variation



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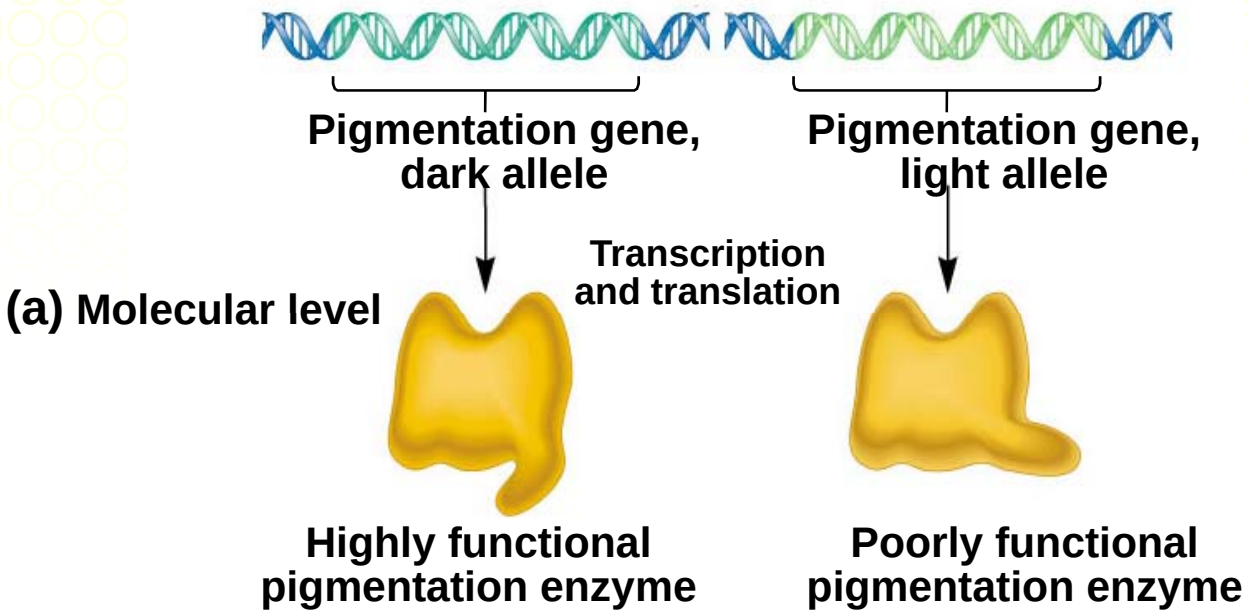

# Mutation & Phenotypic Variation

**Dark butterfly**

**Light butterfly**

**(c) Organismal level**

**Dark butterflies are usually in forested regions.**

**Light butterflies are usually in unforested regions.**

**(d) Populational level**

# References


- Snustad DP, Simmons, MJ (2010) Principles of Genetics Fifth Ed. John Wiley & Sons, Inc., USA.
- Klug WS, Cummings MR, Spencer CA, Palladino MA (2012) Concepts of Genetics. 10th Ed. Pearson, California.
- Hartwell LH, Hood L, Goldberg ML,Reynolds AE, Silver LM (2011) Genetics: From Genes to Genomes. 4th Ed. McGraw-Hill Companies, Inc.,NY