BLG-5070-2

Applied Genetics

Biology

Québec

BLG-5070-2 Applied Genetics

INTRODUCTION

The course entitled *Applied Genetics* is aimed at enabling adult learners to function effectively in situations from the *Research* and *Expertise* families that involve genetics, its applications and their impact.

In this course, adult learners seek answers to problems related to the molecular function of cells and to genetics, genetic diversity and the genetic process of evolution. They process data to solve problems related to genetic crossing, calculate the probability of hereditary diseases or interpret DNA tests. They apply their knowledge to illustrate the effects of a mutation in a cell's DNA sequence, understand the presence of a genetic disease or explain a species' ability to adapt to a change in its environment. They make informed decisions on social, ethical and environmental issues arising from the use of technologies in molecular biology, particularly gene cloning and transgenesis, and evaluate their impact on society and demographics. Lastly, they use various methods to communicate their ideas and the results of their scientific research on applied genetics.

By the end of this course, in *Research* and *Expertise* situations, adult learners will be able to:

- explain the mechanisms involved in the transmission of hereditary characteristics through genetic crossing based on Mendel's laws or sex-linked inheritance
- interpret information on allelic diversity within a population (e.g. ABO and Rh blood groups)
- interpret a family tree and human karyotypes to answer questions on heredity
- analyze an issue related to a genetic engineering application
- justify the decision of whether or not to produce genetically modified organisms (GMOs) and describe their uses and the risks they pose, taking into account biotechnical, social, ethical and environmental factors
- explain the usefulness of genetic diversity as opposed to the homogeneity of crops and livestock
- discuss ethical issues that may arise from genetic screening for hereditary diseases
- use biotechnical results (for example, to establish a genetic profile or to detect a genetic anomaly during a diagnostic test for hereditary diseases)
- use concrete cases to analyze certain mechanisms of microevolution and their impact on biodiversity or on the survival of a species

SUBJECT-SPECIFIC COMPETENCIES

The following table lists, for each competency, the key features studied in the course. The manifestations of the key features are presented in Appendix 4.

Competency 1 Seeks answers or solutions to problems involving biology	Competency 2 Makes the most of his/her knowledge of biology	Competency 3 Communicates ideas relating to questions involving biology, using the languages associated with science and technology
▪ Defines a problem ▪ Develops a plan of action ▪ Carries out the plan of action ▪ Analyzes his/her results	▪ Puts issues in context ▪ Analyzes a phenomenon or an application from a biological point of view ▪ Explains an issue from the standpoint of biology ▪ Forms an opinion about an issue	▪ Interprets scientific and technological messages ▪ Produces scientific and technological messages

PROCESSES

The investigative processes enable adult learners to examine biological issues, solve problems and study applications. The following are the steps in an investigative process:

- Define the problem
- Formulate a hypothesis
- Test the hypothesis
- Draw conclusions and communicate

The most appropriate investigative processes for this course are modelling, documentary research and the observation method. It is during hypothesis verification that these methods become distinguishable. Section 3.5 and Appendices 2 and 3 present these investigative processes with their respective characteristics.

CROSS-CURRICULAR COMPETENCIES

The cross-curricular competencies supplement the subject-specific competencies. The development of one contributes to the development of the others. Course BLG-5070-2 allows for all the cross-curricular competencies to be put into practice. The sample learning situations presented in this course place particular emphasis on those indicated in grey shading in the table below.

Cross-Curricular Competencies			
Intellectual	**Communication-Related**	**Personal and Social**	**Methodological**
Uses information	Communicates appropriately	Achieves his/her potential	Adopts effective work methods
Solves problems		Cooperates with others	Uses information and communications technologies
Exercises critical judgment			
Uses creativity			

SUBJECT-SPECIFIC CONTENT

A) KNOWLEDGE

The compulsory concepts represent the specific knowledge to be acquired in this course. They are presented in the tables in the following section.

1. Concepts

The knowledge written in italics have been acquired in the Science and Technology programs of the Québec Education Program and must be mobilized again in this course.

<table>
<tr><th colspan="2">The Living World</th></tr>
<tr><td colspan="2">General concept: Genetics</td></tr>
<tr><td colspan="2">Genetics is the study of the hereditary transmission of traits and genetic variation among individuals. Genes, the basic units of inheritance, are found at specific locations on chromosomes. Each gene in a eukaryotic cell has two alleles that undergo segregation and independent assortment during gamete formation.

In applying Mendel’s laws of inheritance, it is necessary to take into account an individual’s phenotype in order to determine the underlying genotype. The Punnett square is used to predict the outcome of controlled crosses. Lastly, the family tree represents unions and descendants over many generations for a given trait, and makes it possible to predict the risk of transmitting a hereditary disease.

For autosomal dominant diseases such as Huntington’s disease to manifest, only a single copy of the mutated gene is required. However, autosomal recessive diseases, such as cystic fibrosis, sickle cell anemia, β-thalassemia, lactic acidosis and phenylketonuria, only develop in homozygous individuals. Heterozygous individuals, who have only one mutated gene, will generally not be affected by these diseases. They are called “healthy carriers.” Sex-based anomalies (e.g. colour blindness, hemophilia, Duchenne muscular dystrophy) are more common in males than in females.</td></tr>
<tr><td>COMPULSORY CONCEPTS</td><td>PREVIOUSLY ACQUIRED KNOWLEDGE</td></tr>
<tr><td>Blood group compatibility</td><td>Determines the compatibility or incompatibility of blood groups (e.g. blood group A individuals can only receive blood from individuals with blood type O or A)</td></tr>
<tr><td>COMPULSORY CONCEPTS</td><td>KNOWLEDGE TO BE ACQUIRED</td></tr>
<tr><td>Heredity</td><td>Defines heredity as the transmission of traits from one generation to the next</td></tr>
<tr><td rowspan="3">Chromosomes</td><td>Describes the role of chromosomes as carriers of genetic information responsible for transmitting hereditary information</td></tr>
<tr><td>Uses a karyotype to distinguish sex chromosomes from autosomes</td></tr>
<tr><td>Establishes the relationship between sex chromosomes and gender determination in humans</td></tr>
<tr><td>Alleles</td><td>Defines an allele as one of the possible forms of a gene</td></tr>
<tr><td>Characteristics</td><td>Recognizes hereditary characteristics in an individual or population (e.g. eye colour, blood group, hereditary diseases)</td></tr>
</table>

Genetics (continued)	
COMPULSORY CONCEPTS	**KNOWLEDGE TO BE ACQUIRED**
Homozygous and heterozygous	Defines an individual who is homozygous for a gene as being a carrier of two identical alleles of the gene
	Defines an individual who is heterozygous for a gene as being a carrier of two different alleles of the gene
Dominance and recessiveness	Describes the phenomena of genetic dominance and recessiveness
Genotypes and phenotypes	For a given hereditary characteristic, associates the genotype with the combination of two alleles of the corresponding gene
	Associates the phenotype with observable characteristics of an individual
	Distinguishes the genotype and phenotype of an individual for one or more characteristics using the Punnett square (diagram illustrating crosses) or a karyotype
Mendel's law o Law of dominance o Law of segregation o Law of independent assortment	Describes the main mechanisms of heredity proposed by Mendel and highlights their importance for understanding heredity: law of dominance, law of segregation and law of independent assortment
	Illustrates how independent assortment increases genetic variation due to the existence of many potential chromosome combinations during gamete formation
Genetic Crosses o Autosomal inheritance o Sex-linked inheritance	Determines the proportions of monohybrid or dihybrid crossing outcomes using the rules of probability or the Punnett square
	Uses a family tree to identify the mode of transmission of a hereditary characteristic (autosomal dominant, autosomal recessive or sex-linked)
Hereditary diseases	Uses a family tree to determine the probability of a couple having a child with a hereditary disease (e.g. Huntington's disease, cystic fibrosis, sickle cell anemia, phenylketonuria, colour blindness, hemophilia or Duchenne muscular dystrophy)
	Uses a karyotype to determine whether an individual has or is a carrier of a hereditary disease

General concept: Molecular biology

Molecular biology studies processes such as replication, transcription and translation of DNA, the carrier of genetic information.

The semiconservative model of DNA replication is a highly accurate mechanism involving the pairing of the nitrogenous bases in the nucleotides. It is the process by which DNA is copied. If errors occur during the replication process, the damaged strands are repaired by enzymes.

The transcription of DNA into RNA involves its translation into proteins. In the cell nucleus, DNA is first transcribed into messenger RNA (mRNA). Translation to an amino acid chain then takes place on a ribosome in the cytoplasm using transfer RNA (tRNA). The genetic code specifies the correlation between each mRNA codon and its translation into an amino acid. The same gene can produce different proteins. However, most human DNA is noncoding.

The integrity of the DNA molecules and of the replication, transcription and translation processes ensures the health of an organism, since even a minute change in the nucleotide sequence can lead to the production of an altered protein, have negative physiological effects or produce a lasting change in the phenotype. Mutations are changes in the DNA sequence of a cell.

Many mutations are naturally occurring; they are called spontaneous mutations. Others can be caused by mutagenic agents, such as radiation, chemicals, or viral, bacterial or parasitic infections. Some genetic mutations can cause hereditary diseases or create new alleles. They also contribute to genetic diversity, which is essential for evolution.

COMPULSORY CONCEPTS	**PREVIOUSLY ACQUIRED KNOWLEDGE**
DNA	*Describes the structure of DNA (double helix)*
	Explains the role of DNA (molecule carrying the genetic code of an individual. This information can be found in every cell of the human body)
COMPULSORY CONCEPTS	**KNOWLEDGE TO BE ACQUIRED**
Genome	Defines the genome as the complete set of genetic material or as all the DNA molecules of a cell
DNA replication	Associates DNA replication with the process by which genes are duplicated and passed on to two daughter cells
Gene	Defines a gene as a DNA segment that carries the code needed for the synthesis of one or more proteins
	Describes the composition (nitrogenous bases, sugar and phosphate) and the general structure (base pairing in a double helix) of a DNA molecule
Protein synthesis o Transcription o Translation	Associates protein synthesis with the transcription of a DNA strand into mRNA followed by its translation into an amino acid sequence using tRNA
Genetic code	Uses a standard genetic code table to determine the amino acids that correspond to a codon or the coding DNA sequence for a given polypeptide sequence
Mutation	Recognizes the substitutions and the insertions or deletions of base pairs as the two main categories of point mutations occurring during DNA replication
	Identifies a few causes of mutations: errors during genetic replication, repair or recombination and exposure to mutagenic agents (e.g. radiation, chemical products or infections)
	Explains how a mutation can lead to the creation of a different protein or the absence of a protein and influence the onset of a disease or cancer

General concept: Evolution

Evolution is a process of adaptation involving the transformation of organisms over the course of the Earth's history, from the origin of life to the diversity we see today. It explains both the diversity and unity of life. Evolution can occur in response to anything that causes a change in the genetic composition of a population. On a smaller scale, it involves changes in allele frequency in a population over successive generations.

Mechanisms that promote genetic variation have variable consequences for the survival of individuals and species: they can be beneficial, harmful or neutral and, in some cases, lead to speciation. The crossbreeding of plants and animals can lead to the emergence of useful characteristics in the majority of individuals of the same species. This method, known as "artificial selection," is widely used in agronomy. It generates evolutionary changes at a much faster pace than that of the natural selection process. For example, the use of antibiotics increases the frequency of resistant bacteria arising from natural selection. In the same way, the use of chemical substances to fight insect pests promotes the emergence of resistant insects.

Interventions modifying the genetics of a species, combined with the many actions that transform the environment of living organisms, create challenges and problems for the maintenance of biodiversity. The appearance of new species or, conversely, the extinction of certain species, are issues that must be addressed by society.

COMPULSORY CONCEPTS	**PREVIOUSLY ACQUIRED KNOWLEDGE**
Genetic diversity	*Associates genetic diversity with sexual reproduction (the combination of genes from the mother and father ensures diversity)*
COMPULSORY CONCEPTS	**KNOWLEDGE TO BE ACQUIRED**
Biological evolution	Describes biological evolution as an observable process by which the hereditary characteristics of organisms change, from one generation to the next, as a result of mechanisms that promote genetic variation
Genetic diversity	Defines genetic diversity as the variability of genes within the same species
	Recognizes the impact of human activity on the distribution of living organisms and on biodiversity (e.g. there is greater genetic diversity in wild species than there is in domesticated species that have undergone artificial selection)
Gene pool	Defines a gene pool as all of the genetic information of a population at a given time
Mechanisms of microevolution ○ Natural or artificial selection ○ Gene flow ○ Genetic drift ○ Random mating ○ Mutation	Describes how selection (natural or artificial), gene flow, genetic drift (founder effect, bottleneck effect), random mating and mutations affect the gene pool of a population, from one generation to the next (e.g. high frequency of hereditary diseases in the population of Saguenay–Lac-Saint-Jean)
Adaptation	Illustrates how a population adapts to its environment by increasing the frequency of resistant forms through natural selection (e.g. antibiotic-resistant bacteria or an insect population with decreased sensitivity to an insecticide)
Interventions modifying the genetics of a species	Describes the effects of genetic techniques on the biodiversity of a species (e.g. seed selection, hybridization or cellular cloning)

<table>
<tr><th colspan="2">The Technological World</th></tr>
<tr><td colspan="2">General concept: Genetic engineering</td></tr>
<tr><td colspan="2">Almost all living organisms, whether animals, plants or bacteria, use the same genetic code: one codon codes for one amino acid. The universal nature of the genetic code makes it possible to manipulate genes and transpose them from one organism to another.

The techniques used in molecular biology to sequence, recombine, transfer and analyze genes of living organisms come under genetic engineering. Through gene insertion or modification, genetic engineering makes it possible for plants and animals to lose or acquire traits in just one generation.

The field of genetic engineering is growing at a rapid pace, and new techniques are constantly being developed. For instance, the genetic profile of an individual can be determined by analyzing specific DNA sequences that are unique to each individual. Applications in genetic engineering are used in a variety of fields, including agriculture, medicine and criminology; they tend to transform society and may even transform the human species itself.

These new techniques and their applications raise many social, ethical and environmental issues that society must address.</td></tr>
<tr><th>COMPULSORY CONCEPTS</th><th>PREVIOUSLY ACQUIRED KNOWLEDGE</th></tr>
<tr><td>Genetic transformation
○ Genetically modified organisms (GMOs)</td><td>Names the main advantages and disadvantages of genetic transformations (cancer treatments, pest-resistant and herbicide-tolerant plants, vitamin enrichment or changes in the nature of certain foods, regulations and controls)</td></tr>
<tr><th>COMPULSORY CONCEPTS</th><th>KNOWLEDGE TO BE ACQUIRED</th></tr>
<tr><td>Gene manipulation tools</td><td>Describes the main tools used to manipulate genes (e.g. a restriction enzyme cuts specific base pair sequences on both DNA strands; a cloning vector—a small piece of DNA into which a foreign DNA fragment can be inserted—is capable of autonomous replication in a host cell; recombinant DNA is used to produce therapeutic proteins)</td></tr>
<tr><td>DNA sequencing</td><td>Identifies a moral, ethical or social issue associated with genetic profiling (e.g. eugenics following screening for a hereditary disease, gene patenting or the disclosure of personal information)</td></tr>
<tr><td rowspan="4">Genetic engineering applications
○ Cloning
○ Transgenesis</td><td>Defines cloning as a process used to make identical copies of genes, cells or entire organisms</td></tr>
<tr><td>Recognizes the usefulness of genetic cloning (e.g. the production of vaccines insulin, EPO, somatotropin for dairy production, growth hormone)</td></tr>
<tr><td>Defines transgenesis as the means used to produce a GMO through the transfer of one or more foreign genes into a cell in order to modify its genome</td></tr>
<tr><td>Identifies an issue associated with applied genetic engineering (e.g. the impact of GMO production on biodiversity, the adoption of laws prohibiting human cloning, the use of gene therapy to treat genetic diseases)</td></tr>
</table>

B) CULTURAL REFERENCES

Cultural references make learning situations more meaningful. The following table presents some of the references related to this course. It is neither exhaustive nor compulsory.

<table>
<tr><th colspan="5">CULTURAL REFERENCES</th></tr>
<tr><td colspan="2">Technical objects, technological systems, processes and products</td><td colspan="3">Genetics
- Genetic testing (hereditary diseases)
Molecular biology
- DNA database (CODIS index)
- DNA samples (genetic profile)
Evolution
- Antibiogram
- Genomic selection (dairy cows)
Genetic Engineering
- Agronomy: production of transgenic crops (corn, soy)
- Glofish® (genetically modified zebrafish)
- Harvard OncoMouse
- Production of human insulin by microorganisms
- Golden rice</td></tr>
<tr><th>Area</th><th>Scientists</th><th>Community Resources</th><th>Applications</th><th>Events</th></tr>
<tr><td>The Living World</td><td>Charles Darwin
Frederick Griffith
Barbara McClintock
Gregor Mendel
Thomas Hunt Morgan
Reginald Punnett</td><td>Association de l’acidose lactique du Saguenay–Lac-St-Jean
National DNA Data Bank
Genetic Databases
Corporation de recherche et d’action sur les maladies héréditaires (CORAMH)
Génome Québec
Canadian Animal Genetic Resources
BALSAC database
Svalbard Global Seed Vault</td><td></td><td>Discovery of DNA double helix
Human Genome Project</td></tr>
<tr><td>The Technological World</td><td>Stanley Cohen and Herbert Boyer
Alec Jeffreys
Kary Mullis
Michael Smith
James Watson and Francis Crick</td><td>Association de thérapie génique du Québec
Commission de l’éthique en science et en technologie du Québec
Armand-Frappier Museum
www.ogm.gouv.qc.ca (information on GMOs)
Network of Applied Genetic Medicine</td><td>DNA Identification Act (S.C. 1998, c. 37)
Monsanto Protection Act</td><td>Cloning of Starbuck
Cartagena Protocol</td></tr>
</table>

FAMILIES OF LEARNING SITUATIONS

The learning situations in this course, derived from the *Research* and *Expertise* families, involve genetic applications and their impact on society and biodiversity. These situations cover various general concepts. The following paragraphs contain examples of tasks that could be assigned to adult learners in learning situations involving one or more general concepts.

A situation involving general concepts in molecular biology and genetic engineering might require adult learners to determine an individual's genetic profile. Adult learners could identify and describe genetic engineering techniques used to manipulate genetic information. In the case of a mutation, they would be able to demonstrate that a change in a specific DNA or RNA sequence can lead to changes in gene expression.

A situation involving molecular biology might require adult learners to reflect on the origin of a genetic disease and ask questions about the expression of genetic information, such as the relationship between DNA and protein synthesis. Making adult learners aware of the consequences of exposure to mutagenic agents enables them to assess the effects of such agents. The study of genetics can help them understand that not all genetic diseases are necessarily hereditary.

In a learning situation involving the general concepts of evolution and genetics, adult learners might process qualitative and quantitative data that reflect a change in the genetic makeup of a population over time. Using the different mechanisms of evolution, they could explain these changes or make predictions about the gene pool of a population. The study of the general concept of molecular biology would enable them to use data to make connections between DNA sequences and different traits observed in individuals.

A situation involving genetics might require adult learners to solve a problem related to heredity. Using a karyotype model, they could model the processes that ensure the transmission of genetic information. They could describe the genotype and phenotype for a specific allele or identify the maternal or paternal origin of a chromosome. They would apply their knowledge to explain how parents transmit some of their traits to their children. Adult learners could use the general concept of genetic engineering to explain how DNA is analyzed and how this permits defective genes to be identified.

In a learning situation involving the general concept of genetic engineering, adult learners might justify their opinion of a genetic engineering technique, such as genetic cloning or transgenesis. They could describe certain genetic manipulations and recognize their therapeutic applications. Using the general concept of evolution, adult learners could explain how interventions that modify the genetics of a species may have an impact on the maintenance of biodiversity.

BROAD AREAS OF LEARNING

Learning situations will have more meaning for adult learners if they are related to the broad areas of learning. The broad areas of learning that are most readily applicable to the learning situations for the course BLG-5070-2 are *Health and Well-Being*, *Environmental Awareness and Consumer Rights and*

Responsibilities, *Media Literacy* and *Citizenship and Community Life*. The examples following the presentation of the families of learning situations for this course reflect the educational aim of the broad areas of learning *Environmental Awareness and Consumer Rights and Responsibilities*, *Media Literacy* and *Citizenship and Community Life*.

Broad Areas of Learning
Health and Well-Being
Career Planning and Entrepreneurship
Environmental Awareness and Consumer Rights and Responsibilities
Media Literacy
Citizenship and Community Life

EXAMPLES OF LEARNING SITUATIONS

In the following examples of learning situations, the main tasks to be carried out help adult learners develop of the three subject-specific competencies. They fall under the *Research* and *Expertise* families.

Research Family: Looking for the Guilty Party

A few strands of hair were found at a crime scene. Police suspect two individuals and have asked you to compare the DNA collected at the crime scene with the DNA of the suspects. At the lab, they have determined the genetic profile of the DNA found at the crime scene, and the technician hands you the results of the gel electrophoresis. You have discovered the suspects' genetic profiles in their files. You must identify the guilty party.

To support your decision, your file must include:

- a presentation of the problem supported by a description of the DNA structure and its replication
- an explanation of the scientific principles used to determine an individual's genetic profile
- a DNA analysis of the suspects using restriction enzymes
- a comparison of the results of the gel electrophoresis with the analysis of the suspects' genetic profiles

Expertise Family: Transgenic plants

Joe Green's family has been growing corn for several generations and recently set aside part of their farmland for growing soybeans. However, the significant changes in precipitation and temperature of the last few years are forcing this Québec business to review its agricultural practices. Its corn crops are frequently attacked by pests, and its soybean crops are overrun with weeds.

Since the last general meeting of grain producers, Joe Green has been looking into new transgenic seeds: Bt corn seeds are pest resistant and transgenic soybeans are herbicide resistant. Would it be a good idea for Joe Green to use GMOs? Why? What are the advantages and the risks?

In your justification, be sure to provide:

- a description of what a GMO is and an explanation of the genetic code and protein synthesis
- information on transmission mechanisms for hereditary traits
- the factors that influence evolution mechanisms and the social and environmental repercussions of artificial selection following the use of GMOs
- a justification of the advantages and disadvantages of using GMOs in agriculture

END-OF-COURSE OUTCOMES

Learning situations are administered on the premise that the adult learner will become familiar with an investigative process involving the experimental method, modelling, documentary research and the observation method. In biology, these learning situations enable adult learners to apply their problem-solving skills and knowledge, and to produce messages.

Adult learners solving a problem related to genetics, its applications and their impact develop a representation of the problem based on their reading and interpretation of scientific messages. They develop an experimental protocol or a model based on one of their hypotheses, applying their knowledge of genetics, molecular biology, evolution and genetic engineering. They plan the steps of their research and select those available resources that will enable them to find answers to the questions raised. They implement a plan of action by carrying out selected activities or describe a plan of action whose activities have already been completed. In the laboratory, they demonstrate their ability to prepare and observe samples. When necessary, they use a modelling approach to solve the problem. Adult learners may also use data collections to find solutions to genetic problems. For example, they may use their understanding of genetic replication, transcription and translation to analyze data collected from a DNA test. They use concepts or laws to explain the results of genetic crosses, which they illustrate with appropriate models while applying the writing conventions for genetics. If necessary, they make corrections to the planned steps using the appropriate techniques. In a summary report, they use the results obtained, sometimes presenting the information in charts or graphs. They provide explanations that take the results into account, and check whether the hypothesis is consistent with the analysis of the results. They recognize the relationship between solving scientific problems and the development of biotechnologies.

Adult learners who study an issue or technological application involving genetics or molecular biology formulate questions related to social, ethical or environmental issues. They identify the characteristics of the issue or application in order to understand the underlying scientific principles. They explain the importance of genetic diversity for the evolution of a population and its capacity to adapt to changes in its environment. Lastly, they defend an opinion on issues related to progress in molecular biology and its resulting applications. By relying on their knowledge of genetics, they propose various explanations or solutions that take into account the issue as a whole.

EVALUATION CRITERIA FOR SUBJECT-SPECIFIC COMPETENCIES

Evaluation Criteria for Competency 1	Evaluation Criteria for Competency 2	Evaluation Criteria for Competency 3
▪ Appropriate representation of the situation ▪ Development of a suitable plan of action ▪ Appropriate implementation of the plan of action ▪ Development of relevant explanations, solutions or conclusions	▪ Formulation of appropriate questions ▪ Relevant use of scientific and technological knowledge ▪ Appropriate formulation of explanations or solutions	▪ Accurate interpretation of scientific messages ▪ Appropriate production or transmission of scientific messages

Éducation
et Enseignement
supérieur
Québec