EDEXCEL GCSE BIOLOGY GENETICS Part 2

The genome is the entire DNA of an organism.

All genetic variation arises in mutation, most have no effect on phenotype, some influence but very rarely a single mutation determines phenotype.

Variation

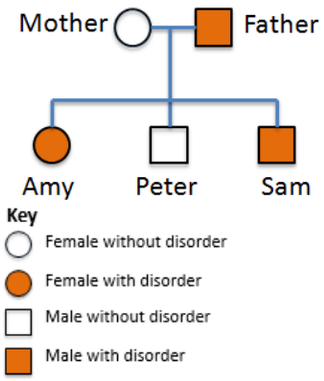
Variation: difference in the characteristics of individuals in a population may be due to		
	Genetic causes (inheritance)	There is usually extensive genetic variation within the population of a species e.g. hair colour, skin colour, height that can also be affected by environment e.g. nutrition, sunlight.
	Environmental causes (condition they have developed in)	
	A combination of genes and environment	

Inherited disorders

The whole human genome has now been studied.

It is of great importance for future medical developments

- Searching for genes linked to different types of disease.
- Understanding and treatment of inherited disorders.
- Tracing migration patterns from the past.



Using a family tree: If the father was homozygous dominant then all of the offspring would have the disorder. He must be heterozygous

(Biology HT) Some disorders are inherited on the chromosomes that determine sex (sex linked genetic disorders)

Colour blindness in men.

The X chromosome carries one normal or one faulty allele (the normal is dominant). The shorter Y chromosome does not carry the allele and so X chromosome allele is always expressed in men.

One pair of chromosomes carry the genes that determine sex

Female	*Male*
XX	XY

Gametes	X	Y
X	XX	XY
X	XX	XY

The probability of a male of female child is 50%. The ratio is 1:1

Sex determination

Blood groups

Determined by multiple alleles (A,B,O) and codominance

- AO, AA blood group A
- BO, BB blood group B
- OO blood group O
- AB blood group AB. A and B are codominant

Genetic inheritance

Using a punnet square (using mouse fur colour as an example)

Parent phenotype	Black fur 	White fur 
Parent genotype	BB	bb
What gametes are present	In each egg B B	In each sperm b b

Gametes	b	b
B	Bb	Bb
B	Bb	Bb

The probability of black fur offspring phenotype is 100%. All offspring genotypes are heterozygous (Bb).

Crossing two heterozygous mice (Bb)

Gametes	B	b
B	BB	Bb
b	Bb	bb

The probability of black fur is 75% and white fur 25%. The ratio of black to white mice is 3:1

Dominant and recessive allele combinations

Dominant	*Recessive*
Represented by a capital letter e.g. B.	Represented by a lower case letter e.g. b.

3 possible combinations:
Homozygous dominant BB
Heterozygous dominant Bb
Homozygous recessive bb

Some characteristics are controlled by a single gene e.g. fur colour, colour blindness.

The alleles present, or genotype operate at a molecular level to develop characteristics that can be expressed as a phenotype.

Most characteristics are as a result of multiple genes interacting.

The understanding of genetics (biology only)

Gregor Mendel	*In the mid 19th century carried out breeding experiments on plants*	Inheritance of each characteristic is determined by units that are passed on to descendants unchanged.

Chromosomes had not yet been discovered so the mechanism for inheritance was still unknown.

Define terms linked to genetics	
Gamete	Sex cells produced in meiosis.
Zygote	Single cell that results from fusion of egg and sperm cell.
Chromosome	A long chain of DNA found in the nucleus.
Gene	Small section of DNA that codes for a particular protein.
Allele	Alternate forms of the same gene.
Dominant	A type of allele – always expressed if only one copy present and when paired with a recessive allele.
Recessive	A type of allele – only expressed when paired with another recessive allele.
Homozygous	Pair of the same alleles, dominant or recessive.
Heterozygous	Two different alleles are present 1 dominant and 1 recessive.
Genotype	Alleles that are present for a particular feature e.g. Bb or bb
Phenotype	Physical expression of an allele combination e.g. black fur, blonde hair, blue eyes.