Family Secrets A Problem-Based Learning Case

Introduction

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Intro: 1

The **Family Secrets** problem-based learning case was developed in as a joint venture between the University of Rochester's Center for Science Education and Outreach and the New York State Biology-Chemistry Professional Development Network. The goals of this project were to develop and disseminate problem-based learning curricula and associated classroom resources that focus on critical thinking skills and hands-on activities pertaining to genetic technology and its associated ethical, legal, and social implications (ELSI).

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Family Secrets Coach's Guide

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Family Secrets Part 1: A Family Disease

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Introduction

The Human Genome Project has provided, and will continue to provide, detailed information on "the code of life." As a result of this information, genetic research has begun to change our lives. Genetic advances make daily headlines and provide new challenges for educators. Today's students need to understand these genetic advances and their ethical, legal and social implications,

Family Secrets is designed to provide a starting point for capturing and expanding on student interest in genetic technology. *Family Secrets* uses a Problem-Based Learning (PBL) approach to illustrate some of the ethical, legal and social implications of modern genetic technology. It incorporates a gel electrophoresis lab to help students understand the technology of genetic testing.

The learning objectives for the *Family Secrets* PBL focus on developing students' understanding of:

- Classical and molecular genetics of human genetic disorders
- Ethical, Legal, and Social Implications (ELSI) of genetic disorders for individuals and families
- Simple molecular techniques for genetic testing
- Decision-making when dilemmas involve ELSI issues and potential risks and benefits.

Family Secrets includes numerous resources designed to support many different classroom approaches to teaching the *Family Secrets* PBL. Teachers are encouraged to select and modify these resources to accommodate the culture of the school. The *Family Secrets* Virtual Laboratory is an interactive tutorial on genetic testing for Huntington's disease.

Overview

The Family Secrets PBL consists of five parts:

- **Part 1: A Family Disease** introduces Jenny, a teenager with a family history of Huntington's disease. Students learn about Huntington's disease and its impact on families.
- **Part 2: The Dilemma** follows Jenny as she talks with her family doctor about genetic testing for Huntington's disease. Students learn about genetic testing (the process and its risks, benefits, and limitations).
- **Part 3: A Difficult Choice** follows Jenny as she talks with her father about whether she should or should not be tested for the Huntington's disease gene. Students learn about a process for making decisions involving issues that have ethical, legal, and social implications.
- **Part 4: Testing for the HD Gene** simulates the laboratory techniques used in testing family members for the Huntington's disease gene. Students learn about the molecular techniques used to test patients for the Huntington's disease gene.
- **Part 5: Making Decisions** follows Jenny as she talks with her brother about their family's future following the results of the genetic testing. Students consider the short and long term consequences and societal implications and of knowing the results of genetic tests.



Family Secrets Resources

Family Secrets includes extensive resources to support the implementation of the problem-based learning activity, including: detailed coaching guides, all student hand-outs for Parts 1-5 of the PBL case, workshop PowerPoints, and information on the PBL process.



Family Secrets Virtual Laboratory

The *Family Secrets* Virtual Laboratory is an interactive tutorial on genetic testing for Huntington's disease. There are three sections to the Virtual Laboratory:

1. Background on Huntington's Disease - This section includes information on the symptoms and molecular basis of Huntington's disease. It also provides a brief introduction to genetic testing for Huntington's disease.

2. Virtual genetic testing procedure for Huntington's disease - This section includes four subsections:

- 1) **Isolating DNA from a Blood Sample** This section starts with taking a blood sample from a patient, and allows the user to centrifuge a blood sample to isolate white blood cells, and treat the white blood cells with protease and detergent to release its DNA.
- 2) **Conducting a Polymerase Chain Reaction (PCR)** This section starts with the DNA sample isolated in the previous section, and allows the user to assemble the components of a PCR reaction, and use a thermal cycler to conduct the reaction. This reaction is which is illustrated in an animated sequence.
- 3) Preparing The Gel This section allows the user to melt agarose, assemble a gel electrophoresis apparatus, and pour the melted gel into the electrophoresis apparatus. It includes an animated sequence that illustrates how the melted agarose polymerizes to form a matrix of agarose polymers.
- 4) Loading Samples Into an Agarose Gel This section allows the user to load the PCR products produced in subsection b and additional controls, into the agarose gel. It also allows the user to go through the electrophoresis process, and includes an animated sequence that illustrates how DNA samples would be separated during electrophoresis. The results of electrophoresis are also discussed in this section.

3. The Family Secrets laboratory activity*

- Gel Electrophoresis: Family Secrets This section allows the user to load PCR products from genetic tests done for Jenny, Jeremy and James Lanahan. This section also allows the user to perform agarose gel electrophoresis of the PCR products, positive and negative controls, and DNA size standards. "gelsix" is the password. The password for this section is "gelsix"
- 2) **Data Analysis:** *Family Secrets* The results of the electrophoresis performed in the previous section are discussed here.
- * This section is an alternative version of the hands-on laboratory activity for *Family Secrets*. It should NOT be used as a replacement for the hands-on laboratory activity, unless the hands-on laboratory activity is not feasible for the class. The password for this section is "gelseven".

Problem-Based Learning

Problem-based learning (PBL) is an instructional approach which enables learners to simultaneously develop problem solving strategies, content knowledge, and research skills. Students become active learners as they work to solve a real-world problem. Teachers assume the role of cognitive coaches rather than knowledge disseminators.

Prior to a teacher beginning *Family Secrets*, we highly recommend that both the teacher and the students be familiar with cooperative learning strategies and the problem based learning (PBL) process. Teachers who are not familiar with the PBL process and who have not used PBL in their classrooms should familiarize themselves with the information, sample PBL activities, and sample rubric in the found in the *Family Secrets* Introduction folder in the Resource CD. It is assumed that students will have completed one introductory PBL activity before beginning the *Family Secrets* PBL. The *Penguin Peril* PBL was specifically designed to introduce students to the PBL process.

Steps in the PBL process



Constructivist and Traditional Approaches

The approach used to integrate *Family Secrets* into the curriculum is the teacher's decision.

Constructivist Approach:

Start *Family Secrets* at the beginning of the genetics unit. Use each part of the PBL as a "hook" to engage student interest in the genetics content topics. Integrate supporting content instruction, when needed, <u>within</u> each part of the PBL. For example:

- Part 1: Classical and molecular genetics.
- Part 2: Biotechnology.
- Part 3: ELSI issues and bioethics
- Part 4: Biotechnology
- Part 5: Scientific research and reproductive technologies

Traditional Approach:

Provide all instruction in genetics content topics <u>prior to</u> starting *Family Secrets*. Then use *Family Secrets* as a culminating experience to encourage students to apply their learning to real-life issues.

Sample Lesson Sequence

Those involved in the development of *Family Secrets* hope that teachers recognize the value of all five parts of the PBL in developing students' genetic literacy. Based on 40-minute class periods, a sample 11-class period lesson sequence, with additional optional alternative assessments, is described in the Parts 1 through 5 Coach's Guides. Teachers may modify this sequence to meet the needs of their students and/or curricular time constraints.

Class 1	Part 1: A Family Disease							
Class 2								
Class 3	Part 2: The Dilemma							
Class 4								
Class 5								
Class 6	Part 3: A Difficult Choice							
Class 7								
Class 8								
Class 9	Part 4: Testing for the HD Gene							
Class 10]							
Class 11	Part 5: Making Decisions							
optional	Alternative Assessments							

Formative and Summative Assessment

Opportunities for both group and individual formative assessment opportunities are provided in Parts 1-5. See a sample "grade book" in Introduction: Appendix A. Part 5 also includes a sample "Genetics Test" and suggestions for alternative, summative assessments.

Correlation with New York State Math, Science, and Technology (MST) Learning Standards

<u>Standard 1: Analysis, Inquiry, and Design</u> – Students will use mathematical analysis, scientific inquiry, and engineering design, as appropriate, to pose questions, seek answers, and develop solutions.

Science relies on logic and creativity. Science is both a body of knowledge and a way of knowing – an intellectual and social process that applies human intelligence to explaining how the world works. Scientific explanations are developed using both observations (evidence) and what people already know about the world (scientific knowledge). All scientific explanations are tentative and subject to change. Good science involves questioning, observing and inferring, experimenting, finding evidence, collecting and organizing data, drawing valid conclusions, and undergoing peer review. Understanding the scientific view of the natural world is an essential part of personal, societal, and ethical decision making. Scientific literacy involves internalizing the scientific critical attitude that it can be applied in everyday life, particularly in relation to health, commercial, and technological claims.

Key Idea 1: The central purpose of scientific inquiry is to develop explanations of natural phenomena in a continuing and creative process

Performance Indicator 1.1: Elaborate on basic and personal explanation of natural phenomena, and develop extended visual models and mathematical formulations to represent one's thinking.

Major Understanding 1.1.c: Science provides knowledge, but values are also essential to making effective and ethical decisions about the application of scientific knowledge.

Performance Indicator 1.2: Hone ideas through reasoning, library research, and discussions with others, including experts

Major Understanding 1.2a: Inquiry involves asking questions and locating, interpreting, and processing information from a variety of sources

Major Understanding 1.2b: Inquiry involves making judgments about the reliability of the source and relevance of information

Performance Indicator 1.3: Work towards reconciling competing explanations; clarify points of agreement and disagreement

Major Understanding 1.3b:All scientific explanations are tentative and subject to change or improvement. Each new bit of evidence can create more questions than it answers. This leads to increasingly better understanding of how things work in the living world.

Performance Indicator 1.4: Coordinate explanations at different levels of scale, points of focus, and degrees of complexity and specificity, and recognize the need for alternative explanations of the natural world.

Major Understanding 1.4a: Well-accepted theories are ones that are supported by different kinds of scientific investigations often involving the contributions of individuals from different disciplines.

Key Idea 2: Beyond the use of reasoning and consensus, scientific inquiry involves the testing of proposed explanations involving the use of conventional techniques and procedures and usually requires considerable ingenuity.

Performance Indicator 2.2: Refine research ideas through library investigation, including electronic information retrieval and reviews of literature, and through peer feedback obtained from review and discussion

Key Idea 3: The observations made while testing proposed explanations, when analyzed using conventional and invented methods, provide new insights into natural phenomena.

Performance Indicator 3.1: Use various methods of representing and organizing observations (e.g., diagrams, tables, charts, graphs, equations, matrices) and insightfully interpret the organized data.

Major Understanding 3.1a: Interpretation of data leads to development of additional hypotheses, the formulation of generalizations, or explanations of natural phenomena.

Performance Indicator 3.4: Based on the results of the test and thorough public discussion, revise the explanation and contemplate additional research.

Major Understanding 3.4a: Hypotheses are valuable, even if they turn out not to be true, because they may lead to further investigation.

Major Understanding 3.4b: Claims should be questioned if the data are based on samples that are very small, biased, or inadequately controlled or if the conclusions are based on the faulty, incomplete, or misleading use of numbers.

Major Understanding 3.4c: Claims should be questioned if fact and opinion are intermingled, if adequate evidence is not cited, or if the conclusions do not follow logically from the evidence given

Performance Indicator 3.5: Develop a written report for public scrutiny that describes the proposed explanation, including a literature review, the research carried out, and suggestions for further research.

Major Understanding 3.5a: One assumption of science is that other individuals could arrive at the same explanation if they had access to similar evidence. Scientists make the results of their investigations public; they should describe the investigations in ways that enable others to repeat the investigations.

Major Understanding 3.5b: Scientists use peer review to evaluate the results of scientific investigations and the explanations proposed by other scientists. They analyze the experimental procedures, examine the evidence, identify faulty reasoning, point out statements that go beyond the evidence, and suggest alternative explanations for the same observations.

<u>Standard 4: Science Content</u> - Students will understand and apply scientific concepts, principals, and theories pertaining to the physical setting and the living environment and recognize historical development of ideas in science.

Key Idea 1: Living things are both similar to and different from each other and from non-living things.

Living things are similar in that they rely on many of the same processes to stay alive, yet are different in the ways that these processes are carried out.

Nonliving things lack certain features of living organisms, such as the ability to maintain a cellular organization, carry out metabolic processes while maintaining internal stability (homeostasis), and pass on hereditary information through reproduction.

In most biological respects, humans are like other living organisms. For instance, they are made up of cells like those of other animals, have much the same chemical composition, have organ systems and physical characteristics like many others, reproduce in a similar way, carry the same kind of genetic information system, and are part of a food web.

The components of living systems, from a single cell to an ecosystem, interact to maintain balance. Different organisms have different regulatory mechanisms that function to maintain the level of organization necessary for life. Diversity is evident and important at all levels of organization, from a single cell to a multi-cellular organism to an ecosystem.

Performance Indicator 1.2: Describe and explain the structures and functions of the human body at different organizational levels (e.g., systems, tissues, cells, organelles).

Major Understanding 1.2a: Important levels of organization for structure and function include organelles, cells, tissues, organs, organ systems, and whole organisms.

Major Understanding 1.2b: Humans are complex organisms. They require multiple systems for digestion, respiration, reproduction, circulation, excretion, movement, coordination, and immunity. The systems interact to perform the life functions.

Major Understanding 1.2d: If there is a disruption in any human system, there may be a corresponding imbalance in homeostasis.

Major Understanding 1.2j: Inside the cell a variety of specialized structures, formed from many different molecules, carry out the transport of materials (cytoplasm), extraction of energy from nutrients (mitochondria), protein building (ribosomes), waste disposal (cell membrane), storage (vacuole), and information storage (nucleus).

Key Idea 2: Organisms inherit genetic instructions in a variety of ways that result in continuity of structure and function between parents and offspring.

Organisms from all kingdoms possess a set of instructions (genes) that determines their characteristics. These instructions are passed from parents to offspring during reproduction. Students are familiar with simple mechanisms related to the inheritance of some physical traits in offspring. They are now able to begin to understand the molecular basis of heredity and how this set of instructions can be changed through recombination, mutation, and genetic engineering.

The inherited instructions that are passed from parent to offspring exist in the form of a code. This code is contained in DNA molecules. The DNA molecules must be accurately replicated before being passed on. Once the coded information is passed on, it is used by a cell to make proteins. The proteins that are made become cell parts and carry out most functions of the cell.

Throughout recorded history, humans have used selective breeding and other biotechnological methods to produce products or organisms with desirable traits. Our current understanding of DNA extends this to the manipulation of genes leading to the development of new combinations of traits and new varieties of organisms.

Performance Indicator 2.1: Explain how the structure and replication of genetic material result in offspring that resemble their parents.

Major Understanding 2.1a: Genes are inherited, but their expression can be modified by interactions with the environment.

Major Understanding 2.1b: Every organism requires a set of coded instructions for specifying its traits. For offspring to resemble their parents, there must be a reliable way to transfer information from one generation to the next. Heredity is the passage of these instructions from one generation to another.

Major Understanding 2.1c: Hereditary information is contained in genes, located in the chromosomes of each cell. An inherited trait of an individual can be determined by one or by many genes, and a single gene can influence more than one trait. A human cell contains many thousands of different genes in its nucleus.

Major Understanding 2.1e: In sexually reproducing organisms, the new individual receives half of the genetic information from its mother (via the egg) and half from its father (via the sperm). Sexually produced offspring often resemble, but are not identical to, either of their parents.

Major Understanding 2.1f: In all organisms, the coded instructions for specifying the characteristics of the organism are carried in DNA, a large molecule formed from subunits arranged in a sequence with bases of four kinds (represented by A, G, C, and T). The chemical and structural properties of DNA are the basis for how the genetic information that underlies heredity is both encoded in genes (as a string of molecular "bases") and replicated by means of a template.

Major Understanding 2.1g: Cells store and use coded information. The genetic information stored in DNA is used to direct the synthesis of the thousands of proteins that each cell requires.

Major Understanding 2.1h: Genes are segments of DNA molecules. Any alteration of the DNA sequence is a mutation. Usually, an altered gene will be passed on to every cell that develops from it.

Major Understanding 2.1i: The work of the cell is carried out by the many different types of molecules it assembles, mostly proteins. Protein molecules are long, usually folded chains made from 20 different kinds of amino acids in a specific sequence. This sequence influences the shape of the protein. The shape of the protein, in turn, determines its function.

Major Understanding 2.1j: Offspring resemble their parents because they inherit similar genes that code for the production of proteins that form similar structures and perform similar functions.

Major Understanding 2.1k: The many body cells in an individual can be very different from one another, even though they are all descended from a single cell and thus have essentially identical genetic instructions. This is because different parts of these instructions are used in different types of cells, and are influenced by the cell's environment and past history.

Performance Indicator 2.2: Explain how the technology of genetic engineering allows humans to alter genetic makeup of organisms.

Major Understanding 2.2c: Different enzymes can be used to cut, copy, and move segments of DNA. Characteristics produced by the segments of DNA may be expressed when these segments are inserted into new organisms, such as bacteria.

Major Understanding 2.2d: Inserting, deleting, or substituting DNA segments can alter genes. An altered gene may be passed on to every cell that develops from it.

Major Understanding 2.2e: Knowledge of genetics is making possible new fields of health care; for example, finding genes which may have mutations that can cause disease will aid in the development of preventive measures to fight disease. Substances, such as hormones and enzymes, from genetically engineered organisms may reduce the cost and side effects of replacing missing body chemicals.

Key Idea 3: Individual organisms and species change over time.

Performance Indicator 3.1: Explain the mechanisms and patterns of evolution.

Major Understanding 3.1b: New inheritable characteristics can result from new combinations of existing genes or from mutations of genes in reproductive cells.

Major Understanding 3.1c: Mutation and the sorting and recombining of genes during meiosis and fertilization result in a great variety of possible gene combinations.

Major Understanding 3.1d: Mutations occur as random chance events. Gene mutations can also be caused by such agents as radiation and chemicals. When they occur in sex cells, the mutations can be passed on to offspring; if they occur in other cells, they can be passed on to other body cells only.

Major Understanding 3.1g: Some characteristics give individuals an advantage over others in surviving and reproducing, and the advantaged offspring, in turn, are more likely than others to survive and reproduce. The proportion of individuals that have advantageous characteristics will increase.

Key Idea 4: The continuity of life is sustained through reproduction and development.

Species transcend individual life spans through reproduction. Asexual reproduction produces genetically identical offspring. Sexual reproduction produces offspring that have a combination of genes inherited from each parent's specialized sex cells (gametes). The processes of gamete production, fertilization, and development follow an orderly sequence of events. Zygotes contain all the information necessary for growth,

development, and eventual reproduction of the organism. Development is a highly regulated process involving mitosis and differentiation. Reproduction and development are subject to environmental impact. Human development, birth, and aging should be viewed as a predictable pattern of events. Reproductive technology has medical, agricultural, and ecological applications.

Performance Indicator 4.1: Explain how organisms, including humans, reproduce their own kind.

Major Understanding 4.1a: Reproduction and development are necessary for the continuation of the species.

Major Understanding 4.1c: The processes of meiosis and fertilization are key to sexual reproduction in a wide variety of organisms. The process of meiosis results in the production of eggs and sperm which each contain half of the genetic information. During fertilization, gametes unite to form a zygote, which contains the complete genetic information for the offspring.

Major Understanding 4.1d: The zygote may divide by mitosis and differentiate to form the specialized cells, tissues, and organs of multi-cellular organisms.

Major Understanding 4.1h: In humans, the embryonic development of essential organs occurs in early stages of pregnancy. The embryo may encounter risks from faults in its genes and from its mother's exposure to environmental factors such as inadequate diet, use of alcohol/drugs/tobacco, other toxins, or infections throughout her pregnancy.

Key Idea 5: Organisms maintain a dynamic equilibrium that sustains life.

Life is dependent upon availability of an energy source and raw materials that are used in the basic enzyme-controlled biochemical processes of living organisms. These biochemical processes occur within a narrow range of conditions. Because organisms are continually exposed to changes in their external and internal environments, they must continually monitor and respond to these changes. Responses to change can range in complexity from simple activation of a cell chemical process to elaborate learned behavior. The result of these responses is called homeostasis, a "dynamic equilibrium" or "steady state" which keeps the internal environment within certain limits. Organisms have a diversity of homeostatic feedback mechanisms that detect deviations from the normal state and take corrective actions to return their systems to the normal range. These mechanisms maintain the physical and chemical aspects of the internal environment within narrow limits that are favorable for cell activities. Failure of these control mechanisms can result in disease or even death.

Performance Indicator 5.1: Explain the basic biochemical processes in living organisms and their importance in maintaining dynamic equilibrium.

Major Understanding 5.1c: In all organisms, organic compounds can be used to assemble other molecules such as proteins, DNA, starch, and fats. The chemical energy stored in bonds can be used as a source of energy for life processes.

Major Understanding 5.1f: Biochemical processes, both breakdown and synthesis, are made possible by a large set of biological catalysts called enzymes. Enzymes can affect the rates of chemical change. The rate at which enzymes work can be influenced by internal environmental factors such as pH and temperature.

Major Understanding 5.1g: Enzymes and other molecules, such as hormones, receptor molecules, and antibodies, have specific shapes that influence both how they function and how they interact with other molecules.

Performance Indicator 5.2: Explain disease as a failure of homeostasis.

Major understanding 5.2a: Homeostasis in an organism is constantly threatened. Failure to respond effectively can result in disease or death.

Major Understanding 5.2h: Disease may also be caused by inheritance, toxic substances, poor nutrition, organ malfunction, and some personal behavior. Some effects show up right away; others may not show up for many years.

Major Understanding 5.2j: Biological research generates knowledge used to design ways of diagnosing, preventing, treating, controlling, or curing diseases of plants and animals.

Key Idea 7: Human decision and activities have had a profound impact on the physical and living environment.

Population growth has placed new strains on the environment - massive pollution of air and water, deforestation and extinction of species, global warming, and alteration of the ozone shield. Some individuals believe that there will be a technological fix for such problems. Others, concerned with the accelerating pace of change and the ecological concept of finite resources, are far less optimistic. What is certain, however, is that resolving these issues will require increasing global awareness, cooperation, and action.

Since the students of today will be the elected officials and informed public of tomorrow, the teacher should encourage a diversity of activities that will allow students to explore, explain, and apply conceptual understandings and skills necessary to be environmentally literate.

Performance Indicator 7.3: Explain how individual choices and societal actions can contribute to improving the environment.

Major Understanding 7.3a: Societies must decide on proposals which involve the introduction of new technologies. Individuals need to make decisions which will assess risks, costs, benefits, and trade-offs.

Major Understandings 7.3b: The decisions of one generation both provide and limit the range of possibilities open to the next generation.

Introduction: Appendix A – Sample "Grade Book."

The left-hand column of the chart below represents some of the possible individual or group (student PBL teams) assignments that may become part of the overall assessment package associated with *Family Secrets*. Other assignments and assessments are possible.

	Part #	Individual or Team					
Record of Individual and Team Work-Facts/Questions	1	Individual or Team					
Record of Individual and Team Work-Facts/Questions	1	Individual or Team					
Class participation	1	Individual					
Individual Student Research	2	Individual					
Category Specialist Report Plan	2	Individual or Team					
Participation in class or JigSaw presentations	2	Individual or Team					
Informed Consent Form	2	Individual					
PBL Rubrics #1 and #2 "early PBL" reflection	2	Individual & Team					
Class Participation	2	Individual					
Bioethical Decision-Making Steps 1-3, then 4-6	3	Team					
Bioethical Decision-Making 7-8 (position statement)	3	Individual					
Class Participation	3	Individual					
Laboratory report team	4	Team					
Class Participation	4	Individual					
Technician's report	4	Team					
Imagining Futures	5	Individual or Team					
PBL Rubrics # 1 and #2 "end PBL" reflection	5	Individual & Team					
Class Participation	5	Individual					
Genetics Test	5	Individual					
Alternative Assessment Options	5	Individual or Team					
Other	-						

Students / Teams