Inheritance and variation L105-113

# AQA GCSE INHERITANCE, VARIATION AND EVOLUTION Part 1

## Meiosis

**Meiosis halves the number of chromosomes**

**Gametes are made in reproductive organs (in animals ovaries and testes)**

*Cells divide by meiosis to form gametes*

- Copies of the genetic information are made.
- The cell divides twice to form four gametes each with single set of chromosomes.
- All gametes are genetically different from each other.

**Gametes join at fertilisation to restore the number of chromosomes**

The new cell divides by mitosis. The number of cells increase. As the embryo develops cells differentiate.

**Meiosis leads to non-identical cells being formed while mitosis leads to identical cells being formed**

## Sexual and asexual reproduction

| | | |
|---|---|---|
| **Sexual reproduction involves the fusion of male and female gametes.** | *Sperm and egg in animals.* / *Pollen and egg cells in flowering plants.* | Produced by meiosis. There is mixing of genetic information which leads to a variety in the offspring. |
| **Asexual reproduction involves only one parent and no fusion of gametes.** | *e.g. cloning of females only in an aphid population.* | Only mitosis is involved. There is no mixing of genetic information. This leads to genetically identical clones. |

## Advantages and disadvantages of sexual and asexual reproduction (Biology only)

**Reproduction advantages/disadvantages**

| *Sexual* | *Asexual* |
|---|---|
| Needs two parents. | Only one parent needed (quicker). |
| Produces variation in the offspring. | Identical offspring (no variation). |
| If the environment changes variation gives a survival advantage by natural selection. | Vulnerable to rapidly changing conditions due to lack of variation. |
| Negative mutations are not always inherited. | Negative mutation can affect all offspring. |
| Natural selection can by speeded up using selective breeding to increase food production. | Food/medicine production can be extremely quick. |

| | | | |
|---|---|---|---|
| **Some organisms use both methods depending on the circumstances** | *Malarial parasites* |  | Asexually in the human host but sexually in a mosquito. |
| | *Fungi* |  | Asexually by spores, sexually to give variation. |
| | *Plants* |  | Produce seeds sexually, asexually by runners in strawberry plants, bulbs division in daffodils. |

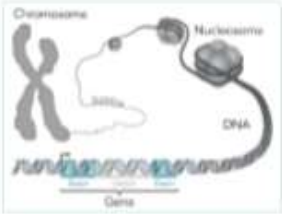
## DNA and the genome

**Genetic material in the nucleus is composed of a chemical called DNA.**

**DNA structure**

*Polymer made up of two strands forming a double helix.*

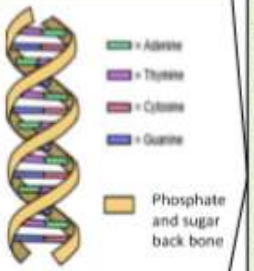
Contained in structures called chromosomes. A gene is a small section of DNA on a chromosome. Each gene codes for a sequence of amino acids to make a specific protein.




The genome is the entire genetic material of an organism.

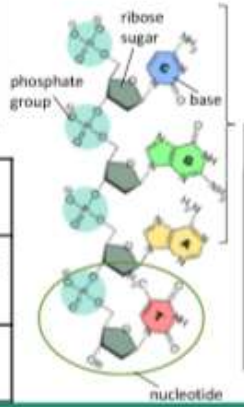
**(HT only)** Not all parts code for proteins. Non-coding parts can switch genes on and off. Mutations may affect how genes are expressed.

## DNA structure (Biology only)




**DNA is polymer made from four different nucleotides. Each nucleotide consists of a common sugar, phosphate group and one of 4 different bases A, C, G & T**

Repeating nucleotide units.




| | | |
|---|---|---|
| **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. |

## Mutations occur continuously (HT only)

When the protein chain is complete it folds to form a unique shape. This allows proteins to do their job as enzymes, hormones or new structures such as collagen.

Some change the shape and affect the function of proteins e.g. and enzyme active site will change or a structural protein loses its strength

Most do not alter the protein so that its appearance or function is not changed.

## Protein synthesis (HT only)

In DNA the complementary strands C, A, T, G always link in the same way. C always linked to G on the opposite strand and A to T.

## (HT) Making new proteins (protein synthesis)

*Composed of chains of amino acids. A sequence of 3 bases codes for a particular amino acid.*

DNA in the nucleus unravels.

⇩

Enzymes make a copy of the DNA strand called mRNA.

⇩

mRNA moves from the nucleus to ribosome in the cytoplasm.

⇩

Ribosomes translate each 3 bases into amino acids according to mRNA template

⇩

Ribosomes link amino acids brought by carrier proteins.

⇩

A long chain of amino acids form. Their specific order forms a specific protein.

A sequence of 3 bases is the code for a particular amino acid. The order of bases controls the order in which each amino acid is assemble to produce a specific protein.

# AQA GCSE INHERITANCE, VARIATION AND EVOLUTION PART 2

## 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* | |

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

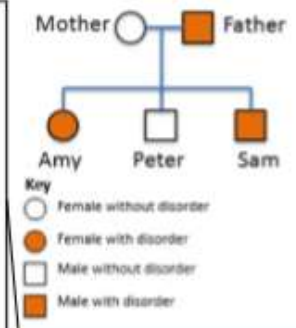
**Mutations occur continuously**

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.

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

## Inherited disorders

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




**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. |

**Embryo screening and gene therapy may alleviate suffering**

*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 /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. |

## 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

## 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

**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

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.

## 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. |