

Genetics

What is genetics?

- Genetics is the study of genes, and tries to explain what they are and how they work
- Genes are how living organisms inherit features from their ancestors
- Genetics tries to identify which features are inherited, and explain how these features are passed from generation to generation

- Genes are made from a long molecule called DNA, which is copied and inherited across generations
- DNA is made of simple units that line up in a particular order within this large molecule
- The order of these units carries genetic information, similar to how the order of letters on a page carries information

- The language used by DNA is called the genetic code, which lets organisms read the information in the genes
- This information is the instructions for constructing and operating a living organism.

Genotype & Phenotype

- The genotype is the genetic constitution of an organism
- The phenotype of an organism is, roughly, the characters (or traits) of an organism
 - It does not mean just 'what you can see on the surface'
 - Example: blood groups are definitely a part of the phenotype but they are not visible just by looking at a person

Dominance

- An allele is a form of a gene at a particular position on a chromosome
- In a heterozygote the effect of one allele may completely 'mask' the other
- The allele that masks the other is said to be **dominant**
- The alternative allele is said to be **recessive**

- Dominant characteristic
 - Dimples = D
- Recessive characteristic
 - No dimples = d
- These can combine as follows:
 - DD = dimples
 - Dd = dimples
 - dD = dimples
 - dd = no dimples





Gregor Mendel

- Mendel demonstrated that the inheritance of certain traits in pea plants follows particular patterns

Inheritance

- Punnett squares can be used to predict the results of single trait crosses
 - Shows all of the possible combinations of the genes
 - Shows the probability of each combination

- Crossing between two pea plants for purple (B, dominant) and white (b, recessive) blossoms

		pollen ♂	
		B	b
pistil ♀	B	 BB	 Bb
	b	 Bb	 bb

- Homozygous
 - Both alleles are the same
 - BB, bb
- Heterozygous
 - Both alleles are different
 - Bb, bB

Gender in Humans

- Gender is determined by one pair of chromosomes
- There are two types of sex chromosome
 - X, Y
- Male = XY
- Female = XX

- Males can contribute either an X or Y chromosome
- Females can only contribute an X chromosome
- Males, therefore, determine the sex of the offspring

		Father	
		X	Y
Mother	X	XX	XY
	x	Xx	Xy

Sex-linked Traits

- Sex chromosomes carry genes for many other characteristics besides gender
- Such characteristics are said to be sex-linked
- Most sex-linked traits are recessive and are carried on the X chromosome
- Some sex-linked traits
 - Hemophilia, Red-Green Color Blindness, Congenital Night-Blindness, Muscular Dystrophy

Example - Hemophilia

- Hemophilia is the inability for blood to clot
- Recessive gene on the X chromosome
- X^H = normal clotting (Dominant)
- X^h = Hemophilia (Recessive)

- Female genotypes/phenotypes
 - $X^H X^H$ = normal
 - $X^H X^h$ or $X^h X^H$ = carrier
 - $X^h X^h$ = hemophilia
- Male genotypes/phenotypes
 - $X^H Y$ = normal
 - $X^h Y$ = hemophilia

Inheritance Probabilities

- Father is normal and Mother has hemophilia

		Father	
		X^H	Y
Mother	X^h	X^hX^H	X^hY
	X^h	X^hX^H	X^hY

- Male offspring have hemophilia
- Female offspring are carriers

- Father is normal and Mother is a carrier

		Father	
		X^H	Y
Mother	X^H	X^HX^H	X^HY
	X^h	X^hX^H	X^hY

- Female offspring
 - 50% chance normal (X^HX^H)
 - 50% chance carrier (X^hX^H)
- Male offspring
 - 50% chance normal (X^HY)
 - 50% chance hemophilia (X^hY)

- Father has hemophilia and mother is normal

		Father	
		X^h	Y
Mother	X^H	X^HX^h	X^HY
	X^H	X^HX^h	X^HY

- Male offspring are normal
- Female offspring are carriers

- Father has hemophilia and mother is a carrier

		Father	
		X^h	Y
Mother	X^h	X^hX^h	X^hY
	X^H	X^HX^h	X^HY

- Female offspring
 - 50% chance hemophilia (X^hX^h)
 - 50% chance carrier (X^HX^h)
- Male offspring
 - 50% chance normal (X^HY)
 - 50% chance hemophilia (X^hY)

- Both parents have hemophilia

		Father	
		X^h	Y
Mother	X^h	X^hX^h	X^hY
	x^h	X^hx^h	X^hY

- All offspring have hemophilia