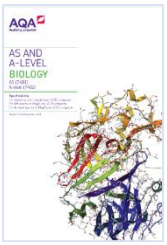
St. Ambrose College

A Level Biology (Year 12)

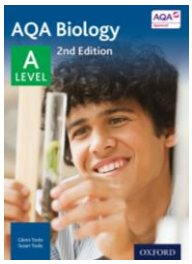
Knowledge Organiser: Unit 4 Genetic information, variation and relationship between organisms (4.1-4.3)

4.1 DNA, genes and chromosomes
4.2 DNA and protein synthesis
4.3 Genetic diversity, mutation and meiosis

For every 1 hour A Level Biology lesson you are expected to spend at least 1 hour independently reviewing the subject content. The following resources should be referred to regularly to support your independent work.



You have been provided with a printed copy of the full subject specification (also available on the AQA website https://www.aqa.org.uk/subjects/science/as-and-a-level/biology-7401-7402/specification-at-a-glance). Use this to follow the learning in lessons…track your progress and be aware of what is still to come.



kerboodle

Use the textbook on www.kerboodle.com after every lesson to develop your understanding. Read the relevant pages, add detail to your class notes and complete the summary tasks. Create your own summary notes/flashcards for future use in the run up to exams.

Unit 4 Genes and Variation on pages

SENECA
www.senecalearning.com

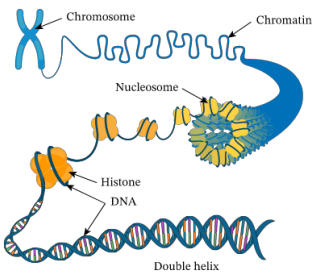
Use regularly between lessons to review basic content and to become more familiar with key terminology. https://senecalearning.com/en-GB/

PMT
resources · tuition · courses

Access detailed revision notes, key definitions, flash cards, past paper questions and mark schemes.
https://www.physicsandmathstutor.com/biology-revision/a-level-aqa/

As an A Level student you are expected to take a proactive approach to your studies; arrive to lessons fully equipped and prepared for what you will be learning about (read ahead in the specification/textbook), focus and participate in lessons, ask for help/clarification when you are unsure and spend time after the lesson consolidating/embedding new learning.

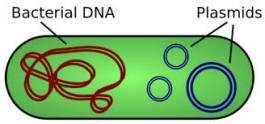
4.1 DNA, genes and chromosomes



DNA is stored differently in eukaryotes vs. prokaryotes

- **Eukaryotic DNA:** Long, linear, associated with proteins called histones, tightly coiled into chromosomes (DNA molecule + its associated proteins)

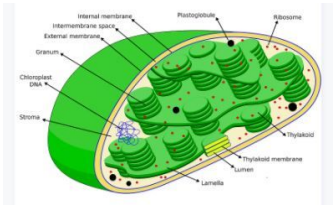
- **Prokaryotic DNA:** Short, circular, not associated with proteins/histones

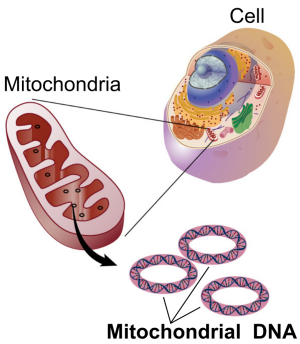


Mitochondria and chloroplasts in eukaryotic cells have their own DNA

- Similar to prokaryotic DNA – short, circular, not associated with proteins/histones

This is evidence for endosymbiotic theory of eukaryotic evolution





The Genetic Code:

- **Sequence of DNA bases** that codes for:
  - The amino acid sequence (primary structure) of a polypeptide
  - A functional RNA e.g. ribosomal RNA and tRNAs
- A gene occupies a fixed position, called a **locus**, on a particular DNA molecules
- Sequence of DNA **bases read in triplets**
  - a DNA base triplet (or mRNA codon) : sequence of 3 bases coding for a specific amino acid
    - e.g. UAU codes for tyrosine
- **Universal**
  - The same specific DNA base triplets code for the same amino acids in all living organisms
    - e.g. UAU codes for tyrosine in all organisms
- **Non-overlapping**
  - Discrete, each base can only be used once and in only one triplet
- **Degenerate**
  - The same amino acid can be coded for by more than one base triplet
    - e.g. tyrosine can be coded for by UAU or UAC

In eukaryotes, much of the DNA doesn't code for polypeptides

- Between genes…
  - Non-coding **multiple repeats** (or Variable Number Tandem Repeats (VNTRs))
- Within genes
  - Only **exons** code for amino acid sequences ***(exons expressed)***, which are separated by one or more non-coding sequences, called **introns** ***(interfering introns)***

More important definitions

- **Genome:** the complete set of genes in a cell, including those in mitochondria and/or chloroplasts
- **Proteome:** The full range of proteins that a cell/genome is able to produce
- **Alleles:** different version (sequence of bases / triplets) of the same gene
- **Homologous pair of chromosomes:** same size chromosomes with same genes, but different alleles

4.2 DNA and protein sysnthesis

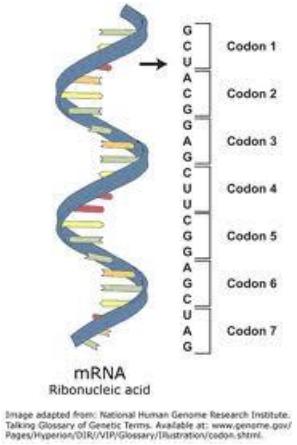
Protein synthesis overview

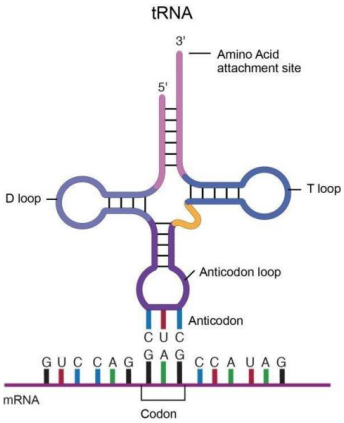
Two main stages:

1. **Transcription**
   - Production of mRNA from DNA (making a 'gene copy')
   - Occurs in the nucleus
2. **Translation**
   - Production of polypeptides (joining together amino acids) from the sequence of codons carried by mRNA (reading the mRNA 'gene copy')
   - Occurs in the cytoplasm on/at ribosomes

Messenger RNA (mRNA)

- Made by transcription in the nucleus
- Acts as a template for translation in the cytoplasm
- Sequence of bases on RNA determines sequence of amino acids in polypeptide chain
- Short, straight chain molecule - single stranded (chemically unstable/short lived)
- Sequence of bases (A,C,**U,**G) on RNA determined by sequence of bases on the template DNA strand (antisense strand)
- Triplet code = codon





Transfer RNA (tRNA)

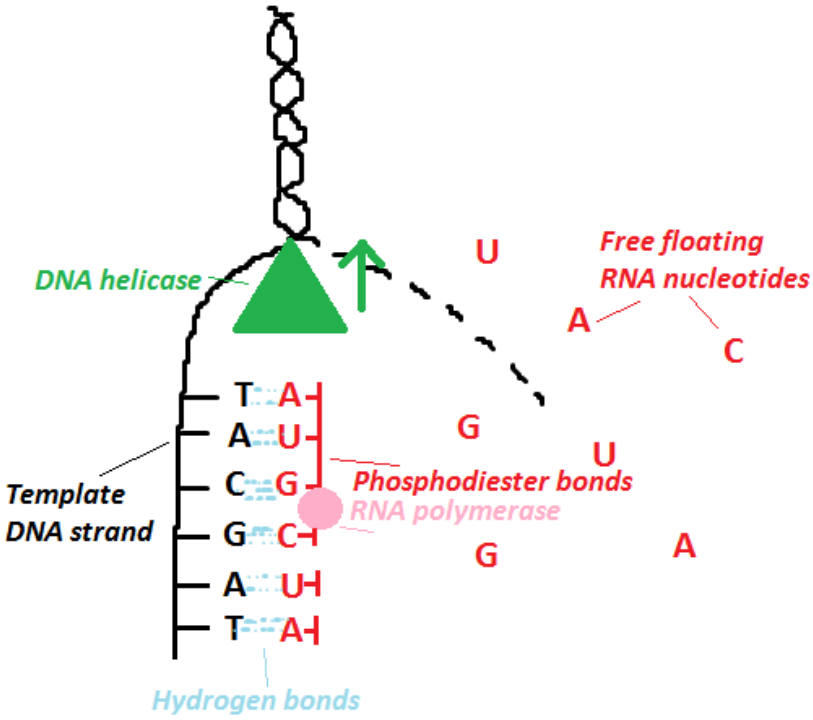
- Short, single polynucleotide strand
  - Folded (3 hairpin loops) into a three-leafed clover shape held together by hydrogen bonds
  - Amino acid binding site
  - Carries an amino acid
- Anticodon = 3 exposed bases
  - Anticodon bases complementary to mRNA codon
- Each tRNA specific to one amino acid, in relation to its anticodon

Comparing the structure of messenger RNA (mRNA) and Transfer RNA (tRNA)

- Similarities
  - Both RNA = made of RNA nucleotides
  - single stranded
- Differences
  - mRNA straight, whereas tRNA folded into clover shape
  - mRNA is a longer, variable length, whereas tRNA is shorter
  - mRNA contains no paired bases or hydrogen bonds, whereas tRNA has some paired bases and hydrogen bonds

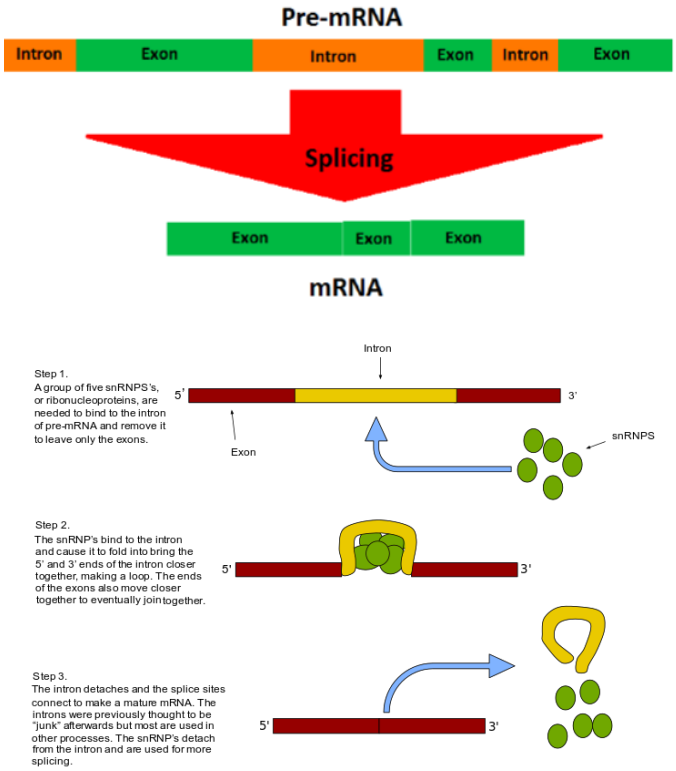
Transcription

- Occurs in the **nucleus**
- required section of DNA double helix **unzipped / unwound** by helicase
    - Hydrogen bonds broken
- RNA nucleotides align next to their **complementary bases** on the template strand forming (**temporary**) hydrogen bonds *(Uracil replaces thymine in RNA)*
- **RNA polymerase** joins adjacent nucleotides - condensation reaction forming phosphodiester bonds
- When RNA polymerase reaches stop codon, mRNA (prokaryotes) or pre-mRNA (eukaryotes) detaches from DNA



Post transcriptional modification

- Eukaryotic genes contain
    - Exons – coding regions
    - Introns – non-coding regions
- The whole gene is copied (transcribed) druring transcription creating pre-mRNA ***(Pre-mRNA contains introns & exons)***
- **Splicing has to occur before the mRNA can leave** the **nucleus** via nuclear pores in the envelope
    - Introns removed
    - Exons spliced together
      (Exons can be spliced together in a variety of different combos increasing the number/variety of different proteins that can be produced from a single gene!!)



- Because Prokaryotic DNA doesn't contain introns, the mRNA produced directly from DNA during transcription can be used in translation
    - **No splicing required in prokaryotes!**

Translation

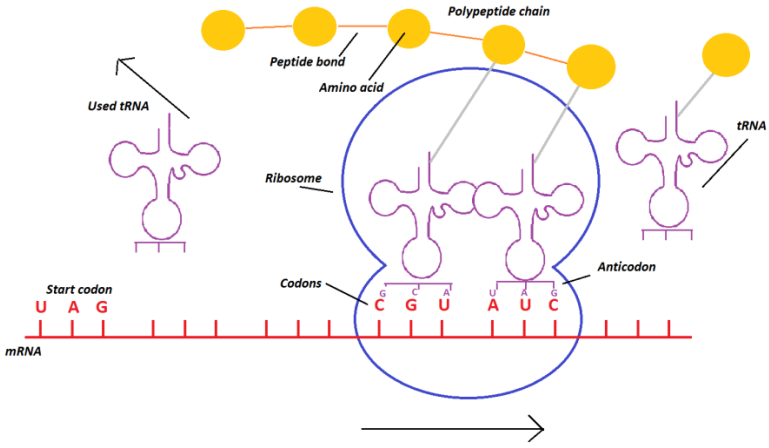
- Sequence of mRNA codons (base triplets) determines sequence of amino acids in a polypeptide chain.

- mRNA attaches to a ribosome
- the ribosome covers 6 bases (2 codons) at a time

- tRNA molecules carry specific amino acids, in relation to their anticodon
- At the ribosome, tRNAs bind to mRNA codons
    - complementary base pairing of tRNA anticodon to mRNA codon
    - Hydrogen bonds form between complementary bases (temporarily hold the tRNAs in place)

- The two adjacent amino acids joined by condensation, forming a peptide bond
    - Using energy from ATP
    - amino acids detach from tRNAs
- tRNA (without its amino acid) leaves the ribosome, ribosome moves along mRNA to next codon

- Stages repeated and continues until stop codon (completed polypeptide chain released)

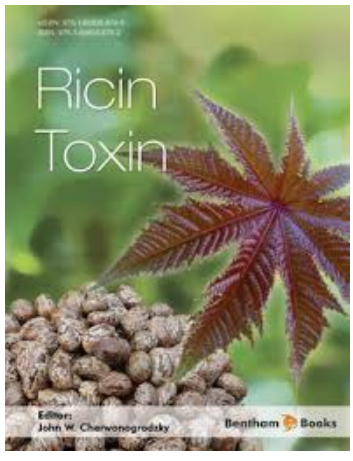


Example Exam Question

Ricin is a protein produced by some plants. In animal cells, ricin acts as an enzyme. This enzyme removes the adenine molecule from one of the nucleotides in the RNA of ribosomes. As a result, the ribosome changes shape.

(a) Ricin causes the death of cells and is very poisonous to many animals.
Suggest how the action of ricin on ribosomes could cause the death of cells. (2)

- ✓ Stops translation / formation of a protein / identified protein
- ✓ Prevents mRNA / tRNA binding
- ✓ Prevents formation of peptide bond
- ✓ Consequence of loss of identified protein



(b) Ricin is found in high concentrations in the seeds of some species of plant.
Suggest and explain one advantage of this to the plant. (2)

- ✓ Animals that can eat the plant get ill / killed / avoid plant
- ✓ So seeds / plants / species more likely to survive

4.3 Genetic diversity can arise as a result of mutation or during meiosis

What is a gene mutation?

- A change in the base sequence of DNA (on chromosomes)
- Can arise spontaneously during DNA replication (S phase of interphase)
- May involves base deletion / insertion or substitution
- Increase the rate of gene mutation (above the rate of naturally occurring mutations)
- E.g. ultraviolet light or alpha particles

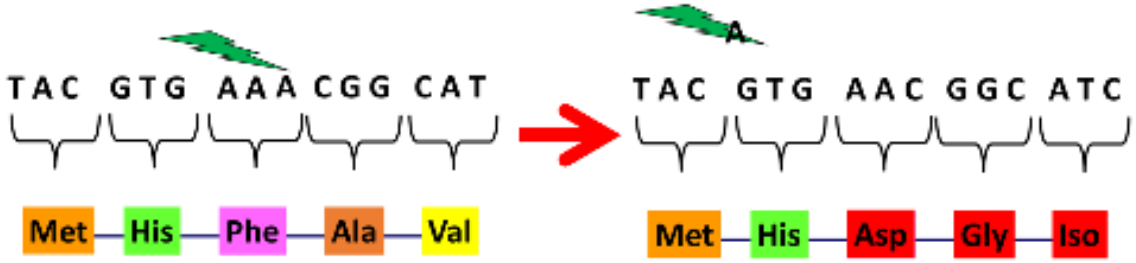
A mutation can lead to the production of a non-functional protein (eg. enzyme)

Change in base / triplet sequence of DNA / gene...

- Changes the **codons on mRNA**
- Changes sequence of amino acids in the **primary structure** of the polypeptide
- Changes position of hydrogen / ionic / disulphide **bonds in tertiary structure** of protein
- Changes **tertiary structure / shape** of the protein (eg. active site if enzyme...substrate no longer complementary and can not bind to active site to form an enzyme-substrate complex

Base deletion

- One nucleotide / base removed from DNA sequence
- Changes triplet / codon sequence from the point of mutation **(frameshift)**
- Changes sequence of codons on mRNA after point of mutation
- Changes sequence of amino acids in primary structure of polypeptide
- Changes position of hydrogen / ionic / disulphide bonds in tertiary structure of protein
- Changes tertiary structure / shape of protein i.e. non-functional or new and superior (converse for insertion, but similar effect = frameshift)



Base substitution

- Nucleotide / base in DNA replaced with another nucleotide / base
- Change in one base → changes one triplet

1. Changes one mRNA codon and one amino acid → sequence of amino acids in primary structure of polypeptide changes etc.

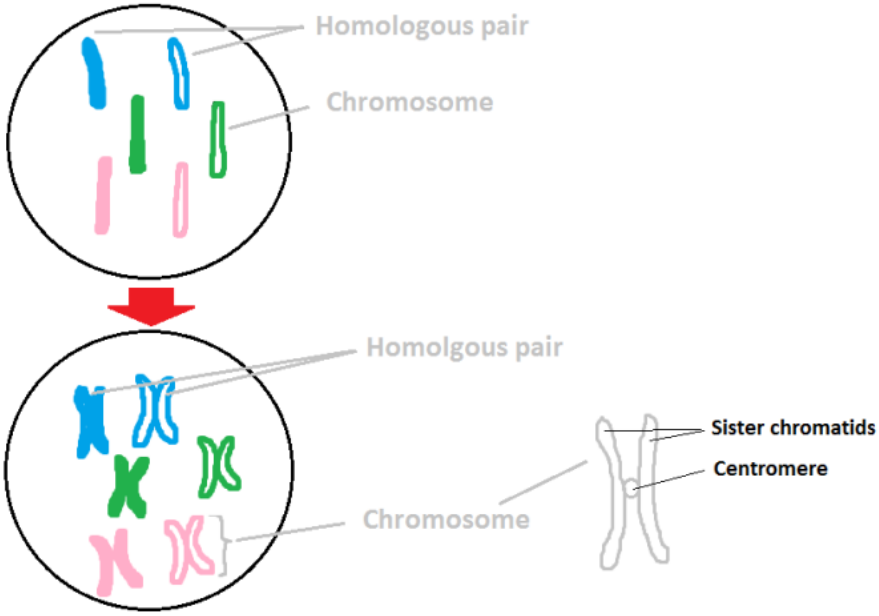
OR

2. Due to the degenerate nature of the genetic code, the new triplet may still code for the same amino acid so the sequence f amino acids in the primary structure of the polypeptide remains unchanged

Meiosis

- Meiosis is a special type of **cell division** that produces **gametes** (haploid cells for sexual reproduction).
- In humans meiosis only occurs in the ovary and the testis to produce eggs or sperm.
- Chromosome number halves (2n → n)
- OÒÏÄÕÃÉÎÇ ÈÁÐÌÏÉÄ ÇÁ Í ÅÔÅÓ ÅÎÓÕÒÅÓ ÄÉÐÌÏÉÄ ÎÕ Í ÂÅÒ ÉÓ ÒÅÓÔÏÒÅÄ ÁÔ ÆÅÒÔÉÌÉÓÁÔÉÏÎ ÁÎÄ ÍÁÉÎÔÁÉÎÓ ÃÈÒÏÍ Í ÏÏÍ Í Å ÎÕ Í ÂÅÒ ÆÒÏÍ Í ÏÎÅ ÇÅÎÅÒÁÔÉÏÎ ÔÏ ÔÈÅ ÎÅØÔ

- Before any cell division (mitosis or meiosis) all the DNA in the nucleus is replicated. Each chromosome has two identical copies, called **sister chromatids**, joined by a **centromere**



Meiosis involves two separate divisions (P1,M1, A1, T1 then P2, M2, A2, T2)

- Meiosis I (first division) = **SPLIT THE HOMOLOGOUS PAIRS**
  - Chromosomes arrange into homologous pairs
  - Crossing over (prophase I and metaphase I) creates genetic variation in gametes
  - Independent segregation (metaphase I) increases genetic variation in gamete

- Meiosis II (second division) = **SPLIT THE CHROMATIDS**

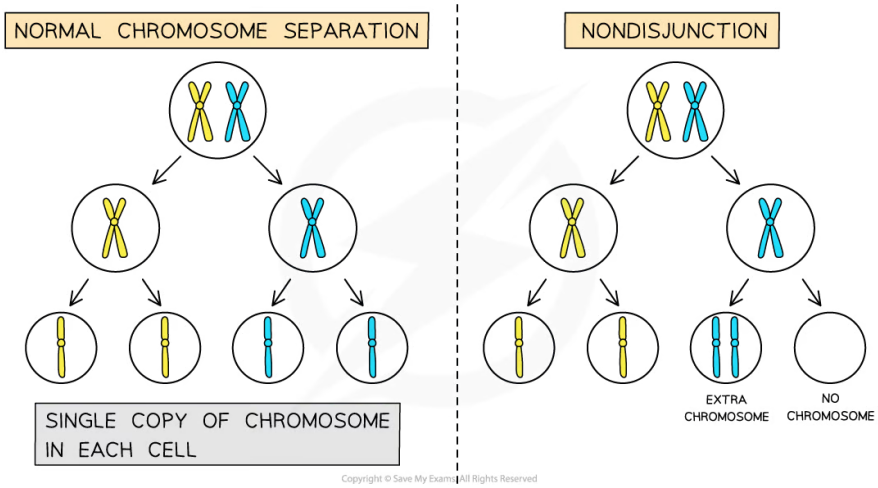
Meiosis creates 4 haploid cells (from a single diploid parent cell) that are genetically varied

How meiosis creates genetic variation

- **Crossing over** between homologous chromosomes
  - Alleles exchanged between non-sister chromatids of homologous chromosomes
  - Creates new combinations of maternal and paternal alleles on chromosomes
- **Independent segregation** of homologous chromosomes
  - Random alignment of homologous pairs at equator → random which chromosome from each pair goes to each daughter cell
  - Creates different combinations of maternal and paternal chromosomes and alleles in daughter cells
- **Random fertilisation** when two gametes fuse to form a zygote

Mutations in the number of chromosomes

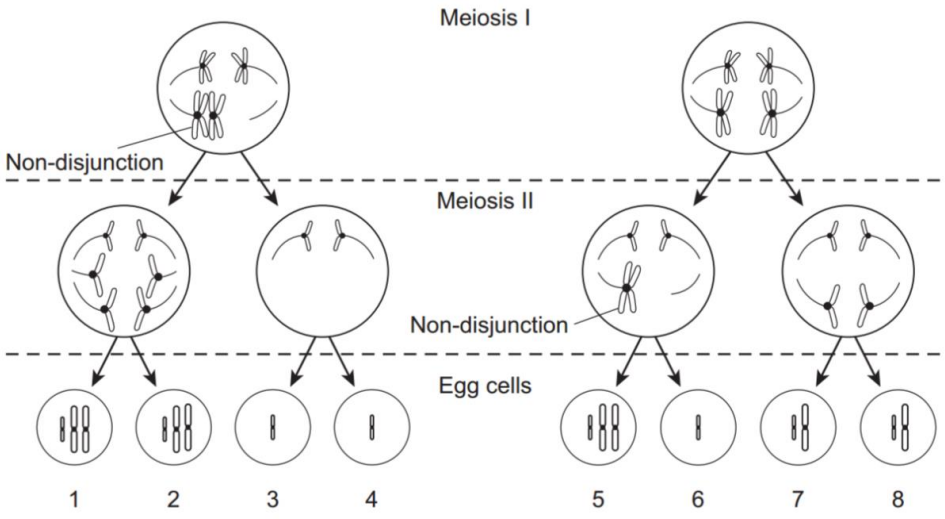
- Homologous chromosomes fail to separate during meiosis I OR sister chromatids fail to separate during meiosis II = **non-disjunction**
- One gamete has an extra copy of this chromosome and the other has none
- Upon fertilisation, zygote has one fewer or one extra chromosome
- Arises spontaneously
- Cause of some genetic diseases e.g. down's syndrome in humans – extra copy of chromosome 21



Example Exam Question

In humans, non-disjunction involving chromosome 21 can lead to a child being born with Down's syndrome. Figure 2 shows two ways in which non-disjunction can take place during meiosis.

Figure 2



(a) **Use Figure 2 to explain what is meant by non-disjunction. (2 marks)**

- ✓ Failure of chromosome (pairs) to separate
- ✓ During anaphase 1
- ✓ Failure of chromatids to separate
- ✓ During anaphase 2
- ✓ (So) both chromosomes/ chromatids go to one pole of spindle
- ✓ incorrect final chromosome number

(b) More than one of the egg cells, numbered 1 to 8, could produce zygotes that would develop into a child with Down's syndrome. **Give the numbers of these cells and explain the reason for your choices. (3 marks)**

- ✓ 1, 2 and 5
- ✓ They contain an extra chromosome (homologous pair still present after meiosis)
- ✓ At fertilisation, gives produces zygote with 3 copies of chromosome (21)/47 chromosomes in total

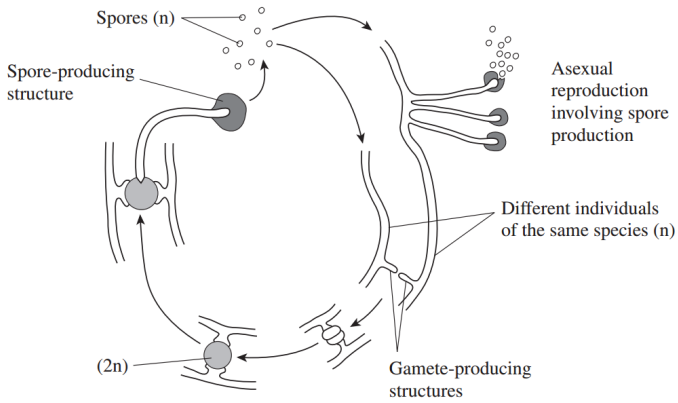
Comparing mitosis and meiosis

- **Mitosis produces diploid cells *whereas* meiosis produces haploid cells**
  - *Two divisions in meiosis whereby homologous chromosomes separate then chromatids separate, whereas one division in mitosis whereby only sister chromatids separate*
- **Daughter cells genetically identical to each other and parent cell in mitosis *whereas* in meiosis, daughter cells are genetically varied**
  - *Crossing over and independent segregation during meiosis I whereas no crossing over in mitosis*
- **Mitosis produces 2 daughter cells *whereas* meiosis produces 4 daughter cells**
  - *Two divisions in meiosis whereas only one division in mitosis*

You need to be able to recognise where meiosis occurs when given information about an unfamiliar life cycle

Example Exam Question

The diagram shows the life cycle of a fungus



(a) **On the diagram mark with (i) the letter F where fertilisation takes place (ii) the letter M where meiosis takes place. (2 marks)**

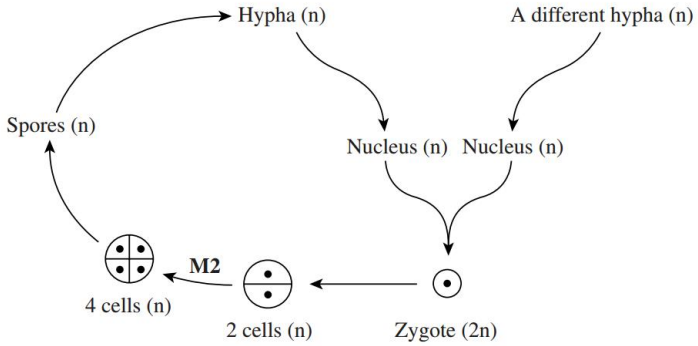
- ✓ M between 2n and spores
- ✓ F between gamete producing structure and zygote

(b) When large amounts of nutrients are present, the fungus reproduces asexually. **Explain one advantage of this. (2 marks)**

- ✓ Many spores released nearby
- ✓ So more of nutrient sourced used / more rapidly used
- ✓ OR produces genetically identical fungi
- ✓ So all well suited to nutrient source / environment

Example Exam Question

The diagram shows the life cycle of a fungus that reproduces sexually



(a) **Explain why the arrow labelled M2 represents the second division of meiosis (2 marks)**

- ✓ Division of zygote / previous stage chromosome number halved / 2n to n (so must be first stage of meiosis)
- ✓ In second sage separation of chromatids occurs (so chromosome number remains constant)/(2 haploid cells go to) 4 haploid cells produced

(b) The spores vary genetically. **Describe two ways by which this variation is produced (2 marks)**

- ✓ Independent segregation metaphase 1 of meiosis
- ✓ Crossing over during prophase1/metaphase 1 of meiosis
- ✓ (random) mutation / change in base sequence of DNA