Appendices

Appendix 1: English translation of the used test

Student Survey

1. Informed consent

☐ I declare that I have been informed about the nature of the investigation and that I understand that the data collected will be strictly anonymous and that the investigator vouches for this anonymity.

2. Do you consider the following statements to be always true, not always true, never true? You can also check the "I don't know" box.

	Always true	Not always true	Always wrong	I don't know
A) If the malformation of the ears of mice can be associated with a recessive mutant allele, an individual carrying two mutant alleles (homozygous recessive) will have abnormal ears.	☐	☐	☐	☐
B) Since Huntington's disease is a dominant monogenic disease, it will be with the same severity that two individuals carrying the mutated gene will develop the disease.	☐	☐	☐	☐

3. Check off the activities that were suggested in the biology class and the ones you wish you had in that class.

	Activities in the biology course	Activities you wish you would have liked
A) Debating the free use of genetic testing	☐	☐
B) Interact with a genetics researcher	☐	☐
C) Understanding how knowledge in the biological sciences is built	☐	☐
D) Learning what can possibly impact our health, our fertility... Why and how?	☐	☐
E) Conduct more global reflections by involving other disciplines (e.g., the ethical approach in religion/EPC, argumentation in French)	☐	☐

4. If you have the opportunity to take a genetic test online, would you do it?

☐ YES, to know my origins

☐ YES, to know my state of health

☐ YES, to know my origins and my state of health

☐ NO, because I don't rely on this data.

☐ NO, because I'm not interested.

☐ NO, because it scares me.

☐ Other

Based on your knowledge acquired throughout your school career, answer the following questions.

What justifies or refutes the following claims? For the "yes" or "no", you can tick **one or more explanations.**

5. Each phenotypic trait corresponds to a single gene. *Max 2, Min 1*

☐ YES, because a gene codes for a protein that confers a property on the individual.

☐ YES, because all phenotypic traits are monogenic.

☐ NO, because most phenotypic traits depend on several genes.

☐ NO, because some traits (a minority) are polygenic (depend on several genes).

☐ I don't know.

6. There are only two possible alleles for a trait.

☐ YES, because the allele can be recessive or dominant (two possibilities).

☐ NO, because there can be a multiplicity of alleles for a given gene.

☐ I don't know.

7. A gene systematically allows the production of a single protein. *Max 2, Min 1*

☐ YES, because the gene is a DNA sequence that is transcribed into RNA and then translated into a protein.

☐ YES, because this is indeed the function of the gene.

☐ NO, because the majority of genes are transcribed into RNA but not translated into proteins.

☐ NO, because a minority of genes are transcribed into RNA but not translated into proteins.

☐ NO, because a gene can allow the production of one or more proteins.

☐ I don't know.

8. The more complex an organism is, the higher the number of genes.

☐ YES, because the complexity of an organism depends on the amount of genetic information available.

☐ NO, because it is the multiplicity of interactions between genes, proteins and RNA that creates complexity.

☐ I don't know.

9. Check everything that the wider environment (i.e. the environment of cells, organs, the individual and the population) can influence. *Max 12, Min 1*

☐ Interactions between organisms

☐ Phenotypic characteristics of the organism

☐ The activity of certain enzymes

☐ The development of the body

☐ The characteristics that will be transmitted to the offspring

☐ The survival of an organism and therefore its ability to reproduce

☐ The adaptability of an organism

☐ Pigmentation of the body

☐ Transcription of DNA into RNA

☐ The translation of RNA into protein

☐ The appearance of genetic mutations

☐ Susceptibility to a disease

10. Célian clones his pet of which here is the photo:

Check the photo(s) that could match the clone. *Max 4, Min 1*

☐ ☐ ☐ ☐

Photos of calico cats. Source: Canva Education.

Appendix 2: English translation of the used test

Survey of teachers

1. Have you spotted any misconceptions in your students about the concepts of genetics? If so, name the most recurrent ones.

2. Here are cognitive skills that can be developed in genetics classes. Check off the ones <u>you don't cover</u>, whether it's in basic science, general science, science education, or science training.

☐ Represent the different combinations of possible gametes on a crossover chessboard.

☐ To propose an explanation of phenotypic variability based on identical genotypes.

☐ To compare phenotypes and genotypes for a given gene of healthy and diseased individuals and to make the link between the onset of a genetic disease and a change in the nucleotide sequence of DNA.

☐ Explain how a phenotypic trait is inherited and determine the genotypes and phenotypes of individuals, based on a family tree describing trait transmission.

☐ Characterize a phenotypic trait as innate or acquired.

☐ To highlight the influence of the environment in the broad sense (environment of cells, organs, the individual or the population) on the expression of certain genes.

☐ To conceive that the phenotype is, generally, the result of interactions between genetic inheritance and factors that will influence the activity of genes.

☐ Distinguish a polygenic trait from a monogenic trait.

☐ Conceive of an individual's genotype as the set of alleles of the individual, whether or not they have an effect on the phenotype.

☐ Consider the influence of genetic variants on the penetrance and expressiveness of a phenotype.

3. Some of the cognitive skills listed in the previous question have not been developed in your students.

☐ Due to lack of time and interest

☐ Due to lack of time but with the wish to be able to tackle them another year

☐ Due to lack of interest

☐ Due to a lack of mastery of these concepts

☐ They have all been developed.

☐ Other reason(s):

4. In your courses, do you address phenotypic variability? If so, in what situation(s)?

☐ Diversity of appearances within a population

☐ Differences between two identical twins or two clones

☐ Phenotypic adaptation of an individual according to environmental conditions

☐ No, I am not addressing phenotypic variability.

5. Name the factors you indicate to your students as influencing phenotypic variability.

6. In your courses, do you develop the skill "*Illustrate from an example that the environment can modify certain phenotypes*"?

☐ Yes, I am developing this skill.

☐ Yes, except for the last two years because this skill has not been included in the "Essentials" (school curriculum adapted for the covid period).

☐ No, I don't develop this skill.

7. What example(s) do you use to develop this skill?

☐ Hydrangea colour

☐ Coat colour of the Siamese cat

☐ Coat colour of the calico cat and/or tortoiseshell cat

☐ Severity of phenylketonuria

☐ Development of diseases

☐ Status of bees (workers and queens)

☐ Shape of daphnids

☐ Locust behaviour

☐ Stress management in mice

☐ Cell types

☐ Metabolism of organisms

☐ Other

8. I don't develop this skill...

☐ Due to lack of time and interest

☐ Due to lack of time but with the wish to be able to tackle them another year

☐ Due to lack of interest

☐ Due to a lack of mastery of these concepts

☐ This skill is not required in the school curriculum I am supposed to follow.

☐ Other

9. Prior to 2014, the competency cited in the previous question was worded as follows: "*Illustrate from an example that the environment can modify the expression of certain genes*". After 2014, "*modify the expression of certain genes*" was replaced by "*modify the expression of certain phenotypes*". How do you perceive this change?

☐ This change seems relevant to me.

☐ This change seems relevant to me, but I would have written "*modify the appearance of certain phenotypes*" instead of "*modify the expression of certain phenotypes*".

☐ This change leaves me indifferent.

☐ This change seems to me to be of little relevance.

10. Why? *

11. What example(s) would you choose to illustrate that the environment can modify the expression of certain genes?

☐ Hydrangea colour

☐ Coat colour of the Siamese cat

☐ Coat colour of the calico cat and/or tortoiseshell cat

☐ Severity of phenylketonuria

☐ Development of diseases

☐ Status of bees (workers and queens)

☐ Shape of daphnids

☐ Locust behaviour

☐ Stress management in mice

☐ Adaptation and survival of organisms

☐ Adaptation of organisms and their progeny

☐ Cell types

☐ Metabolism of organisms

☐ Other

12. Do you talk to your students about the free use of genetic tests (to find out their origins, their state of health)?

☐ Yes ☐ No

13. Possible remarks about discussions on this subject with your students:

14. In the context of the chapter on "Genetics", do you promote activities that actively engage students in their own learning?

☐ Yes ☐ No

15. What type(s) of activity?

☐ Debates open to divergent positions

☐ Debates with a guest speaker

☐ Work on contradictory documents

☐ Activities that lead students to distinguish between knowledge and beliefs and facts and opinions

☐ Activities that lead students to decode complex situations (establish causal links, analyse all parameters, etc.)

☐ Activities that lead students to gradually build models that are closer and closer to reality.

☐ Activities for the co-construction of new knowledge (pupils/pupils – pupils/teachers)

☐ Other

16. Briefly explain how one or two of these activities were carried out.

17. When you discuss scientific models, theories, or laws (e.g., Mendel's laws, theories of evolution), do you mention factors that influence the perception, acceptance, and understanding of these patterns, theories, or laws?

☐ Yes ☐ No

18. What factors do you mention?

19. Do you carry out didactic activities in collaboration with teachers from other disciplines?

☐ Yes ☐ No

20. What discipline(s) are they from? What transversal skill(s) do you develop in your students through these activities? What is the added value(s) of this type of activity?

21. Why?

☐ Due to lack of time

☐ This collaboration is complicated to organise.

☐ I don't see any added value in this type of activity.

22. Have people outside the school ever been involved in your biology classes?

☐ Yes ☐ No

23. For what type of expertise?

☐ Genetics

☐ Epigenetics

☐ Evolution

☐ Epistemology

☐ History of science

☐ Philosophy of Science

☐ Bioethics

☐ Critical thinking

☐ Other

24. Do you think this approach would be useful and should be preferred for your students?

☐ Yes ☐ No

25. I would prefer to interact with experts in:

☐ Genetics

☐ Epigenetics

☐ Evolution

☐ Epistemology

☐ History of science

☐ Philosophy of Science

☐ Bioethics

☐ Critical thinking

☐ Other

Appendix 3: Examples of cognitive skills that promote misconceptions in genetics in the school curriculums of subsidized private French-speaking Belgian education:

- Solve a simple **monohybridism** problem (4th General Transition and 4th Technical Transition).
- Based on a given diagram, explain the **conceptual links between chromosome, DNA, gene, protein, and hereditary trait** (5th Technical Qualification and Transition).
- Solve a **simple heredity problem** in humans (5th Technical Qualification and Transition).
- Construct a Punnett square to explain a case of **monohybridism** in humans (5th Technical Qualification and Transition).
- In the case of a genetic disease, establish a **relationship between phenotypes and the DNA sequence** (6th General Transition).
- From a human pedigree, interpret the transmission of a trait and establish the relationship between **phenotypes and the DNA sequence** (6th General Transition).

Examples of excerpts that promote misconceptions in genetics in textbooks:

- **Bio 6 Van In General Sciences (2023)**
    - "The **genotype** of an individual for a given **trait** is the set of two alleles of the studied trait." (p.24).
    - "An individual is **homozygous/heterozygous** for a **trait** …" (p.24).
    - The use of the example of Siamese cat fur to illustrate that the expression of certain alleles depends on the environment (p. 29).
- **Biology 6 de boeck Basic Sciences (2012)**
    - "**Innate trait**: hereditary trait, transmitted over generations / **acquired trait**: non-hereditary trait that depends on environmental conditions." (pp. 24 and 174).
    - Dominant **traits** (= visible traits) / dominant **alleles** (= alleles that mask recessive alleles) (pp. 24 and 25).
    - "The **gene** is the hereditary factor that determines a **trait**." (p. 25).
    - "The **alleles** of a **trait** …" (p. 25).
    - "Recessive **alleles** are only **visible** in a homozygote." (p. 25).
    - "Several **alleles** can define the same **trait**." (p. 25).
    - "The genotype includes all the **alleles** of an organism, their expression generates the **phenotype**." (p. 25).
    - "Hereditary **traits** are determined by **genes**." (p. 31).
- **Essentia 6 Biology Plantyn Basic Sciences (2019)**
    - "An individual who carries two identical alleles for a **gene** is said to be homozygous for that **trait**." (p.39).
    - "An individual who carries two different alleles for a **gene** is said to be heterozygous for that **trait**." (p.39).
    - "The **phenotype** represents the observable traits for the considered **gene(s)** of an individual." (p.39).
    - "The gene 'corn kernel colour'" (p. 39).

Mention of epigenetics in textbooks:

- **Bio 6 Van General Sciences (2023)**
  - The word epigenetics is mentioned only once (p. 71: "Several phenotypes for a genotype": example of the agouti gene).
  - HOX genes are mentioned in Chapter 2 "Genetics" (p. 84: "How is embryonic development controlled?") and in Chapter 3 "Evolution and species diversity" (p. 130: experimental embryology) but it is not specified that they are involved in epigenetic mechanisms.
- **Biology 6 de boeck Basic Sciences (2012)**
  - The word epigenetics is not mentioned.
- **Essentia 6 Biology Plantyn Basic Sciences (2019)**
  - Epigenetics is illustrated by the example of **Calico cat** fur (X chromosome inactivation) (p. 43).
  - Epigenetics is explained in a "**To learn more**" section (epigenetics - epigenetics and diseases - epigenetics and therapies) (pp.47-48).
  - Epigenetics is defined in the **glossary** (Epigenetics: Study of factors that influence the phenotype without affecting the genotype) (p. 213).

Appendix 4:

Analysis of textbook excerpts through the lens of the *knowledge in pieces* framework

Textbook Excerpt	**Interpretation and learning risks**
"the genotype is the set of two alleles for a trait."	May lead students to believe that all traits are controlled by a single gene with two alleles, reinforcing a simplified view of inheritance.
"The gene determines a trait."	Suggests a direct link between gene and trait, ignoring gene regulation and environmental influence.
Dominant **traits** (= visible traits) / dominant **alleles** (= alleles that mask recessive alleles)	May lead to confusion between visibility of a trait and the genetic concept of dominance, potentially reinforcing the idea that dominant always means more frequent or more visible.
"Innate = hereditary / Acquired = environmental."	Reinforces a strict separation between nature and nurture, masking the complexity of gene-environment interactions.

Explanatory Box:

A Brief History of Genetics and Epigenetics

In 1905, William Bateson was the first to suggest the term "**genetics**" to describe the study of heredity. This study led to the introduction of the concept of the gene in 1909.

In the 1940s, the term "genetics" was primarily associated with the transmission of hereditary traits across generations. At that time, genetics was largely viewed through the lens of Mendel's laws on the inheritance of traits determined by specific genes. During this period, Waddington expanded the notion of genetics by incorporating the complex processes that govern the development of organisms. He introduced the term "**epigenetics**" to reconcile two distinct research fields: epigenesis and genetics. Epigenesis refers to Aristotle's idea that living beings develop gradually from undifferentiated materials, with each part forming successively above (epi) the others.

Today, we have a more molecular definition of epigenetics. Within the framework of genetics, it is a discipline that focuses on modifications that can occur at the DNA level and/or at the level of proteins associated with DNA, such as histones. These modifications, induced by stimuli from the organism or the environment, do not alter the DNA sequence but influence gene activity or expression, and thus heredity.

Basic Concepts in Genetics

- The **genome** is the entire genetic material of an individual, encompassing all DNA sequences, including those corresponding to genes.
- A **gene** is a sequence of DNA (polynucleotide sequence), part of which codes for one or more functional molecules (polypeptide, tRNA, rRNA, or other types of RNA).
- In humans, of the entire DNA contained in a cell nucleus:
    - 1 to 2% (about 20,000 segments of the DNA molecule) code for polypeptides via mRNA. A segment can code for one or sometimes several polypeptides. There is a multiplicity of possible alleles for each of these segments.
    - 98 to 99% do not code for polypeptides. These regions have other important functions, such as regulating gene expression. Some of these regions are transcribed into RNA, other than mRNA, while others consist of inactive genes or regions with unknown functions.
- The **genotype** of an individual is the set of alleles of the individual, whether or not they affect the phenotype.
- The **phenotype** is the result of interactions between genetic inheritance and factors that influence gene activity.

  Most phenotypic traits are **polygenic**, meaning they result from the combination of alleles from multiple genes that together contribute to the phenotype.
- The **penetrance** of a phenotype refers to the probability that a genetic mutation will result in the associated phenotype.
- The **expressivity** of a phenotype refers to the variability in the severity of a phenotype caused by a genetic mutation.
- The **modifications in gene expression** can be genetic or epigenetic:

- **Genetic modifications**: Changes induced by uncorrected replication errors or mutagenic agents that alter the DNA sequence, which are reversible with an extremely low probability and are transmitted during cell divisions.
- **Epigenetic modifications**: Changes induced by stimuli from the organism or the environment that do not alter the DNA sequence, are reversible with a high probability, and can be transmitted during cell divisions. These modifications are due to various, often complex mechanisms, sometimes incompletely characterized (with variable consequences on gene expression), like chemical modification of DNA bases (e.g. Methylation of cytosines), or post-translational modification of histones.