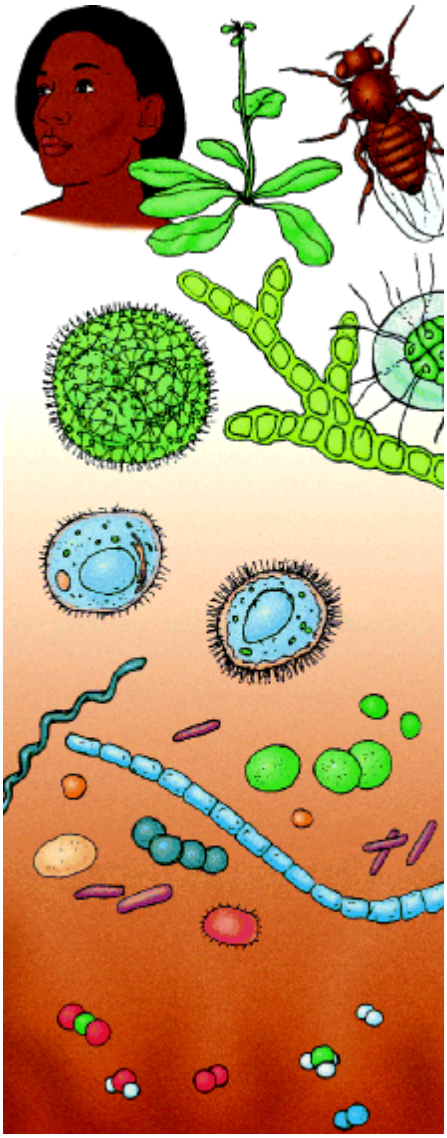
Introduction to Biology

Code: SCI1009



2017/2018

Period 1

Contents

General Information

Introduction, aim and approach

Biology, the science of life, studies the organisms as the basic units of life. How they are evolved, how they are build up, how they act, how they communicate with each other, how they are related to the non-living environment, and how they reproduce. At the start of the academic year the UCM offers students with a direct interest in life sciences, but also those who did not have the opportunity to study biology so far, an interesting course dealing with some of the abovementioned aspects of life: "Introduction to Biology". The central theme will be to gain more insight in the biology of organisms, focusing on different levels of life ranging from the life cycles and reproduction of organisms to the structure and function of the (macro)molecules important in life. The purpose of this course is to increase your appreciation and knowledge of the science of life and, moreover, provide you with a good basic knowledge required to enter more detailed courses in life sciences in the further curriculum, including Cell Biology, Genetics and Evolution, Biochemistry, Human Physiology, Microbiology, Immunology, Pathobiology and Disease, and Metabolism, Nutrition and Exercise.

The course starts with providing insight in how organisms are evolved and nowadays are subdivided. Because organisms are composed of cells, the basic unity of all life forms, the basic structure and function of cells and its organelles are discussed, including the biomolecules essential for life and the generation of these molecules in the cell. This is particularly important, since the causes of many diseases lie within changes in the structure and function of these molecules. As organisms grow and reproduce, the course further focuses on these processes, dealing with topics as basic genetics and the principles of cell growth and differentiation, metabolism and reproduction.

The course consists of 11 main themes that are presented to you in the form of two weekly lectures of one hour each and two tutorial group meetings per week. By means of a presentation the themes and/or additional subjects will be further deepened.

Readings

The main book used in this course is Life: The Science of Biology by Sadava *et al.* (10^{th} edition, but earlier editions can also be used). In addition to the book, other texts are offered. Each task contains a list of recommended readings and additional readings.

As the majority of the literature consists of chapters from books, you may sometimes have the feeling that you lack context, or that names crop up that you have not seen before. Go to any good introduction or handbook on the topic and look these up, if it impedes understanding of the argument. As a last resort you can always try and ask the members of your tutorial group.

Attendance requirement

The attendance requirement for the twelve tutorial meetings is 85%. You are allowed to miss two regular tutorials without further consequences, providing that you have a valid reason. If you miss more meetings, you will have to apply for an additional assignment (request forms are available at the Office of Student Affairs). In order to qualify for an additional assignment you have to have valid reasons for all missed sessions. If you don't meet the attendance requirement, you are not eligible for a resit. You automatically fail the course if you miss over 30% of the scheduled meetings.

Presence at the meeting in which the final presentations take place is mandatory (i.e. 100% attendance requirement).

Assessment

This course contains two elements of assessment:

1. a final exam consisting of open questions (70% of the overall course grade);
2. a presentation (30% of the overall course grade) in groups – for more information, see below.

Exam

The 11 topics discussed in this course will be covered in the exam. Although the attendance for lectures is not mandatory, the content adds to the pre- and post-discussions in the tutorial group meetings. Therefore, the content of the lectures is considered part of the examination.

In addition to the summative elements of assessment, a formative midterm test consisting of multiple choice questions is offered via the Student Portal. This test is not graded and thus does not count towards the final grade. The midterm test will be available via the Student Portal during a fixed timeslot of which you will be clearly informed during the course. The midterm test allows you to check your progress in the course. You will receive immediate feedback after finishing the test.

Presentation

The presentation should take a minimum of 15 minutes and a maximum of 20 minutes, including questions and group discussion. A laptop and beamer will be available.

Outline of the presentation

The presentation should contain at least the following items:

- Introduction
  - Introduction of research question/hypothesis, ± 2 min
  - Explanation of context and relevance, ± 1 min
- State of the art
  - Overview of current research and findings as presented in the literature, ± 5 min
- Conclusion and discussion
  - Conclusion: answer to the research question/hypothesis, ± 2 min
  - Discussion: possible limitations, remaining issues, other interesting points that need to be addressed, ± 2 min
- Group discussion (presenters lead the discussion), +/- 3 min

Assessment of the presentation

The assessment of the presentation will be based on:

- The quality of the introduction (are you able to explain the context and relevance of your research question in such a way that the audience has enough insight to understand the context and reasons for your topic);
- The quality of the presentation of the problem (are you able to explain the problem and capable of explaining why the solutions you provide are the best solutions);
- The handling of questions/discussion (are you able to answer questions and discuss the problem).

The presentation should be theoretically informed. You do not have to explain basic concepts discussed in the course, since you are presenting to students from your own discipline. However, don't take all background knowledge for granted!

The presentation is the result of a cooperation of a group of students – depending on the number of students in the course, the size of the groups may vary between 2 to 6 students. Keep in mind that you will receive a group grade for this assignment. Your tutor will assess the final product; s/he cannot evaluate whether you divided the work equally. It is your own responsibility to assure each team member invests approximately the same amount of work.

Resit

Students who initially fail the course, but who have complied with the compulsory attendance requirement and took part in all of the assessment during the course, are eligible for one resit.

The overall grade for this course consists of a grade for the final exam (70%), and the presentation (30%). The resit serves the purpose of lifting your overall grade to sufficient/above 5.5. It does not replace the overall grade. The resit can be either on the final exam, OR on the presentation. In case of a resit, you will have to redo that part of the assessment for which you received the lowest grade. The resit grade will replace the original grade for that particular element of assessment and will thus count towards 70% (in case of the exam) or 30% (in case of the presentation) of the final grade. In case of redoing the presentation, you have to choose a new topic and give an individual presentation.

In order to receive a grade for the exam you will have to do a serious attempt at passing the exam. If it is not deemed a serious attempt you will not receive a grade and you will not qualify for a resit. The same thing holds for the presentation. If it is not deemed a serious attempt you will not receive a grade and you will not qualify for a resit.

Course coordinators

Questions or issues regarding the course content and e.g. assessment should first be discussed with the tutorial group and tutor. Those questions can also be discussed with the coordinators, but preferably during or directly after the lectures. Please note that the coordinators will not reply to individual e-mails that could have been dealt with during tutorial group meetings or during lectures.

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Overview of meetings, lectures and deadlines

Week	Meeting	Post-discussion	Pre-discussion	Deadlines
1	*Intro lecture*			
	Tutorial group meeting 1		Task 1	
	Tutorial group meeting 2	Task 1	Task 2	
2	*Chemistry: Small molecules* *Chemistry: Molecules of Life*			
	Tutorial group meeting 3	Task 2	Task 3	
	Tutorial group meeting 4	Task 3	Task 4	
3	*The Cell* *Metabolism*			*Distribution of list with possible subjects for presentations*
	Tutorial group meeting 5	Task 4	Task 5	
	Tutorial group meeting 6	Task 5	Task 6	
4	*Introduction to genetics* *DNA-RNA-protein*			*Formative midterm test available on EleUM*
	Tutorial group meeting 7	Task 6	Task 7	Subject declaration
	Tutorial group meeting 8	Task 7	Task 8	
5	*Cell cycle and DNA replication* *Cell Signalling and Communication*			
	Tutorial group meeting 9	Task 8	Task 9	
	Tutorial group meeting 10	Task 9	Task 10	
6	*Defence systems* *Human Physiology*			
	Tutorial group meeting 11	Task 10	Task 11	
	Tutorial group meeting 12	Task 11		
7	Final exam			
	Final presentations			

Please check the schedule provided by the Office of Student Affairs and announcements on the Student Portal as well.

Tasks

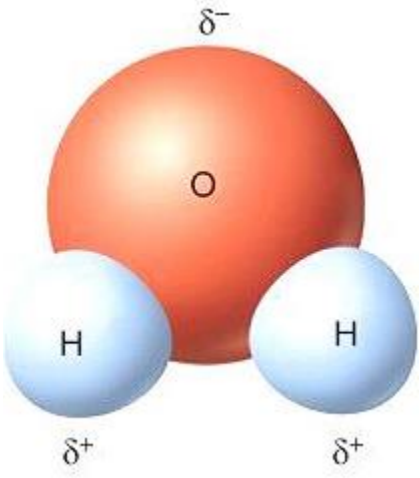
Task 1. The chemistry of life: how important are water and small molecules?

"You are what you eat" is a famous saying about body chemistry. All living things, even those in the most remote and harmful environments (such as hot springs, beneath frozen Antarctic ice, several miles below sea level, acid and salty environments) are composed of the same kind of atoms, and surprisingly of the same elements as the vast non-living portion of the universe. The relative amounts of these atoms eventually combine to form molecules, from simple ones to super-complexes. Each atom has its own specific chemical and physical characteristics which allow them to undergo interactions with the same and/or other atoms resulting in a huge amount of different molecules that support life. An interesting question is how many and which atoms are hereby involved?

Among the many events in cellular life is the multitude of specific chemical transformations and changing partnerships, which provide the cell with usable energy and the molecules needed to form its structure and coordinate its activities. All the biochemical reactions and cellular processes are steered and controlled by basic principles of chemistry.

In general, around 75-80% of living matter by weight is formed by water, inorganic ions and a large array of relatively small organic molecules. Water is the most abundant. Life without water cannot exist. Why is this? Have a look at the structure of water and try to deduce specific features that make this molecule so important for life.

As we will see, the internal environment of a cell is an aqueous solution in which a lot of "things" are solubilised or "seemingly float" around, but always in a very structured order. But not all regions or compartments of a cell and even not all tissues and organs in a multicellular body do have the same features; some are neutral, other are more acidic or basic. What is it that determines this feature and why is it so important to biology?



Recommended Literature

Sadava *et al.*, Life, the science of biology, 8th ed., 2007: Chapter 2

Sadava *et al.*, Life, the science of biology, 9th ed., 2010: Chapter 2

Sadava *et al.*, Life, the science of biology, 10th ed., 2014: Chapter 2

Additional readings

Alberts *et al.*, Essential cell biology, 4th edition, 2014: Chapter 2

Campbell and Reece, Biology, 6th edition, 2002: Chapter 2 & 3 & 4

Task 2: The chemistry of life: from small to big molecules.

Besides the small molecules that you have learned about in the previous task, the cell and its subcellular structures are assembled from large molecules, also known as macromolecules or polymers. These macromolecules are composed of smaller units each with their own properties. Although the amount and diversity of these monomers is quite limited, the diversity of polymers that can be built with these monomers is tremendous. Nevertheless, only correct constructed macromolecules will be functional inside the cell. Each macromolecule will be unique and has its own characteristics and three-dimensional shape. These will be determined by the type of the monomer, the eventual sequence of monomers in a polymer and the interactions between them and with the environment.

Generally, there are four main classes of large biological molecules that form the make-up of living organisms. A unique feature of these four classes of macromolecules is that they are made in a similar way in all living things and are present in roughly the same proportions in all organisms. Because of their specific chemical structure, the cell utilizes each class of polymers for different functions, such as the storage of energy, the storage of genetic information, to catalyse chemical reactions, and for communication. In addition, macromolecules can interact with each other in different ways. Finally, it is important to realize where (at what site) these families of macromolecules are found in the cell and why they are present at those specific sites.

Recommended Literature

Sadava *et al.*, Life, the science of biology, 8th ed., 2007: Chapter 3

Sadava *et al.*, Life, the science of biology, 9th ed., 2010: Chapter 3 & 4

Sadava *et al.*, Life, the science of biology, 10th ed., 2014: Chapter 3 & 4

Additional readings

Alberts *et al.*, Essential cell biology, 4th edition, 2014: Chapter 2 & 4

Campbell and Reece, Biology, 6th edition, 2002: Chapter 5

Task 3. The cell, the basic unit of life

Nowadays, it seems very straightforward that all organisms on earth are composed of cells. However, it still is sometimes difficult to believe that huge mammals (including humans), but also plants as well as for example bacteria's, share many cellular structures and functions. Indeed, it took quite some time in history before two scientists in 1938 proposed one of the fundamental principles of biology, i.e. the cell theory.

One of the earliest scientific investigators who opened our eyes to the world of cells was the 17th century British mathematician Robert Hooke. Using a relatively crude microscope, he looked at everything he could lay a hand on. And in contrast to throwing away the cork of again another empty bottle of wine, as we often do, he sliced pieces of cork and analysed them with his microscope (Figure 1). He observed networks of tiny, boxlike compartments, such as one sees in honeycombs. He called these compartments "cellulae", which we now know as cells. Hooke didn't really see cells, but rather the structures that surround plant cells, because the cellular constituents had disappeared. Particularly by improving the microscopes, e.g. by the excellent work of Antonie van Leeuwenhoek in the 1670s (Figure 2), to the level they nowadays have, more and more cellular secrets have been explored over the past eras. We now know, for example, that there are structural resemblances and differences between prokaryotic cells (e.g. bacteria's) and eukaryotic cells (e.g. mammals), but also between animal cells and plant cells, and that these differences also reflect their function in the different organisms. As an example, schematic drawings of two cells are depicted on page 14 (Figure 3), which may help you in the search for these structures and their basic functions.

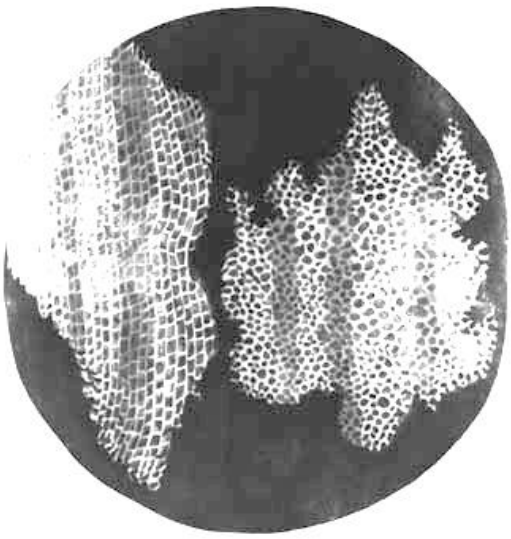


Figure 1 "Cellulae" in cork as observed by Robert Hooke in the 1660s

Figure 2 A sperm cell as seen by Nicolaas Hartsoeker, using a microscope by Antonie van Leeuwenhoek in the 1670s.

Recommended literature

Sadava et al., Life, the science of biology, 8th ed., 2007: Chapter 4 & 5

Sadava et al., Life, the science of biology, 9th ed., 2010: Chapter 5 & 6

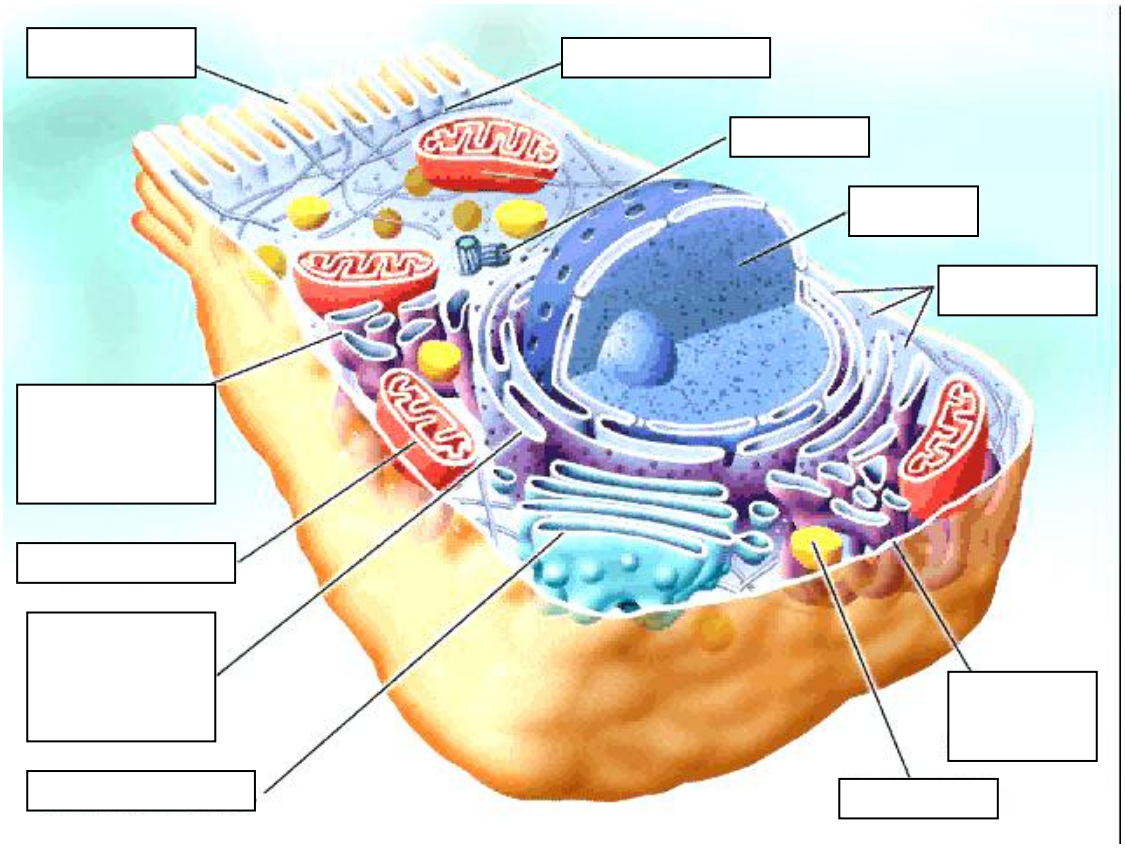
Sadava et al., Life, the science of biology, 10th ed., 2014: Chapter 5 & 6

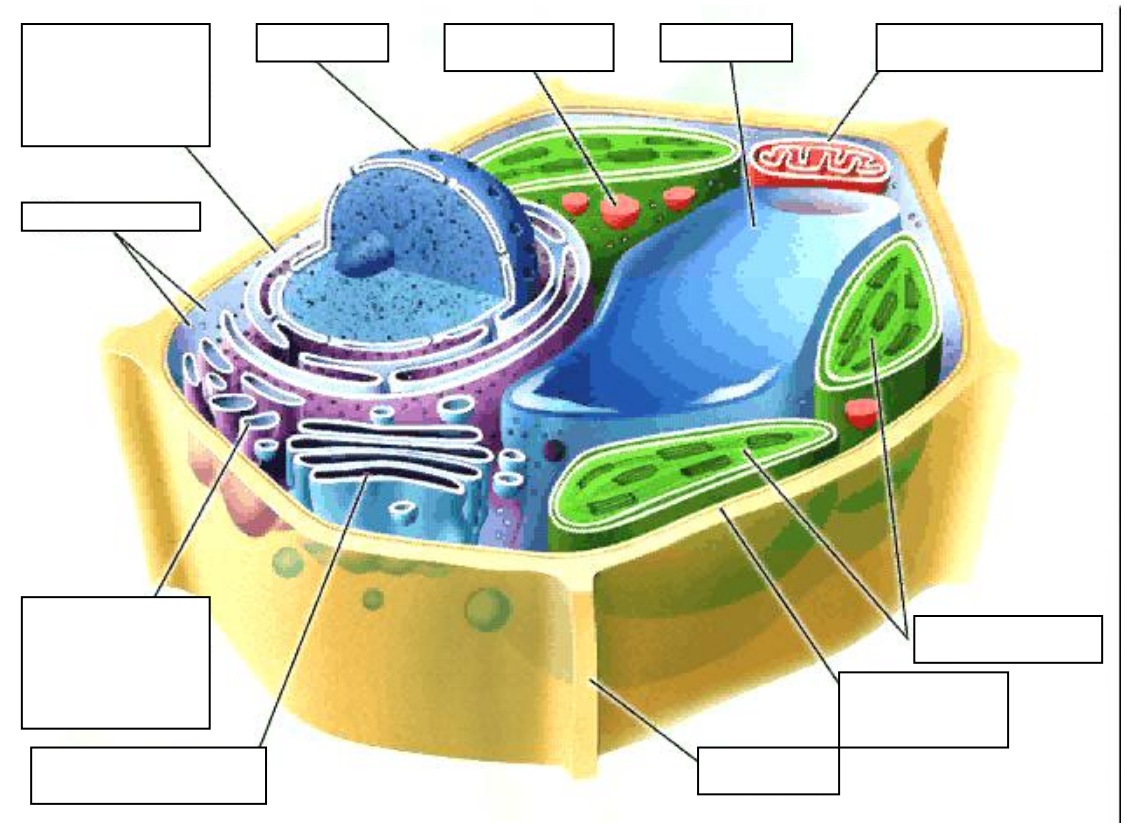
Additional readings

Alberts *et al.*, Essential cell biology, 4th edition, 2014: Chapter 11 & 15

Alberts *et al.*, Molecular biology of the cell, 6th edition, 2015: Chapter 10 & 12

Campbell and Reece, Biology, 6th edition, 2002: Chapter 7 & 8





Task 4. Tired muscles, cheese and wine

Organisms require fuel to live, which must be obtained from foods. Plants make their own fuel through photosynthesis, whereas humans obtain fuel by eating other organisms. In order to perform exercise, energy is required, which is provided exclusively in the form of ATP. Several mechanisms are involved in the synthesis and breakdown of this biologically important molecule in human cells. Sugars and oxygen play an important role in these processes.

During heavy exercise, such as weight lifting and running, muscle cells need to generate a lot of energy. After a while this causes muscle fatigue and soreness, which is the end result of a rather inefficient metabolic process that has occurred in the muscle cells. Like tired muscles, cheese and wine are also products of this metabolic process.

Recommended Literature

Sadava *et al.*, Life, the science of biology, 8th ed., 2007: Chapter 6 & 7

Sadava *et al.*, Life, the science of biology, 9th ed., 2010: Chapter 8 & 9

Sadava *et al.*, Life, the science of biology, 10th ed., 2014: Chapter 8 & 9

Additional reading

Alberts *et al.*, Essential cell biology, 4th edition, 2014: Chapter 13 & 14

Alberts *et al.*, Molecular biology of the cell, 6th edition, 2015: Chapter 14

Campbell and Reece, Biology, 6th edition, 2002: Chapter 9

Task 5. It is all in our genes!

The classical model

The Austrian monk Gregor Mendel played a pivotal role in our understanding of genetics. He spent more than six years working out the principles of inheritance in plants. He made crosses with hundreds of plants and meticulously noted the characteristics of the progeny. He concluded that every cell has two copies of a unit of inheritance (we now call 'gene'). Based on his experiments Mendel postulated a series of laws. The first law states that the two copies of a gene segregate. The second law states that copies of different genes assort independently. Finally the third law states that some genes are dominant and others are recessive. His work was largely ignored at the time he published his results, but gained prominence only years later when the process of meiosis (next task) was described.

Even today the Mendelian laws of inheritance are used in modern medicine. Now we know that a gene is a unit of inheritance, which is responsible for a clearly described phenotypic feature, and inherits according to the Mendelian law (see figure). A gene consists of two alleles that will be separated during the formation of gametes. As a result only one allele of a gene will be present per gamete. During the process of fertilization both alleles re-join.

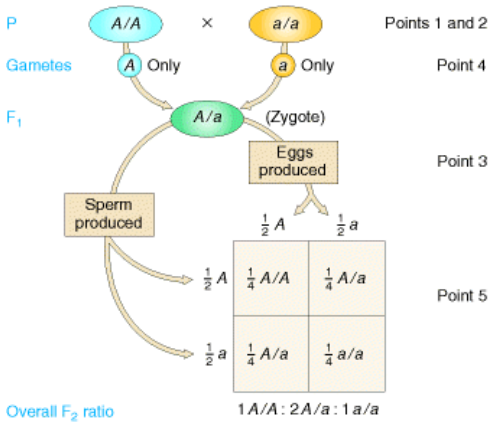
The following examples show how Mendelian laws of inheritance can be used in a clinical setting:

1. A geneticist is asked to determine if two children have probably been exchanged at birth. Examination of the ABO and Rhesus blood group systems led to the results below. The geneticist makes a scheme out of this *(see table on the right)*.

Child X:	B, Rh-
Child Y:	A, Rh+
Father 1:	A, Rh-
Mother 1:	B, Rh+
Father 2:	B, Rh+
Mother 2:	AB, Rh-

2. In case the above investigation is not sufficient to elucidate the problem, one may question whether there are any other systems that can be utilized to do so. One option would be to make use of the HLA gene system, which codes for histocompatibility (tissue or transplantation) antigens. This gene system is very polymorphic, because it consists of four closely linked loci (A – D) that all contain a high number of different alleles. Therefore there is only very little chance that two individuals within a population are identical with respect to their HLA alleles. The individuals in example 1 harboured the HLA combinations shown below. It is now possible to determine which alleles the children received from their parents.

Child X:	A2, A24, B8, B44, C4, D5, D6
Child Y:	A2, A24, B7, B44, C4, C6, D1, D6
Father 1:	A2, A28, B35, B44, C4, C6, D6
Mother 1:	A2, A24, B7, B44, C4, C6, D1, D5
Father 2:	A2, A11, B8, B44, C2, C4, D5, D6
Mother 2:	A24, B8, B14, C4, C6, D3, D6



Mendel's model of the hereditary determinants of a character difference in the P, F_1, and F_2 generations

Recommended Literature

Sadava *et al.*, Life, the science of biology, 8th ed., 2007: Chapter 10

Sadava *et al.*, Life, the science of biology, 9th ed., 2010: Chapter 12

Sadava *et al.*, Life, the science of biology, 10th ed., 2014: Chapter 12

Additional reading

Campbell and Reece, Biology, 6th edition, 2002: Chapter 14 & 19

Task 6. Omnis cellula e cellula *(Every cell is derived from another cell)*

The work of Mendel gained importance only years after its original publication in 1866. It was the German biologist August Weismann who appreciated the significance of meiosis for reproduction and inheritance in 1890. He noted that two cell divisions were necessary to transform one diploid cell into four haploid cells if the number of chromosomes had to be maintained.

Meiosis occurs in eukaryotic life cycles involving sexual reproduction, consisting of the constant cyclical process of meiosis and fertilization. This takes place alongside normal mitotic cell division. In multicellular organisms, there is an intermediary step between the diploid and haploid transition in which the organism grows. At certain stages of the life cycle, germ cells produce gametes.

Somatic cells make up the body of the organism and are not involved in gamete production. They can divide and form to daughter cells. In multicellular organisms this cell division plays an important role in the growth and repair of tissues. In contrast to meiosis, mitosis is a part of the cell cycle process by which chromosomes in a cell nucleus are separated into two identical sets of chromosomes, each in its own nucleus. In general, mitosis (division of the nucleus) is often followed by cytokinesis, which divides the cytoplasm, organelles and cell membrane into two new cells containing roughly equal shares of these cellular components

Although there are lots of different kind of organisms on earth that have their own life cycle, these organisms all have the urge to propagate (reproduce themselves) as this is the driving force in life (and therefore also in evolution). Propagation or reproduction (sexual or asexual) is in essence the production of identical entities and organisms use cell division to achieve this goal.

Recommended Literature

Sadava *et al.*, Life, the science of biology, 8th ed., 2007: Chapter 9

Sadava *et al.*, Life, the science of biology, 9th ed., 2010: Chapter 11

Sadava *et al.*, Life, the science of biology, 10th ed., 2014: Chapter 11

Additional reading

Alberts *et al.*, Essential cell biology, 4th edition, 2014: Chapter 18 & 19

Alberts *et al.*, Molecular biology of the cell, 6th edition, 2015: Chapter 17

Campbell and Reece, Biology, 6th edition, 2002: Chapter 12 & 13

Task 7. "DNA has memory!" *(Isaiah Washington)*

The 21st century model

In 1953 Watson and Crick aided by the work of Rosalind Franklin discovered the (three dimensional) structure of DNA. This discovery led to the 21st century model of a gene as unit of inheritance. Our genetic information is stored in our DNA and a gene is a piece of a double-stranded DNA molecule, which codes for a protein chain via messenger RNA (next task).

As a result of the (almost) complete DNA sequencing of the human genome we now know that the diploid human genome consists of 1) $3x10^9$ base pairs (length: 1 meter), 2) contains about 30,000 genes and 3) is divided over 46 chromosomes. Each human cell contains two copies of each of these chromosomes, except for the germ cells that harbour only one copy of all chromosomes. If one assumes that the mean length of a gene is approximately 10,000 base pairs, it becomes clear that only $3x10^8$ base pairs (10% of the human genome) are harbouring the human genes, and thus the remaining DNA is not coding for essential proteins.

The specific structure of the DNA ensures that during DNA replication the genetic information is faithfully copied. After melting the complementary strands of DNA the single stranded chains serve as template for the synthesis of a new strand. Specific enzymes in the cell catalyse DNA replication. However these enzymes are capable to synthesize DNA in one direction only.

Recommended Literature

Sadava *et al.*, Life, the science of biology, 8th ed., 2007: Chapter 11

Sadava *et al.*, Life, the science of biology, 9th ed., 2010: Chapter 13

Sadava *et al.*, Life, the science of biology, 10th ed., 2014: Chapter 13

Additional reading

Alberts *et al.*, Essential cell biology, 4th edition, 2014: Chapter 5 & 6

Alberts *et al.*, Molecular biology of the cell, 6th edition, 2015: Chapter 4 & 5

Campbell and Reece, Biology, 6th edition, 2002: Chapter 16

Task 8. The central dogma of molecular biology *(DNA-RNA-protein)*

DNA > RNA > protein

The genetic information present in the cell nucleus is required for e.g. the synthesis of proteins in the cell. These proteins are essential for cell structure as well as for the processes occurring within the cell. Protein synthesis takes place in two steps:

1) the transcription (copying) of genetic information from DNA to messenger RNA (mRNA), which takes place in the nucleus.
2) the translation of the mRNA, which takes place in the cytoplasm with the help of ribosomes.

Besides mRNA, two other types of RNA are involved in protein synthesis, i.e. ribosomal RNA (rRNA) and transfer RNA (tRNA). The rRNA is part of the ribosome that together with a number of structural proteins forms a kind of scaffold for protein synthesis. The tRNA transfers the protein building blocks (amino acids) to the right site in the growing protein chain; each of the 20 different types of amino acids, occurring in nature, has its own tRNA type. The information within the mRNA is coded in groups of three nucleotides (a triplet or code). Each code encodes a specific amino acid and is recognized by its complementary anti-code of a tRNA molecule that is bound to an amino acid. A large number of enzymes are involved in the process of protein synthesis. Although we all look alike, we humans do not share an identical genome. Small deviations in our genotype determine the phenotype of an individual.

What follows is an example of how small differences in mRNA can have great consequences for life.

Haemoglobin and its variants

Haemoglobin, the red colour in erythrocytes, which is responsible for oxygen transport in the blood, consists of 4 peptide chains: 2 α-chains (2 x 141 amino acids) and 2 β-chains (2 x 146 amino acids), connected with the so-called "haem" group. The α- and β-chains are encoded by the globin gene clusters present on chromosomes 16 and 11, respectively. The last 25 years hundreds of haemoglobin variants has been discovered and biochemically characterized (by means of electrophoresis and determination of the sequence of amino acids). Some Hb-variants were so much changed in structure, that normal oxygen transport was hampered and/or aggregation could occur. As a consequence, the shape and function of red blood cells may change, which may result in haemolysis and anaemia, such as sickle cell anaemia and the various forms of thalassaemia. Modern technologies in molecular biology and genetics (including recombinant DNA techniques) are available to study the gene mutations that underlie the development of these haemoglobin variants. In this way, the normal biology, gene mutations and the molecular pathology of such gene clusters can be studied in detail. By using this model system, the example below can provide insight in the effect of different gene mutation on the expression of mRNA and protein.

The haemoglobin of a patient with sickle cell anaemia (Hb-S) differs at only one single amino acid when compared with normal haemoglobin (Hb-A). In the β-chain of Hb-S the 6th triplet (GAG) is mutated in GTG. The normal β-chain of Hb-A is:

	1	2	3	4	5	6	7	amino acid
NH_2 –	VAL –	HIS –	LEU –	THR –	PRO –	GLU –	GLU	protein
	GTG –	CAC –	CTG –	ACT –	CCT –	GAG –	GAG	DNA

Which amino acid is encoded by the mutated triplet GTG?

There are more β-chain variants known that are due to a point mutation in the 6th triplet, e.g.:

The C (child) variant: GLU-6 to LYS

The Machida variant: GLU-6 to GLN

Which base mutations have occurred in these cases? What is the consequence of these changes in amino acid with respect to haemoglobin function (consider e.g. the charge and three-dimensional structure of the molecule)?

We will now focus on the carboxy-terminal part of the α-globin. Base mutations at this site will lead to dramatic changes in the haemoglobulin. Two examples are the Icaria- and the Wayne variant.

Normal HbA:

137	138	139	140	141		amino acid
THR –	SER –	LYS –	TYR –	ARG –	terminus	protein
ACA –	UCU –	AAA –	UAC –	CGU		mRNA

	137	138	139	140	141	142	143	144	amino acid
Icaria variant	THR –	SER –	LYS –	TYR –	ARG –	LYS –	ALA –	GLU	protein
Wayne variant	THR –	SER –	ASN –	THR –	VAL –	LYS –	LEU –	…	protein

Which mutations have occurred to explain the above protein products?

Besides mutations on the DNA level, also on the level of the chromosome mutations can be found, such as translocations, inversions, deletions and duplications.

1st position (5' end)	2nd position U	2nd position C	2nd position A	2nd position G	3rd position (3' end)
U	Phe	Ser	Tyr	Cys	U
	Phe	Ser	Tyr	Cys	C
	Leu	Ser	STOP	STOP	A
	Leu	Ser	STOP	Trp	G
C	Leu	Pro	His	Arg	U
	Leu	Pro	His	Arg	C
	Leu	Pro	Gln	Arg	A
	Leu	Pro	Gln	Arg	G
A	Ile	Thr	Asn	Ser	U
	Ile	Thr	Asn	Ser	C
	Ile	Thr	Lys	Arg	A
	Met	Thr	Lys	Arg	G
G	Val	Ala	Asp	Gly	U
	Val	Ala	Asp	Gly	C
	Val	Ala	Glu	Gly	A
	Val	Ala	Glu	Gly	G

Recommended Literature

Sadava *et al.*, Life, the science of biology, 8th ed., 2007: Chapter 12

Sadava *et al.*, Life, the science of biology, 9th ed., 2010: Chapter 14

Sadava *et al.*, Life, the science of biology, 10th ed., 2014: Chapter 14

Additional reading

Alberts *et al.*, Essential cell biology, 4th edition, 2014: Chapter 7

Alberts *et al.*, Molecular biology of the cell, 6th edition, 2015: Chapter 6

Campbell and Reece, Biology, 6th edition, 2002: Chapter 17

Task 9. Cell signalling and communication

Multicellularity was so slow to evolve because it has been related to the difficulty of developing cell communication mechanisms that a multicellular organism needs. Its cells have to be able to communicate with one another in complex ways if they are able to govern their own behaviour for the benefit of the organism as a whole.

Individual cells, like multicellular organisms, needs to sense and respond to their environment. A typical free-living cell – even a primitive bacterium – must be able to track down nutrients, tell the difference between light and dark, and avoid poisons and predators. And if such a cell is to have any kind of "social life", it must be able to communicate with other cells.

In a multicellular organism, things are much more complicated. Cells must interpret the multitude of signals they receive from other cells to help coordinate their behaviour. During animal development, cells in the embryo exchange signals to determine which specialized role each cell will adopt, what position it will occupy in the animal, and whether it will survive, divide or die; later, a large variety of signals coordinate the animal's growth and its day-to-day physiology and behaviour. Later on in life, things can go completely wrong if the communication between the cells does not work properly.

These communication mechanisms depend heavily on extracellular signal molecules, which are produced by cells to signal to their neighbours or to cells further away. They also depend on elaborate systems of proteins that each cell contains to enable it to respond to a particular subset of these signals in a cell-specific way. These proteins include cell-surface receptor proteins, which bind the signal to appropriate parts of the cell. At the end of each intracellular signalling pathway are target proteins, which are altered when the pathway is active and change the behaviour of the cell. Depending on the signal's effect, these target proteins can be regulatory proteins, ion channels, components of a metabolic pathway, parts of the cytoskeleton, and so on.

Signalling is associated with receptors. Once a ligand has been bound to a receptor, several types of highly conserved intracellular signal transduction pathways are activated. Signal transduction pathways are grouped into several basic types, based on the sequence of the intracellular events. Classical players in these pathways are kinases which can be an intrinsic part of the receptor or be tightly bound to it. These kinases will activate a variety of signal transduction proteins including transcription factors, small GTP-ases, larger trimeric GTP binding proteins.

Recommended Literature

Sadava *et al.*, Life, the science of biology, 8th ed., 2007: Chapter 15

Sadava *et al.*, Life, the science of biology, 9th ed., 2010: Chapter 7

Sadava *et al.*, Life, the science of biology, 10th ed., 2014: Chapter 7

Additional reading

Alberts *et al.*, Essential cell biology, 4th edition, 2014: Chapter 16

Alberts *et al.*, Molecular biology of the cell, 6th edition, 2015: Chapter 15

Campbell and Reece, Biology, 6th edition, 2002: Chapter 11

Task 10. Fighting viruses

Winter is nearing, with its frequent problems of running noses, and discussions being interrupted by bursts of coughing and sneezing. But why is it that some seem to catch every cold virus that comes by, whereas others appear to be immune and suffer no consequences of a coughing and sneezing student in their tutorial group?

The immune system consists of physical barriers and molecular and cellular processes within an organism that protects against disease. If a few intruders happen to sneak by your biological sentry, that's OK. Your body deploys a host of additional immune cell forces, including antibodies, that are designed to hunt down the unwanted intruders and ultimately work to destroy them. But with more than 200 viruses that can cause the common cold, our bodies must be packed with defence forces! How else is the body able to recognise all these different intruders?

Recommended Literature

Sadava *et al.*, Life, the science of biology, 8th ed., 2007: Chapter 18

Sadava *et al.*, Life, the science of biology, 9th ed., 2010: Chapter 42

Sadava *et al.*, Life, the science of biology, 10th ed., 2014: Chapter 42

Additional reading

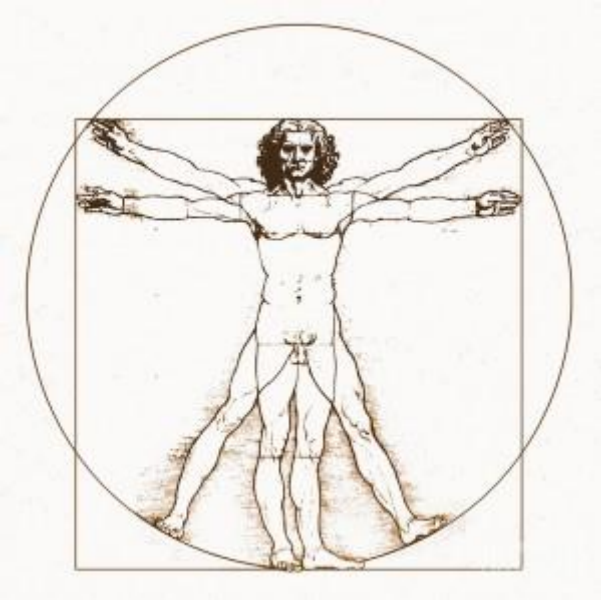
Alberts *et al.*, Molecular biology of the cell, 6th edition, 2015: Chapter 24

Campbell and Reece, Biology, 6th edition, 2002: Chapter 43

Task 11. Putting things into perspective

At the end of the six-week course, with the exam in near sight, a group of students decides it might be worthwhile to recap the topics discussed in the course. One of them suggests making it interesting by putting things into perspective. She suggests looking at the human body: "We have been discussing cells, including the way they divide and multiply and how they communicate. We have studied the molecules of life and how they interact. We know all about DNA and RNA and how they make proteins. I'm pretty sure this all has some relation to the human body. After all, we're not just a collection of individual cells!"

The others agree. They start looking into how different cells make up such a complex multicellular organism like the human being. They come across interesting information on how these different cells and tissues communicate. It is almost a small wonder how our body is able to regulate all its internal systems without us having to think about it, but in essence it is nothing more than a combination of what has been discussed in this course.



Recommended Literature

Sadava *et al.*, Life, the science of biology, 8th ed., 2007: Chapter 40-41

Sadava *et al.*, Life, the science of biology, 9th ed., 2010: Chapter 40-41

Sadava *et al.*, Life, the science of biology, 10th ed., 2014: Chapter 40-41

Additional reading

Campbell and Reece, Biology, 6th edition, 2002: Chapter 40