Edexcel (B) Biology A-level

Topic 8: Origins of Genetic Variation

Notes

Meiosis is a reduction division. The main role of meiosis is the **production of haploid gametes** as cells produced by meiosis have half the number of chromosomes of normal body (somatic) cells.

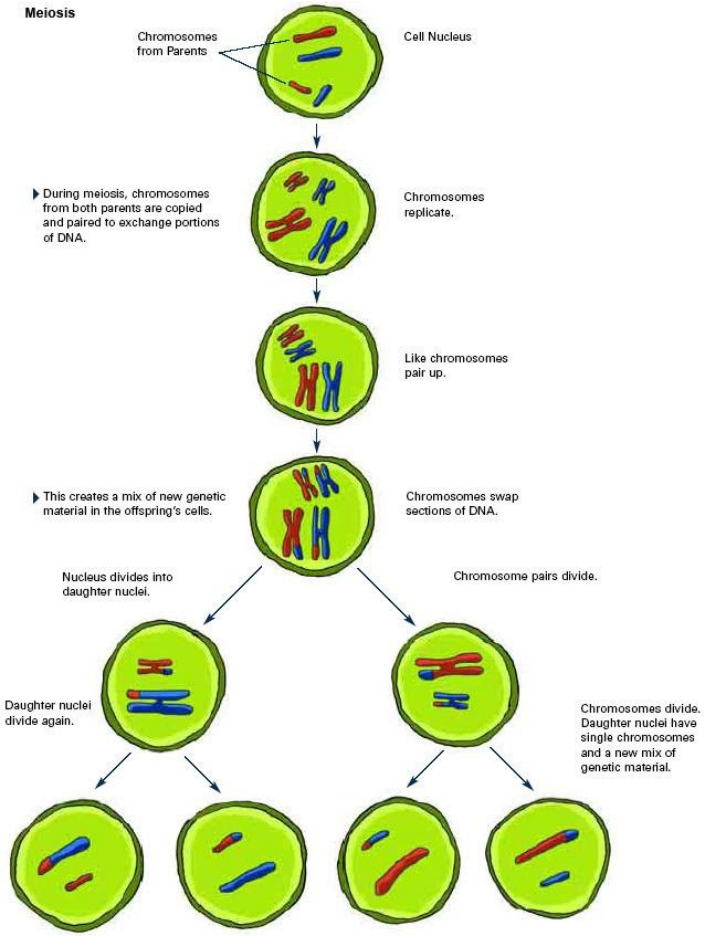

Figure: Biologypost - Meiosis

Mutation is the source of genetic variation. Meiosis introduces variation via the production of gametes with new combinations of alleles. This is achieved via:

- **Crossing over,** where sections of DNA are exchanged between homologous chromosomes.
- **Independent assortment,** which describes the different possible combinations of maternal and paternal chromosomes.

Random fertilisation also introduces variation because the combination of gametes that fuses to form the zygote is random.

Keywords:

Allele – an alternative form of a gene.

Phenotype – observable characteristics of an organism.

Genotype – the alleles present within cells of an organism.

Dominant – an allele that is always expressed in the phenotype, even if only one copy of the allele is present.

Recessive – an allele that is only expressed in the phenotype if two copies of the allele are present i.e. homozygous recessive.

Homozygous – two identical alleles of a gene (a homozygous organism is called a homozygote).

Heterozygous – two different alleles of a gene (a heterozygous organism is called a heterozygote).

Codominance – both alleles contribute to the phenotype without mixing e.g. blood type.

Multiple Alleles - more than two possible versions of an allele.

Chi-Squared Test

$$X^2 = \sum \frac{(\text{observed - expected})^2}{\text{expected}}$$

The **chi-squared test** is a **statistical** test which can be used to establish whether the difference between **observed and expected results** is significant.

- It can only be used if the **sample size** is sufficiently large (>20). It can only be used for **discontinuous variation** data.
- The chi-squared test can be used to determine whether the **null hypothesis** is correct or not. The null hypothesis is the assumption that there is no significant difference between observed and expected results.
- The value obtained is compared to the **critical value**, and in a case where the value obtained is less than the critical value, the null hypothesis is accepted as the difference is not significant.
- In a case where the x^2 value is greater than the critical value, the null hypothesis is rejected, meaning that the difference between observed and expected results is very unlikely to occur due to chance and is significant.

Inheritance

If the null hypothesis is rejected, this implies **autosomal gene linkage** e.g. colour and wing length in *Drosphilia.* This results from the presence of alleles on the **same chromosome** - they are linked and are inherited, to a greater or lesser extent, as if they were the same gene. The closer together the genes are on the chromosome, the more closely linked they are, and the less likely they are to be separated by recombination.

Some genes are **sex-linked** - they **appear on the X chromosome**. For this reason, some genetic disorders are much more common in men because they don't have another copy of the allele for a particular characteristic, and therefore only need one copy for the allele to be expressed e.g. haemophilia (absence of clotting factor VIII and therefore blood that doesn't clot properly), red-green colourblindness.

Natural Selection and Evolution

Allele frequencies change over time due to natural selection leading to evolution via selection pressures acting on the gene pool. There are three types of natural selection:

- **Stabilising** (reduces variation in the population - conserves phenotypes already present - selects against extreme phenotypes).
- **Disruptive** (increase in the diversity of a population - common when conditions are diverse and small subpopulations evolve different phenotypes suited to their niche - selects for extreme phenotypes).
- **Directional** ('classic' natural selection - shows a change from one phenotype to another, which is more advantageous to the environment).

Natural selection and therefore allele frequency can be dramatically affected by certain factors:

- **Population bottleneck** - the effect of a catastrophic event or series of events that dramatically reduces the size of a population (by at least 50%) and causes a severe decrease in the gene pool of the population, resulting in large changes in allele frequencies and a reduction in genetic diversity.
- **Founder effect** - the loss of genetic variation that occurs when a small number of individuals become isolated, forming a new population with allele frequencies not representative of the original population (also referred to as a voluntary bottleneck).

Sometimes change in allele frequency is not due to natural selection:

- **Genetic drift** = random changes in the gene pool of a population that occur by chance, not because they confer any advantage or disadvantage to the offspring. Has a major effect on small populations, e.g. after a population bottleneck.

The **Hardy-Weinberg equation** can be used to **estimate the frequency of alleles in a population**:

p = the frequency of the dominant allele (represented by A)
q = the frequency of the recessive allele (represented by a)
p^2 = frequency of AA (homozygous dominant)
2pq = frequency of Aa (heterozygous)
q^2 = frequency of aa (homozygous recessive)

For a population in genetic equilibrium:

$p + q = 1.0$

$(p + q)^2 = 1$ hence
$p^2 + 2pq + q^2 = 1$

Conditions for the Hardy-Weinberg Equation:

- No mutations
- Random mating
- Large population
- No migration into or out of the population (i.e. the population is isolated)
- No selection pressure