



Very rarely a mutation will lead to a new phenotype which if is suited to environmental change can lead to rapid change in the species.

Embryo screening: small piece of developing placenta removed to check for presence of faulty genes

Gene therapy: replacing the faulty allele in somatic cells with a normal allele

Embryo screening and gene therapy may alleviate suffering

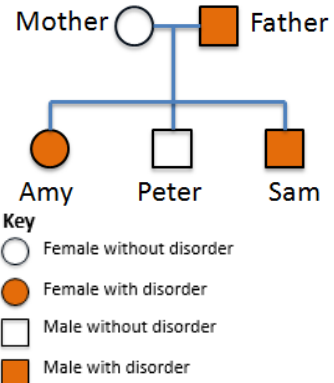
Embryo screening /gene therapy issues	Economic	Costly and not 100% reliable.
	Social	Not available to everyone (due to cost).
	Ethical	Should only 'healthy' embryos be implanted following screening.

Mutations occur continuously

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	

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



Inherited disorders

Some disorders are inherited. They are caused by the inheritance of certain alleles

Polydactyly	Cystic fibrosis
Caused by inheriting a dominant allele.	Caused by inheriting a recessive allele (both parents have to at least carry it).
Causes a person/animal to have extra toes or fingers.	A disorder of the cell membrane. Patients cannot control the viscosity of their mucus.

Sex determination

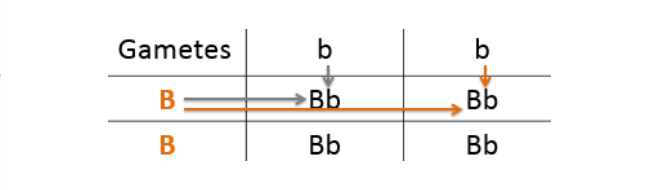
Ordinary human body cells contain 23 pairs of chromosomes

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

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



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

AQA GCSE INHERITANCE, VARIATION AND EVOLUTION PART 2

Variation

All genetic variation arises in mutation, most have no effect on phenotype, some influence but very few determine phenotype.

The genome and its interaction with the environment influence the development of phenotypes

Define terms linked to genetics	Gamete	Sex cells produced in meiosis.
	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.

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.

Genetic inheritance

The concept of probability in predicting results of a single gene cross.

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