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Genetic diseases: challenges and solutions

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BIO-INSPIRED STEM TOPICS FOR ENGAGING YOUNG GENERATIONS
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General topic of the learning path	Bio-technology. Biomanufacturing and Synthetic Biology
Specific name of the learning unit	**Genetic Diseases: Challenges and Solutions**
Age of the target users	14-18 years
Requirements for the learner	• Basic understanding of biology concepts (cells, DNA, genes) • Basic digital skills (using a computer, tablet, or smartphone) • Ability to work individually and collaboratively • Access to the internet and AR-compatible devices • No prior knowledge of genetics, biotechnology, biomanufacturing, or AR content creation required

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Description of the learning unit	This learning unit introduces students to genetic diseases as complex biological challenges caused by changes in DNA and gene function. Learners explore the causes, types, and real-world impact of genetic disorders, as well as the scientific, technological, and ethical challenges related to their diagnosis and treatment. Through bio-inspired thinking and synthetic biology approaches, students discover how natural mechanisms such as DNA repair, gene regulation, and cellular regeneration inspire modern medical solutions, including gene therapy and gene editing. Augmented Reality (AR) is used as a key educational tool to visualize invisible molecular processes and to support the creation of an interactive AR-based quiz, transforming assessment into an active, inquiry-based learning experience.
Subject: Parties involved	Biology, Genetics, Biotechnology, Synthetic Biology, Digital Literacy / Computer Science, Health Education, Ethics
Keywords	Genetic diseases, DNA, genes, mutations, bio-inspiration, synthetic biology, biomanufacturing, gene therapy, gene editing, CRISPR, DNA repair, biotechnology, human health, Augmented Reality (AR), digital assessment
Key qualifications, skills and knowledge that can be acquired	**Qualifications / Attitudes** • Awareness of genetic diversity and health challenges • Responsible and ethical thinking regarding genetic and synthetic biology technologies • Curiosity toward biotechnology and medical innovation **Skills** • Critical thinking and problem-solving • Analysis of cause–effect relationships in genetics and biotechnology • Digital and visual literacy through AR tools • Collaboration, communication, and content creation skills **Knowledge** • Understanding the genetic basis of diseases

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	• Recognition of different types of genetic mutations • Awareness of diagnostic and treatment challenges • Understanding bio-inspired and synthetic biology solutions • Understanding the role of AR in biotechnology education and assessment
Resources and didactic aids used	• Educational texts and scientific articles on genetics, biotechnology, and synthetic biology • Case studies of genetic diseases and medical biotechnologies • Online educational platforms and databases • Augmented Reality tools and applications (Delightex) • AR-compatible devices (smartphones, tablets, computers) • 3D models of DNA, genes, proteins, and cells
Assessment criteria and evaluation	• Active participation in discussions and AR-based activities • Completion of AR-supported exercises related to genetic mechanisms • Scientific accuracy and clarity of the student-created AR quiz • Ability to explain genetic diseases, challenges, and solutions • Reflection on ethical, social, and technological implications of biotechnology and synthetic biology

Introduction

Genetic diseases are conditions caused by changes or errors in our DNA, the biological code that carries instructions for building and maintaining the human body. Some genetic diseases are inherited from parents, while others appear due to spontaneous mutations or environmental influences. These conditions can affect how organs develop, how cells function, and how the body responds to its environment, often creating lifelong health challenges.

In this learning unit, students will explore the causes and consequences of genetic diseases, as well as the scientific and technological challenges involved in understanding, diagnosing, and treating them. By connecting biology with bio-inspired thinking and modern technologies, learners will discover how nature itself offers clues for repairing genetic damage and how innovations such as artificial intelligence, gene analysis, and personalized medicine contribute to new solutions. Through this exploration, students will gain insight into how genetics shapes human health and how science works to turn biological challenges into opportunities for improvement.

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Explore

1. Genetic Diseases: Challenges and Solutions

In this section, students build foundational knowledge about genetic diseases, their causes, and the challenges involved in diagnosing and treating them. This section introduces essential concepts from genetics, molecular biology, and biotechnology, preparing learners for later activities such as AR visualization, modeling, and ethical discussions about emerging medical solutions.

1.1 What Are Genetic Diseases?

A genetic disease is a condition caused by changes in DNA, either through mutations in individual genes or abnormalities in chromosomes. Unlike infectious diseases, genetic disorders are not caused by bacteria or viruses; instead, they are inherited from parents or arise spontaneously during cell division or early development. It is estimated that genetic diseases affect approximately **1 in 25 people worldwide**.

Key characteristics of genetic diseases include:

- They are caused by changes in the DNA sequence (such as point mutations, insertions, or deletions) or by changes in chromosome number or structure.
- They can be **monogenic**, involving a single gene, or **polygenic**, involving multiple genes and environmental factors.
- Their severity varies widely, from mild conditions such as color blindness to life-threatening disorders.

A well-known example is **sickle cell anemia**, where a single nucleotide change in the HBB gene alters the structure of hemoglobin, causing red blood cells to become rigid and sickle-shaped.

1.2 Types of Genetic Mutations

Genetic mutations differ in scale and impact, but all can disrupt how proteins are produced or function.

Mutation Type	Description	Example Disease
Point mutation	Single nucleotide change	Sickle cell (GAG→GTG)
Insertion/Deletion	Added/removed nucleotides	Cystic fibrosis (3bp ΔF508)
Trinucleotide repeat	CAG expansion	Huntington's disease
Chromosomal	Aneuploidy, translocation	Down syndrome (trisomy 21)

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These mutations may result in proteins that are non-functional, partially functional, or harmful to cells.

1.3 Common Genetic Diseases

Genetic diseases can be grouped based on how they arise:

- **Monogenic (Mendelian) diseases** are caused by mutations in a single gene. Examples include cystic fibrosis, Duchenne muscular dystrophy, and hemophilia A.
- **Multifactorial genetic diseases** result from interactions between multiple genes and environmental factors, such as certain metabolic or neurological disorders.
- **Chromosomal disorders**, such as Down syndrome, are caused by extra or missing chromosomes and affect overall development.

Studying these conditions helps scientists understand how specific genetic changes lead to observable biological effects.

1.4 Diagnostic Challenges

Diagnosing genetic diseases is often complex. Many disorders share similar symptoms, and more than 7,000 rare genetic diseases have been identified worldwide. Some conditions appear only later in life, making early detection difficult. Additionally, many people are healthy carriers of genetic mutations without showing symptoms.

Modern diagnostic tools include newborn screening programs, whole-genome sequencing, and emerging AI-based technologies that analyze genetic data and physical features to support earlier and more accurate diagnosis.

1.5 Treatment Challenges

Traditional treatments for genetic diseases usually focus on managing symptoms rather than correcting the underlying genetic cause. While these approaches can improve quality of life, they have important limitations:

- The mutated gene itself remains unchanged.
- Damage to tissues and organs may be irreversible.
- Delivering treatments to specific cells, such as those protected by biological barriers, remains difficult.

These challenges highlight the need for new therapeutic strategies.

1.6 Emerging Solutions

Recent advances in biotechnology offer promising solutions to genetic diseases:

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- **Gene therapy** uses viral vectors to deliver healthy copies of genes into affected cells.
- **CRISPR/Cas9 gene editing** allows scientists to precisely modify DNA, correcting mutations at their source.
- **RNA-based therapies and cell therapies** aim to restore normal protein production or replace damaged cells.

Some of these approaches have already reached clinical use, demonstrating how basic genetic knowledge can lead to real medical breakthroughs.

1.7 Why AR and 3D Modeling Matter

Genetic processes such as mutations, protein folding, and gene editing occur at a microscopic level and cannot be seen directly. Augmented Reality and 3D modeling make these invisible processes visible by allowing students to explore:

- How protein structure changes due to mutations,

- How gene-editing tools modify DNA,
- How chromosomal abnormalities affect cells.

Example: Rotate sickle hemoglobin, simulate editing (https://www.delightex.com/edu/lesson-plans).

Interactive visualization helps students connect abstract genetic concepts with real biological consequences.

Reflection Questions

1- How can a single nucleotide change lead to a serious disease?
2- Why is delivering gene therapy to the correct cells challenging?
Which recent genetic therapy developments do you find most promising, and why?

Execute

2. Bio-Inspired Solutions to Genetic Diseases

In the Execute phase, students move from understanding genetic diseases to actively exploring how nature itself inspires solutions to genetic challenges. Living organisms have evolved powerful mechanisms to protect, repair, and regulate their genetic information. By studying

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these natural processes, students can better understand how modern biotechnology and medicine attempt to mimic, enhance, or adapt them to treat genetic diseases.

2.1 Bio-inspiration in genetic repair and regulation

Nature continuously protects DNA from damage caused by radiation, chemical agents, and replication errors. Cells are not passive systems; they actively monitor and repair their genetic material through highly efficient mechanisms.

Key bio-inspired principles include:

- **DNA repair mechanisms**: Cells use natural repair systems to detect and correct mutations, inspiring modern gene-editing technologies.
- **Selective gene activation**: Not all genes are active at all times; cells regulate gene expression depending on their environment, age, and function.
- **Cell specialization and regeneration**: Stem cells demonstrate how damaged or defective cells can be replaced, inspiring regenerative medicine and cell therapy.
- **Error tolerance and adaptation**: Some organisms tolerate genetic variation and adapt to it, offering insights into resilience and long-term survival.

These natural strategies form the conceptual bridge between biological systems and technological solutions for genetic diseases.

2.2 Practical application through AR-supported learning

Many genetic processes occur at the molecular level and cannot be observed directly. Augmented Reality (AR) allows students to interact with dynamic 3D representations of DNA, genes, proteins, and cells, transforming abstract concepts into tangible learning experiences.

An AR application can support this unit by enabling students to:

- Visualize DNA structure and locate specific genes affected by mutations.
- Observe how mutations alter protein shape and function.
- Simulate natural DNA repair processes and compare them with gene-editing techniques.
- Explore how corrected genes restore normal cellular function.

AR tools provide real-time interaction, rotation, scaling, and step-by-step exploration, supporting inquiry-based and experiential learning.

2.3 Suggested AR-based exercises and activities

- **Exercise 1: DNA Repair – Learning from Nature**
  Students explore an AR model of DNA damage caused by a mutation. They activate natural DNA repair pathways and observe how cells detect and fix errors. Students then

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compare natural repair mechanisms with human-designed gene therapies, identifying similarities and differences.

- **Exercise 2: Protein Structure and Function**
  Using AR, students manipulate 3D protein models to compare healthy and mutated proteins. By rotating and zooming in on molecular structures, learners observe how small genetic changes can lead to large functional consequences, such as protein misfolding.
- **Exercise 3: Gene Editing Simulation**
  Students simulate a gene-editing process inspired by natural DNA repair. They select a faulty gene, apply a virtual "repair tool," and observe the before-and-after effects on gene expression and cell behavior. This exercise emphasizes precision, limits, and ethical considerations.
- **Exercise 4: From Cell to Organism**
  Students follow the effects of a corrected gene from a single cell to tissue and organ level. AR visualization shows how restoring a gene can improve organ function, helping students connect molecular events with real-world health outcomes.
- **Exercise 5: Designing an AR Quiz: Genetic Diseases – Challenges and Solutions**

  In this exercise, students **design and build an interactive AR-based quiz** focused on genetic diseases, their causes, challenges, and possible solutions. Using **Delightex**, learners create a test that combines scientific questions with 3D visualizations of DNA, genes, proteins, and cells.

  Students develop quiz scenarios, write multiple-choice or decision-based questions, and connect each question to AR models that demonstrate genetic mutations, protein misfolding, and gene-editing outcomes. The quiz includes a **points and lives system**, where correct answers earn points and incorrect choices reduce lives, providing immediate feedback and encouraging careful reasoning.

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2.4 Global inspiration and real-world relevance

Similar AR-supported activities are used worldwide in genetics education to help students:

- Understand complex molecular processes through visualization.
- Develop problem-solving skills by testing different intervention strategies.
- Engage collaboratively by discussing outcomes and design choices.

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- Reflect on ethical and societal implications of genetic technologies.

By combining bio-inspired principles with AR-based hands-on activities, students experience genetics not as abstract theory, but as an evolving field where natural mechanisms inspire innovative medical solutions.

Enhance

3. AR for Deeper Learning and Engagement

Genetic diseases involve complex and invisible biological processes that occur at the molecular and cellular level. Concepts such as DNA mutations, protein misfolding, gene regulation, and gene editing are difficult for students to fully understand using traditional textbooks or static images alone. Augmented Reality (AR) significantly enhances the learning process by transforming these abstract processes into interactive, visual, and experiential learning experiences.

3.1 How AR deepens understanding of genetic diseases

AR allows students to visualize genetic mechanisms that cannot be observed directly. By interacting with 3D models of DNA, genes, chromosomes, and proteins, learners can explore how small genetic changes lead to major biological consequences. This spatial and dynamic visualization helps students:

- Understand cause–effect relationships between mutations and disease symptoms
- Connect molecular-level events to cellular, tissue, and organism-level outcomes
- Grasp complex processes such as gene editing and DNA repair step by step

Instead of memorizing definitions, students actively explore how genetic diseases develop and how modern solutions attempt to address them.

3.2 Making complex concepts accessible and engaging

AR reduces cognitive overload by breaking down complex genetic processes into manageable visual stages. Learners can rotate, zoom, and isolate specific structures, allowing them to focus on one concept at a time. This approach supports diverse learning styles and makes advanced biotechnology topics accessible to secondary school students.

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Engagement is further increased through interaction and exploration. AR-based tasks encourage curiosity, experimentation, and discussion, turning students into active participants rather than passive recipients of information.

3.3 Example of a useful AR tool (Deligtex)

Delightex is a particularly effective tool for genetics education. It enables students not only to explore AR content but also to create their own interactive learning materials. Through Delightex, learners can design AR-based tests, simulations, and visual explanations of genetic diseases, reinforcing understanding through creation and reflection.

3.4 Practical suggestions for integrating AR into the learning experience

To strengthen students' connection to the topic and support effective knowledge building, AR can be integrated in the following ways:

- **Exploration-based learning**: Use AR to introduce genetic mutations and visualize their effects before formal explanations.
- **Simulation and experimentation**: Allow students to test different gene-editing or repair scenarios and observe outcomes in a risk-free environment.
- **AR-based assessment**: Replace or complement traditional tests with AR-based quizzes or student-created AR tests that evaluate understanding through application.
- **Collaborative learning**: Encourage students to work in groups to design, test, and improve AR-based learning scenarios, fostering communication and peer learning.
- **Reflection and ethics**: Use AR scenarios as a starting point for discussions on ethical, social, and technological challenges related to genetic interventions.

By integrating AR into genetics education, learning becomes more meaningful, memorable, and motivating. AR supports deeper conceptual understanding, enhances engagement, and empowers students to connect biological theory with real-world medical challenges. In this unit, AR functions not only as a visualization tool but as a bridge between biology, technology, and responsible innovation.

Conclusion

This learning unit explored genetic diseases as complex biological challenges caused by changes in DNA and gene function. Students developed a foundational understanding of how genetic mutations arise, why they are difficult to diagnose and treat, and how nature itself offers

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inspiration for repairing and regulating genetic information. By examining bio-inspired mechanisms such as DNA repair, gene regulation, and cellular regeneration, learners connected biological theory with real-world medical challenges.

The integration of Augmented Reality played a key role in transforming abstract genetic processes into accessible and engaging learning experiences. Through interactive 3D visualizations and the creation of AR-based tests, students moved beyond passive learning to actively explore, apply, and communicate their knowledge. This approach strengthened conceptual understanding, encouraged critical thinking, and highlighted the ethical and societal dimensions of emerging genetic technologies. Ultimately, the unit demonstrates how combining biology, bio-inspiration, and immersive digital tools can empower students to better understand genetic diseases and envision innovative solutions for the future.

Phase	Description
Explore	- Research and Discovery: Students explore the basic principles of genetics, including DNA structure, genes, mutations, and chromosomal abnormalities. They investigate how genetic diseases arise, their global prevalence, and real-life examples to understand the biological and social relevance of the topic.
	- Content Development: Teachers provide accessible learning materials such as short texts, visuals, infographics, and introductory videos explaining genetic diseases, mutation types, and biological consequences. Content focuses on building conceptual understanding rather than clinical detail.
	- Needs Analysis: Students' prior knowledge of genetics and biotechnology is assessed through discussion or short diagnostic questions to identify misconceptions and adapt the learning pace and depth accordingly.
Execute	- Curriculum Implementation: Bio-inspired concepts such as DNA repair, gene regulation, and cellular regeneration are linked to modern genetic solutions. Lessons guide students from understanding natural mechanisms to applying this knowledge through practical AR-supported tasks
	- Interactive Exercises: Students participate in AR-based activities exploring DNA damage, protein malfunction, gene editing, and biological outcomes. A key activity involves **designing and creating an AR-based test in Delightex**, transforming scientific knowledge into interactive assessment content.

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	- Feedback Collection: Peer review, group discussions, and teacher feedback are used to evaluate students' AR exercises and test designs, supporting reflection and iterative improvement.
Enhance	- AR Integration: Augmented Reality is used to visualize invisible genetic processes, such as mutations, protein folding, and gene correction, making abstract biological mechanisms tangible and understandable.
	- Interactive Learning: Students actively manipulate 3D models, simulate biological processes, and explore cause–effect relationships, shifting learning from passive reception to experiential exploration.
	Gamified Content: - **Points and Badges**: Students earn points and digital badges for completing AR exercises, correctly identifying mutations, and designing accurate AR test questions. - **Leaderboards**: Optional leaderboards can be used to encourage motivation and friendly competition based on task completion or quiz performance. - **Quests and Levels**: Learning activities are structured as progressive quests, moving from basic genetic concepts to advanced bio-inspired solutions and test creation. - **Rewards for Exploration**: Bonus points or hidden content are unlocked when students explore additional AR models or investigate advanced genetic scenarios. - **Collaborative Gamified Tasks**: Students work in teams to co-design AR test questions, review peer work, and collectively solve genetic challenges.
	AR-Based Assessments: Assessment is conducted through AR-based quizzes and student-created AR tests, allowing learners to demonstrate understanding by applying knowledge, visualizing biological processes, and explaining genetic challenges and solutions in an interactive format.

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AI-based tools (ChatGPT, Perplexity AI, and NotebookLM) were used to support content drafting, research synthesis, and organization of source materials during the development of this learning unit:

- OpenAI. *ChatGPT* (large language model). https://chat.openai.com
- *Perplexity AI* [AI-powered search and research assistant]. https://www.perplexity.ai
- Google *NotebookLM* [AI-powered research and note-taking tool]. https://notebooklm.google.com

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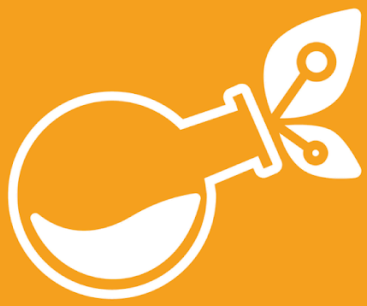

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