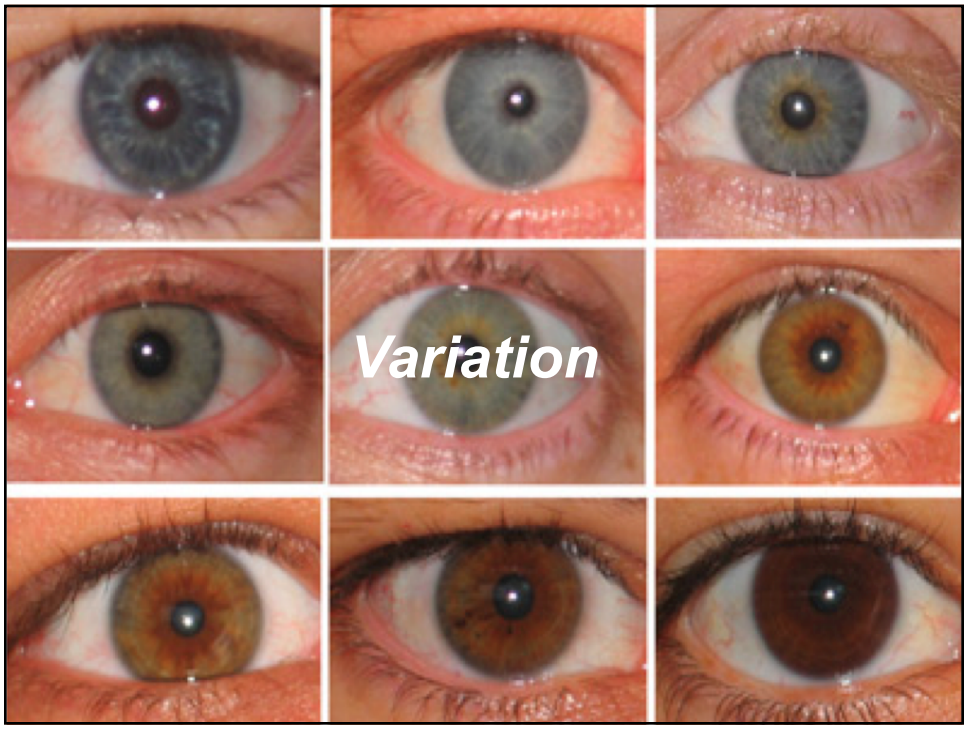

Lecture 4

NeoDarwinism & the evolutionary significance of genetic variation

1. **Where does genetic variation come from?**
2. **How is it inherited?**
3. **How does it influence trait variation?**

Key reading in Coyne – Chapter 3

1

Requirements for Darwin's theory - revisited

GENETICS

- **Variation –** variation among individuals in a population
- **Heredity –** progeny resemble their parents more than unrelated individuals

- **Selection –** some forms better at surviving and breeding in a given environment

But Darwin had no understanding of genetics and the mechanism of inheritance

2

Variation – variation among individuals in a population

Phenotype

The organism as observed – used when discussing a trait or a feature of an organism that varies

Environmental effects

Genetic variation

3

What is a gene?

Complex concept – various definitions

- The functional unit of inheritance
- A unit of hereditary information located on the chromosomes consisting of DNA
- A DNA sequence composed of codons* essential for a specific biological function

*A sequence of 3 nucleotides that makes up the genetic code

4

Some basic terms used in genetics

Genotype

- Genetic constitution of an organism –used in relation to a particular gene or gene combinations e.g. *Aa*, *AaBB*

Allele

- Versions of the same gene that differ in their base sequence

Genome

- The entire organism's DNA including both genes and non-coding regions

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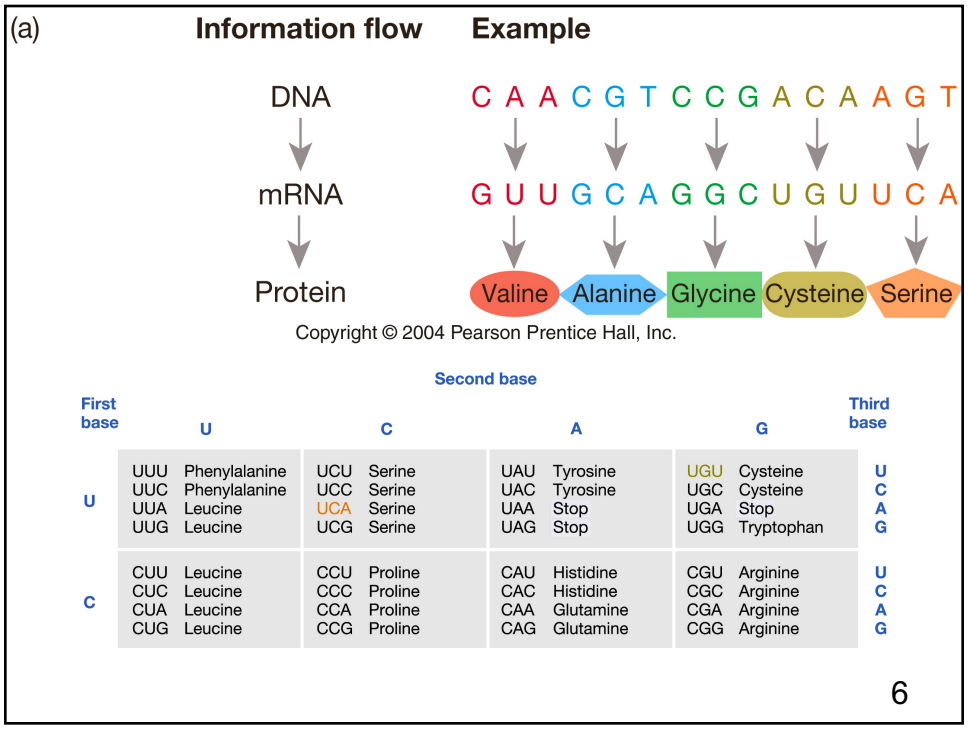

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Evolution requires genetic variation – where does it come from?

- Gene flow
- Hybridization
- Recombination – creation of new combinations of alleles during sexual reproduction
- Independent Assortment - the alleles of two (or more) different genes get sorted into gametes independently of one another
- Mutation – stable change in the base sequence of DNA

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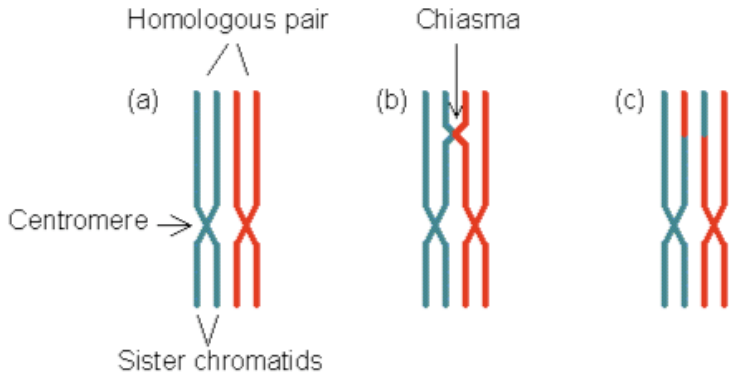
Recombination



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Independent assortment & recombination

- Independent assortment & recombination during meiosis generates enormous diversity
- Most genetic variability in a population results from sexual reproduction; in any given generation input from mutation very small

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Mutation – ultimate source of genetic variation

- Stable change in DNA sequence resulting in a change of genotype
- Occurs at a very low but variable rate in all organisms
- To be important for evolution, must occur in germ cells (sperm/eggs) – somatic mutations NOT inherited

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Mutation and the structure of DNA

	DNA	mRNA	Amino acid	
1. Point mutations	ATG	→ UAC	→ Tyrosine	
ATG → ATA	ATA	→ UAU	→ Tyrosine	silent
	ACG	→ UGC	→ Cysteine	replacement

1. Insertions/deletions (including 'jumping genes')

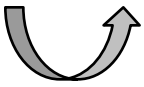
ATG → ATGG

2. Changes in repeat number

ATGATG → ATGATGATG

3. Chromosomal rearrangements

ATG → TGA



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Sickle Cell Anemia

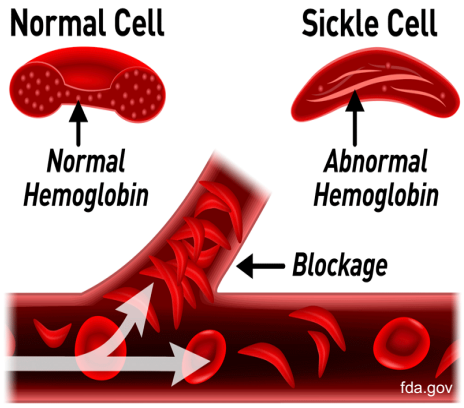
No Sickle Cell Anemia

DNA RNA Protein

CTC → GAG → Glutamic acid

Sickle Cell Anemia

CAC → GUG → Valine



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A mutant in the field – flower colour in an Australian daisy – what will happen to it?

Effects of mutations

- The effect of a mutation on fitness:
  - neutral
  - deleterious
  - lethal
  - beneficial
- In many cases the fitness effect depends on environment

14

Mutation rates in eukaryotes

Organism	Number of fitness-affecting mutations per diploid genome each generation
Drosophila (fruit fly)	1.2
Caenorhabditis (worm)	0.96
Arabidopsis (plant)	0.1 - 0.6
Mouse/Rat	0.91
Human/Chimpanzee	1.6 - 3

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Characteristics of mutation

- Mutation is an unstoppable phenomenon
  - despite cellular mechanisms to correct errors during DNA replication
- Mutation is not directed by the organism or the environment
  - random with respect to effects on fitness
- Rates depends on the type of mutation
  - also varies among genes
- Environmental insults can affect mutation rate
  - mutagens, high temperature

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Inheritance and the transmission of genes among generations

- How is genetic information transmitted from parents to offspring – mechanism of inheritance?
- How are traits expressed in parents and offspring – is this influenced by how many genes control a trait?

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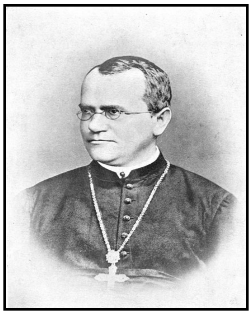

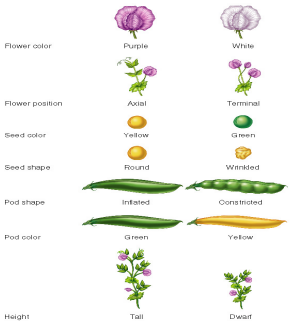

Gregor Mendel

(1822-1884)

- Priest and "father of modern genetics"
- Through controlled crosses with peas established the laws of inheritance
- Mendel's Laws re-discovered at the beginning of the 20th century by Hugo de Vries & Carl Correns

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Design of Mendel's Experiment

P  Cross between two pure-breeding lines

⇩

F_1 selfed

⇩

F_2

Parental plants differ in some observable phenotypic trait e.g. flower colour, plant height, seed shape

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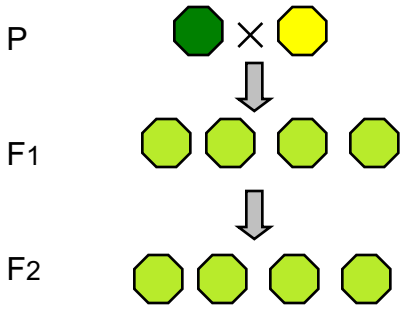
Blending inheritance predicts



Blending inheritance occurs when offspring of a cross show intermediate phenotype

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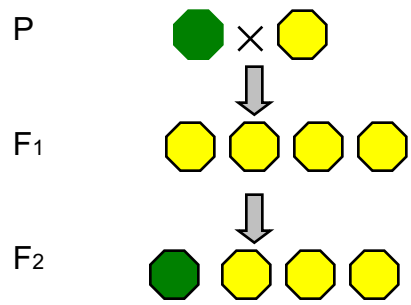
Mendel's results



cross
of pure-breeding lines

selfed

3/4 yellow
1/4 green

Yellow is **dominant** to green which is **recessive**; predictable **3:1 phenotypic ratio** and **1:2:1 genotypic ratio** in F_2

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Main conclusions from Mendel's experiments with peas

- Inheritance determined by discrete particles (genes) – "particulate inheritance"
- Most organisms carry 2 copies of each gene (alleles) and are diploid
- Organisms produce gametes (e.g. sperm vs. eggs) each containing one allele - gametes haploid
- Offspring inherit one allele from each parent at random

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*Genetic Polymorphism**

- **The occurrence of two or more discrete forms of a species in the same locality in such proportions that the rarest cannot be maintained by mutation alone – usually means frequency >5%**
- Involves discrete phenotypes (called forms or morphs) governed by segregation of a small number of alleles at 1-2 major genes

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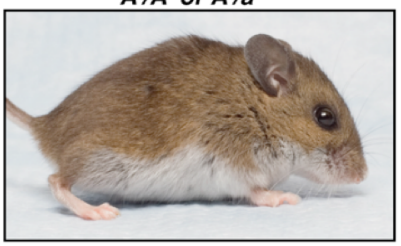
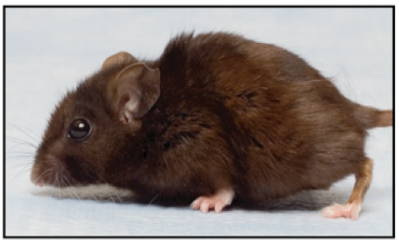
Mouse coat colour polymorphism in contrasting habitats

Hopi Hoekstra - Harvard

A A^+/A^+ or A^+/a^-



B a^-/a^-



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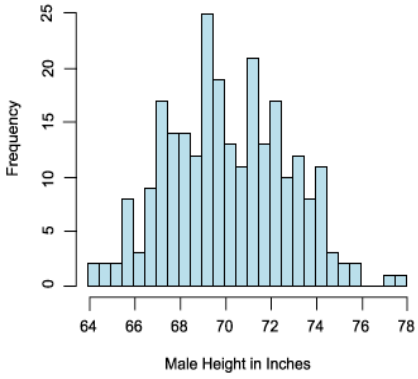
Variation in human height



65% of variation in human height is heritable – an example of quantitative inheritance

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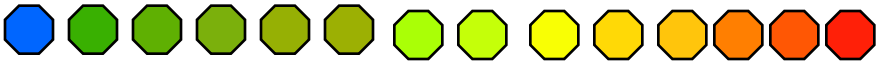
Discrete vs continuous traits

Discrete traits – Simply inherited by 1 or 2 genes (major genes) – Mendelian genetics

Continuous traits – complex inheritance by many genes (polygenes) of small effect – Quantitative inheritance



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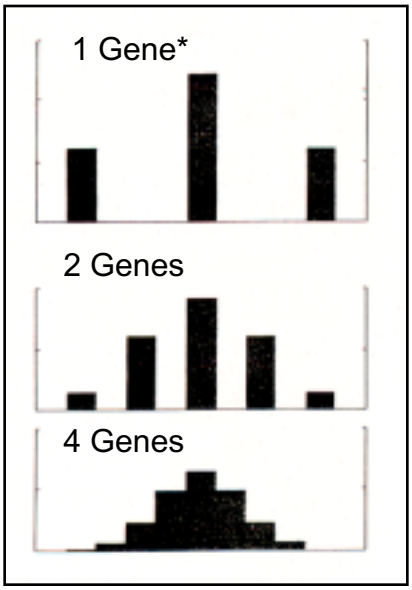
Gene number & phenotypic distribution



Trait distribution

Relation between number of genes controlling a trait and phenotypic variability

Few genes – discontinuous (discrete) variation

Many genes - continuous variation

* Gene action here is co-dominant; heterozygote has intermediate phenotype

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Variation – variation among individuals in a population

Phenotype

The organism as observed – used when discussing a trait or a feature of an organism that varies

Environmental effects Genetic variation

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Genetic analysis of variation

Discontinuous variation - Mendelian genetics

- Genes of major effect, dominance and recessiveness, genetic polymorphism

Continuous variation - Quantitative genetics

- Many genes of minor effect, important environmental effects, selection response

29

Fisher's Fundamental Theorem of Natural Selection

Sir Ronald Fisher
1890-1962

"The rate of increase in fitness of a population at any time is equal to its genetic variance in fitness at that time"

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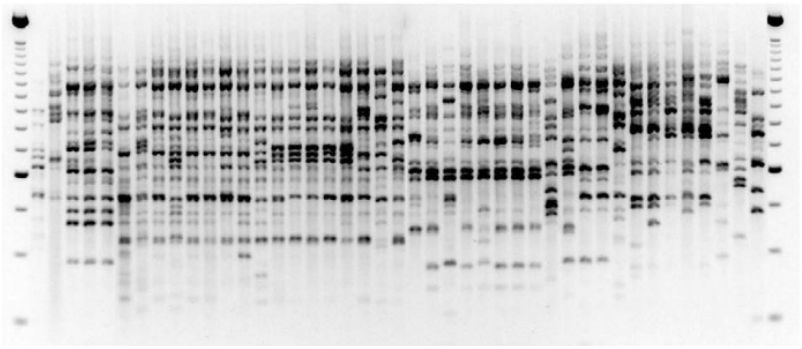
Next Lecture - 7

The maintenance & measurement of genetic variation



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