



Gene expression

Revision notes • IB Biology HL D2.2: regulation of transcription and mRNA breakdown, genome vs transcriptome vs proteome, methylation, epigenetic inheritance and twins.

For IB Diploma Biology, Standard Level and Higher Level (first assessment 2025). **HL only** is additional higher level content, for HL students only.

Gene expression Spec D2.2 HL only

Gene expression and its regulation Spec D2.2.1, D2.2.2, D2.2.3

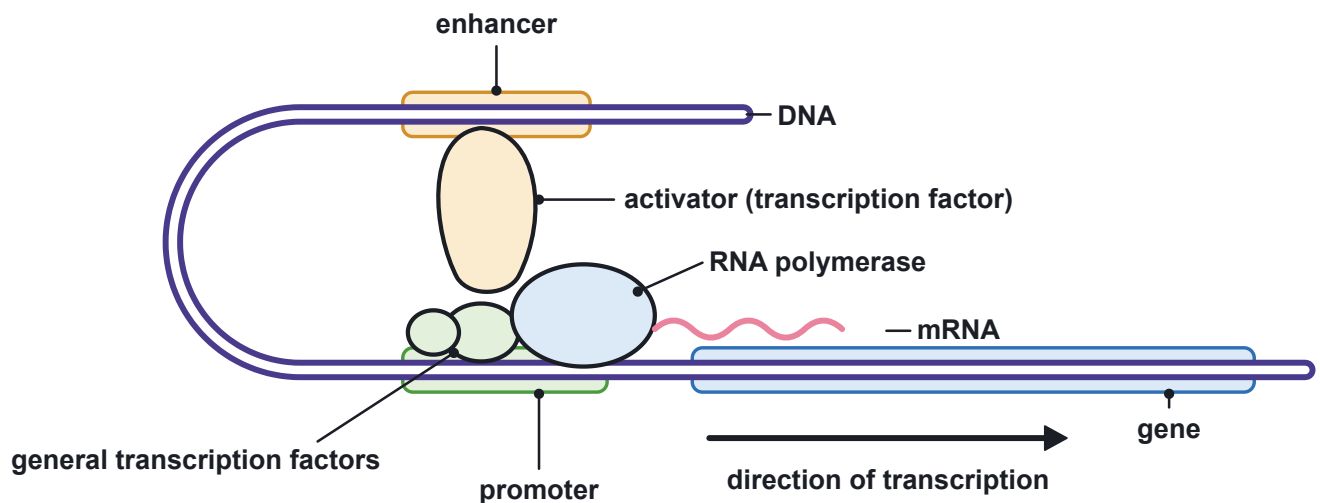
Gene expression is the mechanism by which the information in a gene has an effect on the **phenotype**. The most common stages are **transcription** (making mRNA from the gene), **translation** (making a polypeptide from the mRNA) and the **function of the protein product**, for example an enzyme that catalyses a reaction producing a pigment.

Regulation of transcription

Transcription is regulated by **proteins that bind to specific base sequences in DNA**. Each binding protein has a shape that fits only one base sequence.

Promoter	A base sequence just before (upstream of) a gene where RNA polymerase binds to start transcription.
Transcription factor	A protein that binds to a specific base sequence in DNA, such as a promoter or enhancer, and increases or decreases the rate of transcription of a gene.
Enhancer	A base sequence, which can be some distance from the gene, where activator transcription factors bind to increase the rate of transcription.

RNA polymerase usually cannot bind to a promoter on its own. **General transcription factors** must bind to the promoter first. **Activator** proteins binding to enhancers increase the rate of transcription, while **repressor** proteins can block it. Different cells contain different transcription factors, so they express different genes.



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An **activator** bound to an **enhancer** interacts with the transcription factors and RNA polymerase at the **promoter**, increasing the rate of transcription.

Control of mRNA degradation

The amount of protein made also depends on how long each mRNA molecule lasts, because the same mRNA can be translated many times. mRNA is broken down by enzymes called **nucleases**. In human cells, mRNA may persist for anything from **minutes up to days**. By changing the rate of breakdown of a particular mRNA, a cell regulates how much **translation** of that gene happens.

Exam tip: Linking question: what mechanisms are there for inhibition in biological systems? Repressor proteins blocking transcription and nucleases destroying mRNA both inhibit gene expression; compare them with enzyme inhibition (C1.1).

Epigenesis, the transcriptome and epigenetic tags Spec D2.2.4, D2.2.5, D2.2.6

Epigenesis is the development of patterns of differentiation in the cells of a multicellular organism. A zygote divides to form cells that become specialised in different ways: muscle, nerve, skin. All these cells have the same DNA. What differs is which genes are switched on.

Epigenetic changes **do not alter the DNA base sequence**. So they alter the **phenotype** of a cell, but not its **genotype**.

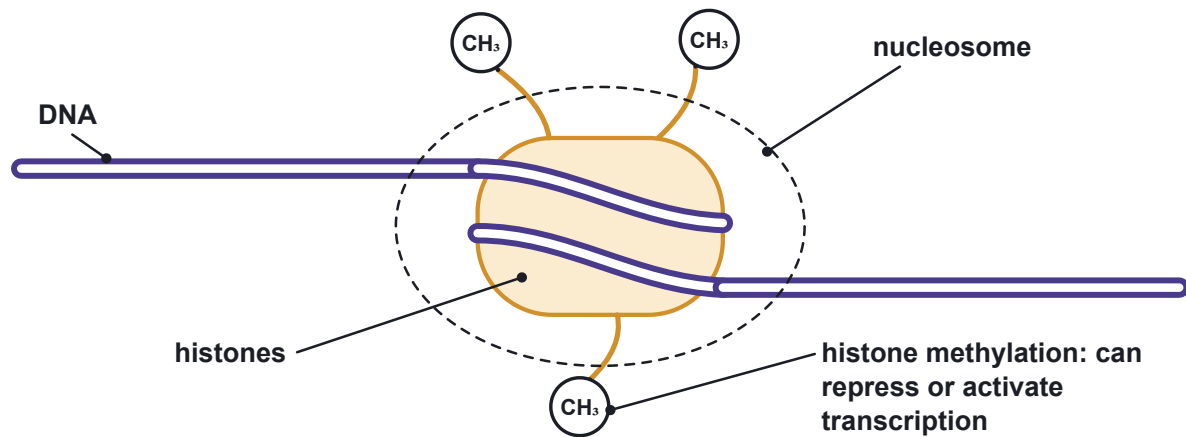
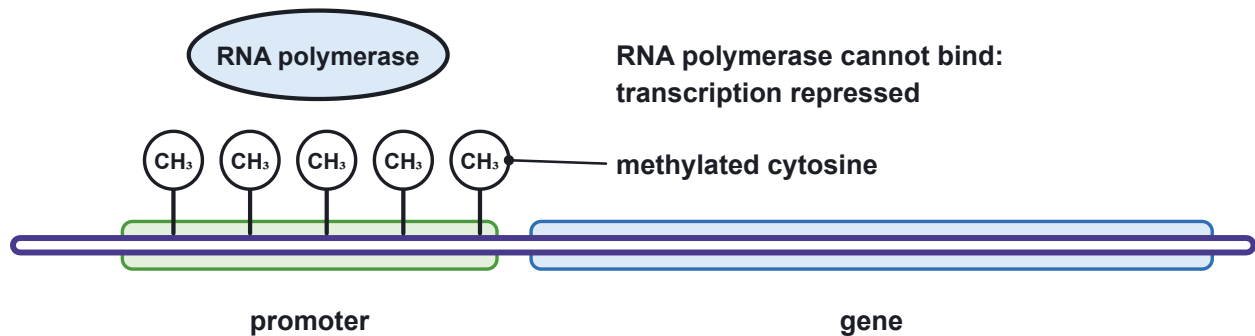
Genome, transcriptome and proteome

Three sets of molecules in a cell		
Term	Meaning	Same in all cells of an organism?
Genome	All of the genetic information (DNA) of the cell	Yes (almost always)
Transcriptome	All the RNA molecules transcribed in the cell at a given time	No: depends on which genes are being transcribed
Proteome	All the proteins produced by the cell at a given time	No: depends on which mRNAs are translated

No cell expresses all of its genes. The pattern of gene expression in a cell determines how it differentiates. A red blood cell precursor transcribes the haemoglobin genes, for example, but not the genes for the digestive enzymes made in the pancreas.

Methylation as an epigenetic tag

- **DNA methylation:** methyl groups are added to **cytosine** bases in the DNA of a **promoter**. This **represses transcription**, so the gene downstream is not expressed.
- **Histone methylation:** DNA is wound around histones in **nucleosomes**. Methyl groups added to amino acids in the histones can cause transcription to be **repressed or activated**, depending on which amino acid is methylated.



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Epigenetic tags: methylation of cytosine in a promoter represses transcription; methylation of histones can repress or activate it.

Common mistake: Methylation does not change a base into another base or change the sequence. The base is still cytosine; it just carries a methyl tag.

Epigenetic inheritance and the environment Spec D2.2.7, D2.2.8, D2.2.9, D2.2.10

Epigenetic inheritance is the passing on of phenotypic changes in a cell or organism to daughter cells or offspring **without changes in the nucleotide sequence of DNA**. This can happen if epigenetic tags, such as DNA methylation or histone modification, **remain in place during mitosis or meiosis**.

In mitosis, methylation patterns are copied onto new DNA, so a differentiated cell such as a skin cell produces more skin cells. This is how a tissue keeps its pattern of gene expression.

Environmental effects on gene expression

The environment can change epigenetic tags. **Air pollution** is the example to learn. Studies of people exposed to high levels of air pollution, such as fine particles and polycyclic aromatic hydrocarbons (PAHs) from traffic and burning fuels, have found altered **methyl tags on DNA** in their blood cells. Some genes gain methyl tags and others lose them. For example, children with asthma living in a city with heavily polluted air had more methylation of the promoter of *FOXP3*, a gene needed by regulatory T

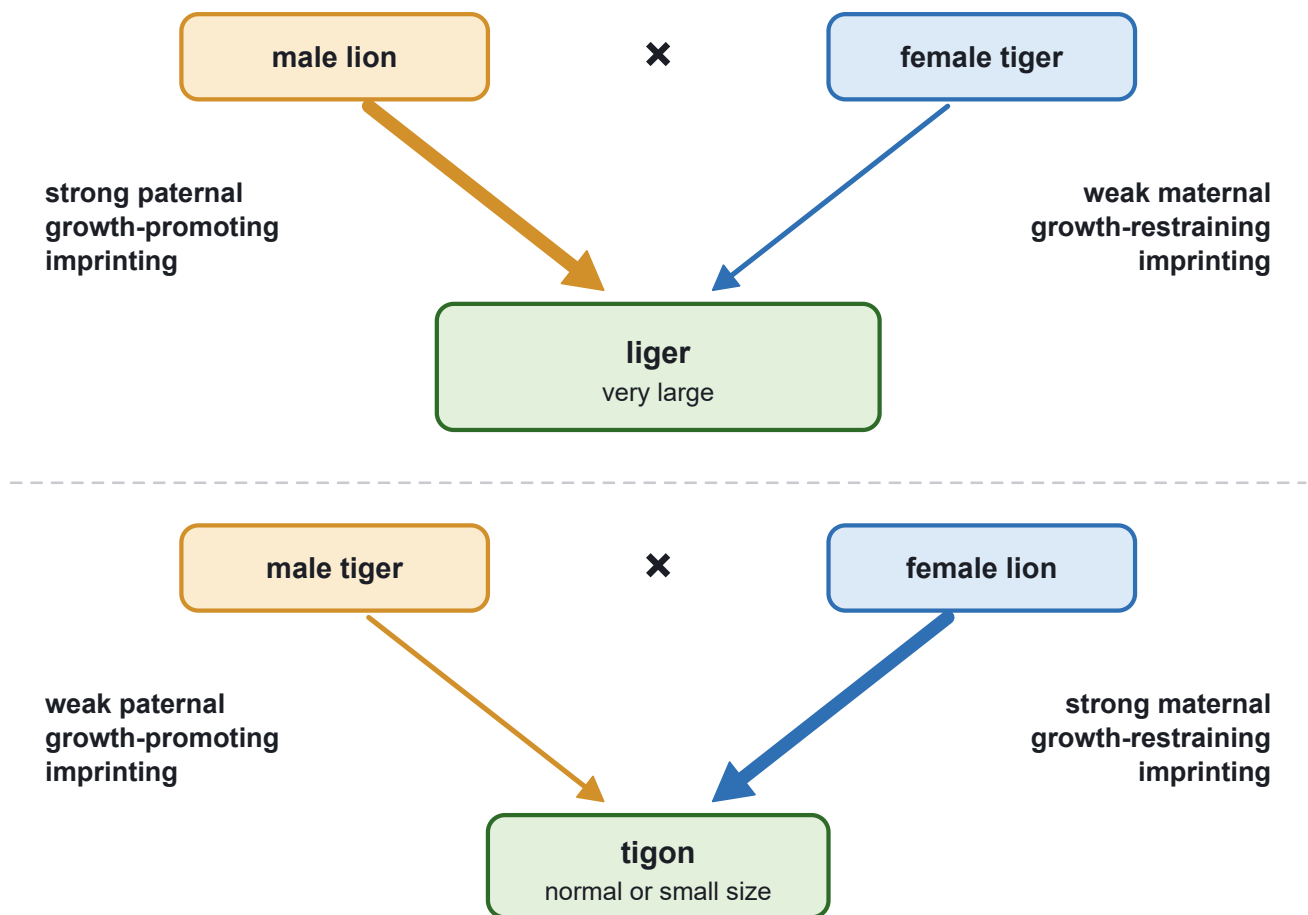
cells that dampen immune responses, than children with asthma in a cleaner city. More methylation means less transcription, so their regulatory T cells worked less well, which is linked to worse asthma symptoms.

Removal of most but not all tags from gametes

During the formation of the **ovum and sperm**, and soon after fertilisation, **most** epigenetic tags are removed, so the zygote can develop all cell types. **Not all** are removed. Some genes keep tags that depend on which parent they came from. This is **genomic imprinting**: for an imprinted gene, the copy from one parent is silenced by methylation, so the gene is expressed only from the mother's copy or only from the father's copy. Several imprinted genes control growth before birth. In general, some **growth-promoting** genes are expressed only from the father's copy, and some **growth-restraining** genes only from the mother's copy.

Tigons and ligers (lion-tiger hybrids)		
Hybrid	Parents	Phenotype
Liger	Male lion × female tiger	Grows much larger than either parent
Tigon	Male tiger × female lion	Similar in size to, or smaller than, the parents

Both hybrids have one set of chromosomes from a lion and one from a tiger, so the difference in size is put down to **epigenetics**: which parent each set of imprinted genes came from. The most widely accepted hypothesis is this. In lions, the paternal growth-promoting imprinting is strong, and it is balanced by strong maternal growth-restraining imprinting in lionesses. In tigers, both effects are weaker. A **liger** inherits strong growth promotion from its lion father but only weak restraint from its tigress mother, so it grows much larger than either parent. A **tigon** inherits only weak growth promotion from its tiger father but strong restraint from its lioness mother, so it is no larger than its parents and is often smaller.



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Genomic imprinting explains why ligers grow much larger than tigons. Arrow thickness shows the strength of each effect.

Monozygotic twin studies

Monozygotic (identical) twins develop from one zygote, so they have the same genome. Any differences between them must come from the **environment** and its effect on gene expression. Studies comparing twins show that their methylation patterns are very similar when they are young but become **more different as they get older**, especially if they have lived apart with different diets, habits and exposures. This shows the environment changing epigenetic tags.

Exam tip: Linking question: in what ways does the environment stimulate diversification? Twin studies are the classic evidence that the same genotype can produce different phenotypes through epigenetic changes.

External factors and gene expression Spec D2.2.11

Cells change which genes they express in response to signals from outside the cell. Two examples are a hormone in animals and a nutrient in bacteria.

A hormone: oestrogen

1. Oestrogen is a steroid hormone, so it diffuses through the cell membrane of target cells, for example cells of the uterus lining.
2. Inside the cell (mainly in the nucleus) it binds to a specific **oestrogen receptor** protein.
3. The hormone–receptor complex acts as a **transcription factor**: it binds to specific base sequences in DNA near target genes.

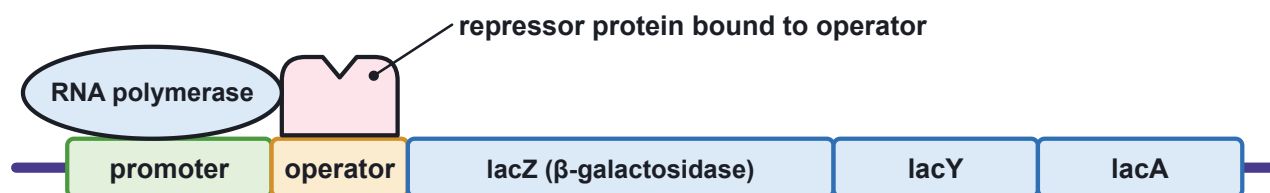
4. Transcription of those genes increases, so the cell makes new proteins, for example those needed for the uterus lining to thicken.

A biochemical: lactose in *E. coli*

The bacterium *Escherichia coli* only makes the enzymes for using lactose when lactose is present. The genes for these proteins (including β -galactosidase, which hydrolyses lactose) are together in the **lac operon**, controlled by one promoter and an **operator** sequence.

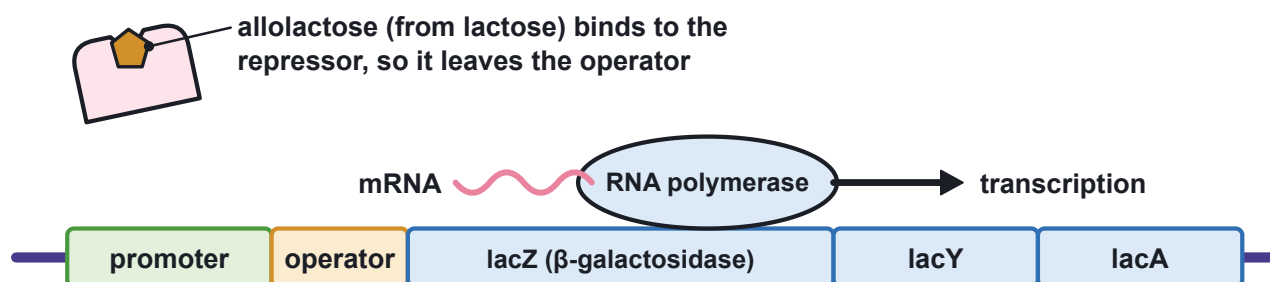
The lac operon		
Condition	What happens	Result
Lactose absent	A repressor protein binds to the operator and blocks RNA polymerase	Genes not transcribed: no enzymes wasted
Lactose present	Allolactose (an isomer formed from lactose) binds to the repressor, changing its shape so it detaches from the operator	RNA polymerase transcribes the genes; enzymes for lactose uptake and breakdown are made

Lactose absent



RNA polymerase is blocked: the genes are not transcribed and no enzymes are made

Lactose present



RNA polymerase transcribes the genes: enzymes for lactose uptake and breakdown are made

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The **lac operon**: with no lactose the repressor blocks transcription; lactose (as allolactose) releases the repressor so the genes are transcribed.

Exam tip: The guide asks for one hormone and one biochemical such as lactose or tryptophan. Learn one of each thoroughly and describe the binding to a specific DNA base sequence.

Quick check

- 1 . What are the three most common stages in gene expression?
- 2 . What is a transcription factor?
- 3 . What effect does methylation of cytosine in a promoter have?
- 4 . Why do monozygotic twins become more different with age?

Quick check answers

Gene expression

- 1 . Transcription, translation and the function of the protein product (such as an enzyme).
- 2 . A protein that binds to a specific base sequence in DNA, such as a promoter or enhancer, to regulate transcription.
- 3 . It represses transcription of the gene downstream.
- 4 . They have identical genomes, but different environments change their epigenetic tags and so their gene expression.