Gene expression, epigenetics and phenotype

How cells specialise through differential gene expression, post-transcriptional changes, the effect of the environment, epigenetics, and multiple alleles and polygenic inheritance.

Differential gene expression

All cells of an organism (apart from gametes and a few others) contain the **same genes**, but different cell types **express different genes**. A cell **specialises** through **differential gene expression**: some genes are **switched on**, producing active **mRNA**, which is translated into proteins. These proteins control cell processes or determine the structure of the cell.

For example, a pancreatic cell expresses the insulin gene, but a developing red blood cell expresses the haemoglobin genes (and later loses its nucleus). **Transcription factors** are proteins that bind to DNA near a gene and switch it on or off.

In animals and in plants, this is how cells become specialised during development from a totipotent or pluripotent stem cell.

One gene, more than one protein

A eukaryotic gene contains **exons** (coding) and **introns** (non-coding). The **pre-mRNA** is copied from the whole gene, then **spliced** to remove introns.

Alternative splicing joins the exons in **different combinations**, so **one gene can give rise to more than one mRNA** and therefore more than one protein. This explains why humans have more proteins than genes. Other post-transcriptional changes include adding a cap and a tail to the mRNA.

Genotype and environment

Phenotype is the result of an interaction between the **genotype** and the **environment**. Examples:

- Human height depends on genes and on nutrition.
- Hydrangea flower colour depends on the soil pH.
- Himalayan rabbits have dark fur on the cool extremities, because the enzyme is active only at lower temperatures.

Epigenetics

Epigenetic modification changes the **activity of genes without changing the DNA base sequence**.

- **DNA methylation**: a methyl group (CH_3) is added to **cytosine** bases (often in promoter regions). This **blocks transcription factors** from binding, so the gene is **switched off**.

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- **Histone modification**: DNA is wrapped round **histone** proteins. **Acetylation** of histones loosens the DNA so the genes can be transcribed (**on**); **deacetylation** and methylation make the chromatin **tightly packed** so the genes cannot be transcribed (**off**).

These changes can be **passed on** when cells divide (so a specialised cell gives rise to cells of the same type), and in some cases between generations.

> **NOTE**
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> Epigenetic effects show how the environment (diet, stress, smoking) can alter gene activity. Identical twins start with the same DNA but their patterns of methylation diverge with age, so they differ in disease risks.

Multiple alleles and polygenic inheritance

- **Multiple alleles**: a gene with **more than two alleles**, although each individual has only two. Human **ABO blood groups** are controlled by three alleles (I^A, I^B and I^O), with I^A and I^B codominant and both dominant to I^O.
- **Polygenic inheritance**: a characteristic controlled by **many genes** with small, additive effects, which gives **continuous variation**, such as height, skin colour and milk yield. The distribution is a **normal curve** (bell-shaped).
- The environment also affects such traits, so the phenotype shows a continuous range.

	Discontinuous variation	Continuous variation
Genes	One or a few genes	Many genes (polygenic)
Environment	Little effect	Large effect
Phenotype classes	Distinct categories (blood group)	A range (height, mass)
Graph	Bar chart	Histogram, normal curve

Explaining continuous variation needs three points: many genes each with a small effect, several alleles at each gene, and environmental influence.

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