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Understanding Genetic Disorders: From DNA to Disease

BIOS4YOU AR 2.0

BIO-INSPIRED STEM TOPICS FOR ENGAGING YOUNG GENERATIONS THANKS TO THE USE OF AUGMENTED REALITY

Project Number: 2023-1-DE03-KA220-SCH-000126516

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General topic of the learning path	Human Genome Project and Genomic Medicine
Specific name of the learning unit	**Understanding Genetic Disorders – From DNA to Disease**
Age of the target users	14-18 years
Requirements for the learner	Basic understanding of DNA and inheritance Ability to use a mobile phone or tablet Interest in health, science, and real-life problems Willingness to discuss ethical questions
Description of the learning unit	This learning unit helps students understand genetic disorders in a clear and engaging way. Learners explore how changes in DNA (mutations) can lead to disease, how genetic disorders are inherited, and how modern medicine identifies and manages them. The unit is built around Delightex Augmented Reality (AR) environments, where students interact with 3D models of DNA, genes, chromosomes, and proteins. Abstract genetic concepts become easier to understand through visual and hands-on exploration. Instead of traditional quizzes, assessment is integrated into AR-based tasks, group discussions, and reflection activities carried out inside Delightex. Ethical issues such as genetic testing, data privacy, and responsibility in medicine are addressed throughout the unit, encouraging critical thinking and informed discussion.
Subject: Parties involved	Biology teachers, Students, AR / ICT coordinators, Healthcare professionals (e.g. genetic counsellors), School technical support staff

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Keywords	DNA, mutation, genetic disorder, chromosome, inheritance, genetic testing, AR in education, bioethics, personalised medicine
Key qualifications, skills and knowledge that can be acquired	Students will develop: a clear understanding of how genes and mutations can cause disease, the ability to distinguish between different types of genetic disorders, practical experience using AR tools to visualise DNA, genes, and proteins, basic bioethical reasoning related to genetic testing and health data, critical thinking and communication skills through group work and discussion, digital skills through the use of AR and interactive learning technologies
Resources and didactic aids used	Chapter texts and figures based on NHGRI resources and Wallis (2018) Augmented Reality applications: MoleculAR, Genome AR Printable case studies and reflection tasks Interactive worksheets and family tree charts Tablets or projectors for AR activities Classroom posters summarising types of genetic disorders
Assessment criteria and evaluation	Student learning is assessed through: - successful completion of AR-based tasks (e.g. identifying gene mutations) - clear and logical participation in group discussions on diagnosis and ethics - short reflective writing on the role of genomics in healthcare - final group or individual presentation using AR visualisations (e.g. protein models or inheritance patterns) - a short quiz checking understanding of genetic disorder types and testing methods

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Introduction:

Genes are biological instructions that control how our bodies grow, develop, and function. They influence everything from eye colour to how our immune system fights illness. Genes are made of DNA, and each gene carries specific information needed to produce proteins that keep the body working properly (Wallis, 2018).

Sometimes these genetic instructions change. These changes are called mutations. While some mutations have no effect, others can disrupt how a protein works or stop it from working completely. When this happens, it can lead to a genetic disorder—a health condition caused by changes in DNA (National Human Genome Research Institute [NHGRI], 2024).

In this learning unit, students explore how genetic disorders develop and why they occur. They learn about different types of genetic disorders, including single-gene disorders, chromosomal disorders, and complex conditions influenced by both genes and the environment. The unit also explains how genetic disorders are inherited and how modern medicine uses genetic testing to diagnose diseases and assess health risks (Wallis, 2018).

In addition, students examine important ethical questions connected to genetic science. As genetic technologies become more powerful and accessible, society must consider issues such as genetic privacy, informed consent, and responsible use of genetic information. These questions are especially important in an era of personalised medicine, where treatments can be tailored to a person's genetic profile (NHGRI, 2024).

By the end of this unit, students gain a clearer understanding of how genes affect health and disease. This knowledge provides a strong foundation for further studies in biology, healthcare, and biotechnology, and helps students become informed citizens who can thoughtfully engage with scientific and ethical discussions in modern society.

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1: Explore

What Are Genetic Disorders?

Genes are like instruction manuals inside every cell of our body. They tell cells how to build proteins — molecules that help us grow, stay healthy, and fight disease (Wallis, 2018). Genes are made of DNA, and their instructions are written in a specific order using four letters: A, T, C, and G.

Sometimes a small change happens in DNA. This change is called a mutation. A mutation can affect how a protein is made or stop the protein from working correctly. When a gene does not function as it should, it can lead to a genetic disorder (National Human Genome Research Institute [NHGRI], 2024).

Mutations can be inherited from one or both parents, or they can happen spontaneously. Some mutations have no effect, but others interfere with normal body processes. In some cases, even a small change in DNA can cause a serious disease, depending on the role of the affected gene (Wallis, 2018).

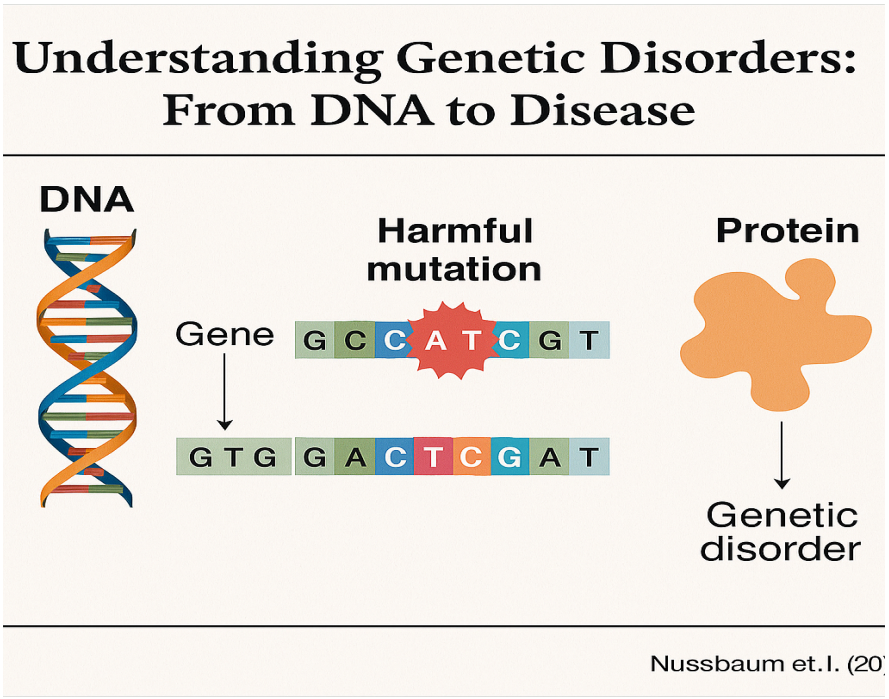

Figure 1. A mutation in DNA can change a protein and lead to disease (generated by AI).

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Types of Genetic Disorders

There are three main types of genetic disorders, based on how they develop and are inherited.

Single-Gene Disorders

Single-gene disorders occur when one specific gene is mutated. The mutation may be passed down in families or appear by chance.

Examples include:

- Cystic fibrosis – a disorder that causes thick, sticky mucus in the lungs and digestive system (NHGRI, 2024).
- Sickle cell disease – a condition that changes the shape of red blood cells, making it harder for them to carry oxygen.

These disorders follow clear inheritance patterns, such as dominant or recessive, which makes them easier to trace using family trees (Wallis, 2018).

Chromosome Disorders

Chromosome disorders occur when a whole chromosome is missing, extra, or rearranged. These changes often lead to serious developmental and health problems.

Examples include:

- Down syndrome – caused by having three copies of chromosome 21.
- Turner syndrome – affects females when one of the X chromosomes is missing or altered (NHGRI, 2024).

Such conditions are often identified through prenatal screening or karyotyping, which allows doctors to examine chromosomes.

Complex Disorders

Complex disorders are caused by a combination of genetic and environmental factors, such as lifestyle, diet, stress, or pollution.

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Examples include:

- diabetes
- heart disease
- autism

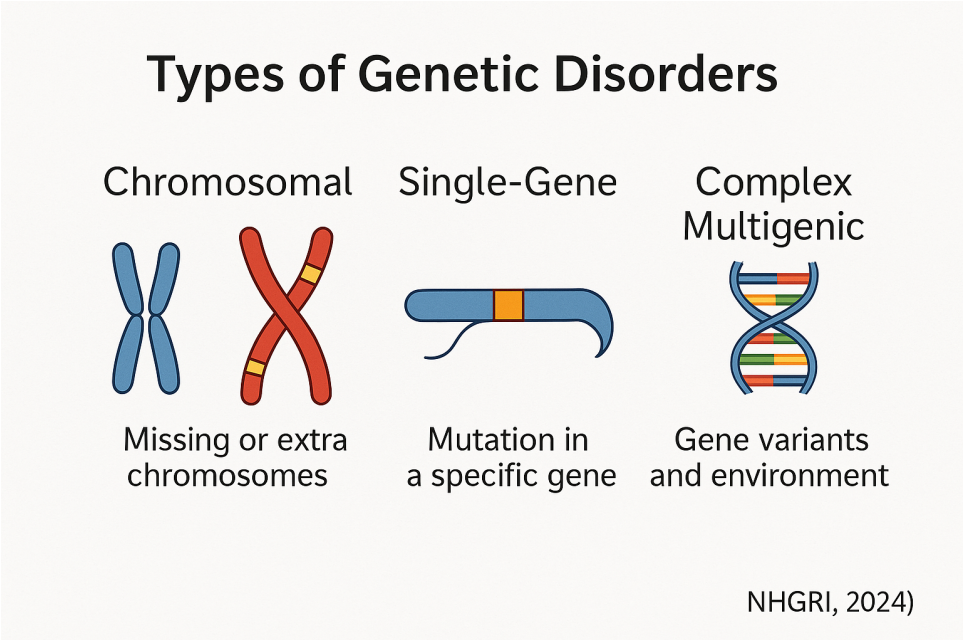

Figure 2. Types of genetic disorders by origin and inheritance (generated by AI).

These disorders do not follow simple inheritance patterns. Scientists study large populations to identify genetic markers that increase the risk of developing these conditions (Wallis, 2018).

Cancer and Genes

Cancer is often linked to mutations in genes that control cell growth and division, although not all cancers are inherited.

There are two important types of genes involved:

- Oncogenes – when activated incorrectly, they cause cells to grow too fast.
- Tumour suppressor genes – normally slow down cell growth; when damaged, they may allow cancer to develop.

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Mutations in genes such as BRCA1 and BRCA2 can significantly increase the risk of breast and ovarian cancer. For this reason, genetic testing is often recommended for people with a family history of these cancers (NHGRI, 2024).

Diagnosing Genetic Disorders

Thanks to large research projects like the Human Genome Project, scientists can now analyse a person's DNA to check for genetic disorders or health risks. This process is known as genetic testing.

Genetic testing is used to:

- diagnose diseases
- identify the cause of symptoms
- guide treatment decisions
- assess family risk (Wallis, 2018)

Modern methods such as whole genome sequencing and exome sequencing allow doctors to examine thousands of genes at the same time. Genetic counselling is also provided to help patients understand test results and make informed decisions.

Before any genetic test is performed, patients must give informed consent, meaning they understand the purpose, risks, and possible outcomes of the test (NHGRI, 2024).

Why This Matters

Understanding genetic disorders helps people make informed decisions about health and medical care. At the same time, it raises important ethical questions. For example:

- Should employers have access to genetic information?
- Can health insurance companies use genetic data to limit coverage?

These issues are part of bioethics, the study of what is right and wrong in science and medicine (Wallis, 2018). Learning about DNA and disease empowers students to take part in informed discussions about science, technology, and society.

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Conclusion

Genetic disorders offer valuable insight into how the human body works and what can happen when genetic instructions are altered. By learning how mutations affect DNA, understanding different types of genetic disorders, and exploring how genetic testing is used in modern medicine, students become better prepared to think critically about biology and personal health.

As genetic technologies continue to advance, ethical decision-making will become increasingly important. Understanding both the scientific and societal impact of genetics helps students navigate life in a modern genomic age.

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2: Execute

In this section, students move from theory to active learning. They apply what they have learned about DNA, mutations, and genetic disorders through hands-on activities, Augmented Reality (AR) exploration, and collaborative tasks inside Delightex.

The main goal of this phase is to help students understand how genetic changes affect the human body by seeing and exploring these processes in a visual and interactive way (Wallis, 2018).

Role of AR in Achieving Learning Objectives

Augmented Reality plays a central role in the Execute phase. Using applications such as MoleculAR and Genome AR, students interact with 3D models of DNA, genes, chromosomes, and proteins. These tools make abstract molecular structures visible and easier to understand (NHGRI, 2024).

With AR, students can:

- observe the DNA double helix in three dimensions
- explore how a mutation changes a gene or protein
- compare healthy and mutated genetic structures
- connect genetic changes to real genetic disorders

This type of spatial and visual interaction helps learners better understand how mutations can lead to disease (Wallis, 2018).

AR-Based Learning Activities

DNA Mutation Exploration (AR Activity)

Students use **MoleculAR** to explore a DNA model and observe how a mutation changes the genetic code. By comparing a normal gene with a mutated one, students can see how a small change in DNA may affect protein function.

Task for students:

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- Explore the DNA model in AR
- Identify the location of the mutation
- Discuss how the mutation could affect the body

This activity supports understanding of the relationship between DNA, proteins, and disease (NHGRI, 2024).

Inheritance Patterns in Families

Using **Genome AR** and simple family tree charts, students explore how genetic disorders are inherited. They trace dominant and recessive traits and predict the likelihood of a disorder appearing in future generations.

Task for students:

- Analyse a family tree
- Identify carriers and affected individuals
- Explain the inheritance pattern

This task helps students connect theoretical inheritance rules to real-life genetic conditions (Wallis, 2018).

Case Study: Diagnose the Disorder

Students are presented with a short patient scenario inside the Delightex environment. The case includes symptoms and basic genetic information related to a genetic disorder.

Example scenario:
A patient experiences breathing problems and frequent lung infections caused by a mutation in one gene.

Students:

- analyse the symptoms
- explore AR models of the affected gene or protein
- identify the most likely type of genetic disorder

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Case-based learning encourages problem-solving and supports deeper understanding of genetic diseases (NHGRI, 2024).

Ethics in Action

Modern genetics also raises important ethical questions. During this phase, students explore short ethical scenarios related to genetic testing, data privacy, and responsible use of genetic information.

Discussion questions:

- Should everyone be tested for genetic disorders?
- Who should have access to genetic test results?
- How can genetic data be protected?

These discussions are linked to bioethical principles and help students reflect on the social impact of genetic technologies (Wallis, 2018; NHGRI, 2024).

Feedback and Support

Throughout the Execute phase, students receive feedback through:

- teacher-guided discussions
- peer feedback during group activities
- short reflection prompts inside Delightex

The focus of this phase is on understanding and application, rather than memorisation. By actively working with AR tools and real-life examples, students are prepared for the reflective tasks in the Enhance phase.

Global Relevance and Student Engagement

The learning activities in this unit reflect modern approaches already used in schools and science centres around the world. Interactive DNA and genetics applications are currently used in genetics education in countries such as Finland, the United States, and Japan, where digital and AR-based learning is integrated into science curricula (NHGRI, 2024).

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Teachers report that student engagement increases when abstract biological concepts become visual and interactive. AR allows learners to move, explore, and manipulate models, which supports deeper conceptual understanding compared to traditional textbook-based learning (Wallis, 2018).

By using AR elements in this unit, students become active participants in scientific exploration rather than passive receivers of information. They do not only *see* genetic mutations but *experience* how changes in DNA can affect human health. This approach prepares students for informed discussions about modern medicine, genetics, and ethical decision-making in real-world contexts (NHGRI, 2024).

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3: Enhance

In this phase, learning is deepened through Delightex as the main interactive environment. All activities take place inside a Delightex scene, where Augmented Reality (AR) elements, information panels, and reflection tasks are combined into one coherent learning experience.

For students aged 14–18, topics such as DNA mutations, inheritance patterns, and gene–protein interactions can be difficult to understand through text alone. Delightex helps bridge this gap by integrating 3D AR models, guided tasks, and reflection prompts into a single immersive space, allowing students to actively explore complex genetic concepts (Wallis, 2018).

Deepening Understanding of Complex Concepts

Within the Delightex scene, students interact with AR elements that support understanding of genetic disorders, including:

- the double-helix structure of DNA and its role in replication and transcription,
- point mutations and their effects on protein synthesis,
- chromosomal changes such as trisomy or deletions linked to disorders like Down syndrome or Turner syndrome,
- the pathway from a DNA mutation to physical symptoms caused by faulty proteins.

These concepts are presented through embedded AR models and visual panels rather than static diagrams. Using tools such as MoleculAR and Genome AR integrated into the Delightex workflow, students can rotate, zoom in, and explore genetic structures step by step. This supports spatial understanding and helps learners see how small genetic changes can lead to significant health effects (NHGRI, 2024).

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Active Learning and Engagement inside Delightex

Delightex transforms the Enhance phase into an active learning mission, not a passive review. Instead of watching videos or reading long texts, students complete guided tasks directly in the Delightex environment.

Learners engage through:

- interaction with AR models placed inside the scene,
- short Delightex information panels guiding observation and thinking,
- mission-style tasks (e.g. analysing a mutation or solving a case study),
- peer discussion based on prompts displayed in the scene.

This approach supports intrinsic motivation and encourages students to take ownership of their learning. Active participation has been shown to improve understanding of complex biological processes, especially in genetics education (Wallis, 2018).

Delightex Tools and Learning Structure

In this phase, Delightex is used to combine multiple learning elements:

- AR objects (DNA, genes, chromosomes, proteins) for exploration and observation
- Information panels explaining key ideas in short, student-friendly language
- Case study panels presenting real-life genetic scenarios
- Reflection panels guiding ethical discussion and personal response

External AR tools such as MoleculAR, Genome AR, or Merge EDU are used as content sources, while Delightex remains the main platform that structures the learning process and assessment.

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Connecting Genetics to Real-World Contexts

Delightex scenes also support real-world application of genetic knowledge. Through AR-supported scenarios, students explore roles such as genetic counsellors or patients and reflect on how genetic information is used in healthcare.

For example:

- students analyse the impact of a BRCA gene mutation using AR models and discussion prompts,
- ethical panels raise questions about genetic testing, privacy, and data protection.

These tasks help learners understand how genomic science affects individuals and society, supporting interdisciplinary learning across biology, ethics, and technology (NHGRI, 2024).

Inclusive and Personalised Learning with Delightex

Delightex supports inclusive learning by offering:

- visual explanations that support learners with different language backgrounds,
- short, layered content instead of long texts,
- self-paced exploration of AR elements within the scene.

Students can revisit panels and models as needed, which supports confidence and independent learning.

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Conclusion:

In this learning unit, students explored the fundamentals of genetic disorders, including what they are, how they are inherited, how they affect the human body, and how they are diagnosed. Through structured lessons and interactive tasks, learners developed a clear understanding of how mutations in DNA can lead to different diseases, such as single-gene disorders, chromosomal disorders, and complex conditions influenced by both genetic and environmental factors (Wallis, 2018).

A key feature of this unit was the use of Delightex as the main learning environment, where Augmented Reality (AR) elements, information panels, and learning tasks were combined into one coherent learning path. AR tools such as MoleculAR, Genome AR, and Merge EDU were used within Delightex to support exploration and understanding. These tools allowed students to visualise DNA structures, observe the effects of mutations, and explore chromosome abnormalities in an interactive way. As a result, abstract genetic concepts became more accessible and easier to understand (NHGRI, 2024).

The unit also supported the development of important 21st-century skills. Through AR-based tasks, discussions, and collaborative activities, students practised ethical reasoning, digital literacy, communication, and teamwork. By exploring real-life applications such as genetic testing, disease prediction, and bioethical decision-making, learners were able to connect scientific knowledge to current societal issues, including personalised medicine and data privacy (Green & Guyer, 2015).

Overall, this learning unit encouraged students to think critically and act as informed learners and responsible citizens. By working inside the Delightex environment, students were not only learning about genetics but also experiencing how genomic knowledge is used in modern science and healthcare. As genetic technologies continue to develop, learners are now better prepared to take part in informed discussions and future challenges related to science, medicine, and society (Collins et al., 2003).

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Phase	Description
Explore	- Research and Discovery: Students are introduced to key concepts such as DNA, genes, mutations, and genetic disorders through visual materials, real-life examples, and short explanatory texts.
	- Content Development: Teachers use student-friendly resources (e.g. NHGRI fact sheets) supported by diagrams, karyotypes, and infographics.
	- Needs Analysis: Diagnostic tasks help identify students' prior knowledge, allowing teachers to adjust the depth of content and provide appropriate support.
Execute	- Curriculum Implementation: Lessons combine teacher guidance, discussion, and AR demonstrations using Delightex with integrated tools such as MoleculAR and Merge EDU.
	- Interactive Exercises: Students complete AR-based tasks including mutation tracing, virtual karyotype analysis, and ethical scenario discussions. Group work and worksheets support collaboration and application of knowledge.
	- Feedback Collection: Learning is supported through peer feedback, teacher feedback, short quizzes, and reflective prompts inside Delightex.
Enhance	- AR Integration: Delightex is used as the core environment where students interact with 3D models of DNA, chromosomes, and mutations. These tools make invisible biological structures visible and interactive.
	- Interactive Learning: Learners complete mission-based tasks such as decoding mutations, analysing virtual patient cases, and explaining genetic mechanisms using AR models.
	Gamified Content: **Points and badges:** Students earn digital badges for completing AR tasks and challenges (e.g. *DNA Detective*, *Chromosome Hero*). **Leaderboards:** Visual progress indicators support motivation and friendly competition. **Quests and levels:** Learning is structured as a journey where new challenges are unlocked as understanding deepens. **Rewards for exploration:** Optional AR content is unlocked through additional exploration and reflection. **Collaborative gamified tasks:** Group challenges include solving genetic mystery cases and creating AR-based posters explaining a specific genetic disorder.
	AR-Based Assessments: Assessment is integrated directly into Delightex and includes: analysing a genetic disorder using AR-supported case studies, completing a virtual Punnett square or inheritance task, explaining mutation effects using 3D genetic models, a final short quiz supported by AR visualisations.

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