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Decoding the Human Genome

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

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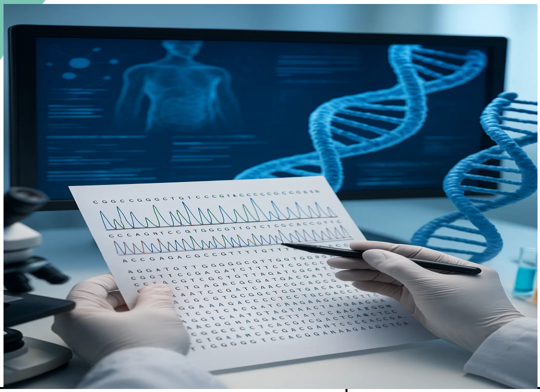

Figure 1. Generated by AI

General topic of the learning path	Human Genome Project and Genomic Medicine
Specific name of the learning unit	Decoding the Human Genome
Age of the target users	14-18 years
Requirements for the learner	Basic knowledge of DNA structure and gene expression. Interest in biology, genetics, or biotechnology. Ability to use digital devices and interact with augmented reality (AR) content.
Description of the learning unit	This learning unit introduces students to the ENCODE Project and the functional elements of the human genome through an interactive and inquiry-based learning approach supported by augmented reality (AR). Students explore key genomic concepts such as coding and noncoding DNA, regulatory regions, gene expression, and chromatin structure. Using **Assemblr EDU** as the main AR platform, learners interact with layered digital models that visualize genome organization and gene regulation processes. AR-based exploration is combined with guided discussions, vocabulary activities, digital quizzes, and collaborative tasks. The unit culminates in an AR-based mini-project or structured AR challenge. The learning unit aims to deepen students' understanding of genome complexity, highlight the importance of noncoding regulatory elements, and emphasize the role of large-scale collaborative research projects such as ENCODE in modern genetics and genomic medicine.
Subject: Parties involved	Biology / Life Sciences; ICT / Digital Competence; Science Teachers; AR platform used: Assemblr EDU
Keywords	Human Genome, ENCODE Project, Functional DNA Elements, Noncoding RNA, Gene Regulation, Chromatin, AR in Education, Bioinformatics, Genomics, Science Learning
Key qualifications, skills and knowledge that can be acquired	Understanding the structure and function of the human genome. Knowledge of coding and noncoding regions and their regulatory roles. Familiarity with genomic research approaches and technologies (e.g., ChIP-seq, RNA-seq). Ability to interpret scientific and visual genomic

	data. Digital literacy through structured AR exploration. Teamwork and scientific communication skills. Critical thinking related to genetic research and its applications in medicine and biotechnology.
Resources and didactic aids used	Adapted scientific texts and summaries. Assemblr EDU augmented reality application. Mobile devices (smartphones or tablets) for AR interaction. Digital or printable AR markers (if required). 3D models of DNA, regulatory regions, and chromatin structures. Interactive worksheets and observation tables. Online quiz tools for formative assessment (e.g., Gimkit)
Assessment criteria and evaluation	Active participation in AR-based exploration and group tasks. Completion of observation tables and reflection questions. Accuracy in explaining genomic elements and their regulatory functions. Results of formative assessments (vocabulary and concept checks). Quality of final AR-based mini-project or structured AR task. Peer feedback and teacher evaluation

Introduction

In the 21st century, the decoding of the human genome has become one of the most significant scientific achievements in biology and medicine. The completion of the Human Genome Project provided researchers with a reference sequence of human DNA, making it possible to study the genetic basis of human development, physiology, and disease. However, while the project delivered a complete sequence, it also raised important new questions. One of the most striking issues was how the relatively small number of human protein-coding genes—approximately 21,000—could account for the remarkable biological complexity of the human organism (Claverie, 2001; Lander et al., 2001).

This unexpected discovery challenged earlier assumptions that organismal complexity is directly related to the number of protein-coding genes. Humans were found to have a similar number of such genes as much simpler organisms, such as the nematode *Caenorhabditis elegans* (Claverie, 2001). As a result, scientists began to shift their attention toward the large portions of the genome that do not encode proteins. These regions, previously referred to as “junk DNA,” were increasingly recognized as functionally important for regulating gene activity and shaping biological complexity.

To address these questions, the ENCODE (Encyclopedia of DNA Elements) Project was launched in 2003 by the National Human Genome Research Institute (NHGRI). The primary goal of the ENCODE Project was to identify and characterize all functional elements within the human genome, including both protein-coding sequences and noncoding regions (ENCODE Project Consortium, 2004). Rather than focusing only on genes, ENCODE aimed to understand how the genome operates as a dynamic and highly regulated system.

This learning unit introduces students aged 14–18 to the core ideas of genomics and functional DNA through an interactive and inquiry-based learning approach. By combining current scientific research with augmented reality (AR), students explore the hidden regulatory layers of the human genome. These include noncoding RNAs, regulatory DNA elements, chromatin modifications, and mechanisms of transcriptional regulation. The initial pilot phase of the ENCODE Project focused on approximately 1% of the human genome (around 30 megabases) and revealed widespread transcriptional activity, including many previously unknown noncoding RNAs and regulatory signals (ENCODE Project Consortium, 2007).

Through AR-based learning stations and digital models, students are able to visualize biological processes that are normally invisible to the naked eye. Interactive three-dimensional models allow learners to explore chromatin structure, observe how regulatory elements interact with genes, and understand how gene expression differs between cell types. Students are also introduced to key genomic technologies such as chromatin immunoprecipitation followed by sequencing (ChIP-seq), DNase I hypersensitive site sequencing (DNase-seq), and RNA sequencing (RNA-seq). These tools help scientists analyze genome activity and identify functional DNA elements (Landt et al., 2012; Djebali et al., 2012; Gerstein et al., 2012).

Research generated by the ENCODE Project has demonstrated that regulatory elements such as promoters and enhancers work together to fine-tune gene expression in a cell-type-specific manner. These interactions depend strongly on chromatin organization and histone modifications, highlighting the importance of genome structure in gene regulation (Kundaje et al., 2012). Such findings have reshaped scientific understanding of how genetic information is controlled within cells.

The project has also emphasized the relevance of noncoding regions in human health and disease. By integrating ENCODE data with genome-wide association studies (GWAS), researchers have been able to functionally annotate a large proportion of disease-associated genetic variants. Many of these variants are located in regulatory regions rather than in protein-coding genes, suggesting that changes in gene regulation can play a critical role in disease development (Schaub et al., 2012; Boyle et al., 2012).

By the end of this learning unit, students will not only gain foundational knowledge in molecular biology and bioinformatics but also develop digital literacy, teamwork, and scientific communication skills. The unit is designed to foster curiosity, critical thinking, and an appreciation for the complexity of the human genome. At the same time, it highlights the collaborative nature of modern scientific research and the importance of large-scale international projects such as ENCODE. While some DNA sequences are directly involved in coding for proteins, a significant portion of the genome consists of noncoding regions that perform essential regulatory and structural roles.

EXPLORE: From Genes to Complexity

What is the Human Genome?

The human genome is the complete set of genetic instructions encoded in DNA. DNA is composed of four types of nucleotides: adenine (A), thymine (T), cytosine (C), and guanine (G). These nucleotides are arranged in a specific sequence of more than three billion base pairs. Together, they store the information required to direct all biological processes in the human body, including growth, development, metabolism, and response to the environment (Lander et al., 2001).

Deoxyribonucleic acid (DNA) is organized into structures called chromosomes, which are located in the nucleus of each cell. Each chromosome contains many genes. A gene is a defined sequence of nucleotides that usually encodes a functional product, most commonly a protein. Proteins perform a wide range of essential functions in cells, such as building cellular structures, catalyzing biochemical reactions, and transmitting signals between cells.

The completion of the Human Genome Project in 2001 provided the first comprehensive overview of human genetic information. One of its most unexpected findings was that the human genome contains approximately 21,000 protein-coding genes. This number is surprisingly similar to that of much simpler organisms, such as the nematode *Caenorhabditis elegans* (Claverie, 2001). This discovery challenged the long-standing assumption that organismal complexity is primarily determined by the number of protein-coding genes.

As a result, scientists began to investigate other sources of biological complexity within the genome. Attention shifted toward the large proportion of DNA that does not encode

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proteins. These regions, referred to as noncoding DNA, make up the majority of the human genome. Although they do not directly produce proteins, they play essential roles in regulating gene expression and maintaining genome structure (ENCODE Project Consortium, 2007).

Noncoding regions include a wide range of functional elements. Among these are promoters, which mark the starting point of transcription; enhancers, which increase the level of gene expression; silencers, which repress transcription; and insulators, which control interactions between regulatory elements and genes. In addition, the genome contains various types of noncoding RNA molecules, such as long noncoding RNAs (lncRNAs) and microRNAs (miRNAs). These molecules influence gene regulation by affecting chromatin structure, RNA stability, and translation.

Together, protein-coding genes, regulatory DNA elements, and noncoding RNAs form a highly coordinated regulatory system. This system allows the same genome to generate a wide diversity of cell types, such as neurons, muscle cells, and immune cells, each with distinct functions. It also enables cells to respond to developmental signals and environmental changes. Understanding this regulatory complexity is fundamental to explaining human development, adaptation, and disease.

What is the ENCODE Project?

The Encyclopedia of DNA Elements (ENCODE) Project was launched in 2003 by the National Human Genome Research Institute (NHGRI). Its primary goal is to identify and characterize all functional elements in the human genome, including those that do not code for proteins (ENCODE Project Consortium, 2004).

To achieve this, the ENCODE Project uses a wide range of experimental and computational methods. These include chromatin immunoprecipitation followed by sequencing (ChIP-seq), RNA sequencing (RNA-seq), and DNase I hypersensitive site sequencing (DNase-seq). These technologies allow scientists to study gene expression, transcription factor binding, and chromatin accessibility across many different human cell types (Landt et al., 2012; Djebali et al., 2012).

The project began with a pilot phase that focused on approximately 1% of the human genome. This phase was designed to test experimental methods and data analysis strategies. After its success, the project expanded to cover the entire genome. The ENCODE Consortium generated thousands of datasets that describe genes, transcripts, chromatin structure, transcription factor binding sites, and other regulatory features.

One of the most important discoveries of the ENCODE Project is that a large proportion of the human genome is transcribed into RNA, even though only a small fraction codes for proteins. Researchers identified thousands of regulatory regions, including promoters, enhancers, and insulators. These elements influence gene expression in a cell-type-specific manner and are closely linked to chromatin structure and histone modifications (Gerstein et al., 2012; Kundaje et al., 2012).

The ENCODE Project also highlighted the importance of noncoding RNAs. Thousands of long noncoding RNAs were identified, many of which appear to have regulatory functions. These RNAs are often expressed in specific tissues and play roles in transcriptional regulation, chromatin remodeling, and RNA processing (Djebali et al., 2012).

In addition, ENCODE research demonstrated that many genetic variants associated with human diseases are located in noncoding regulatory regions rather than within protein-coding genes. By integrating ENCODE data with genome-wide association studies (GWAS), scientists were able to functionally annotate many disease-associated variants (Schaub et al., 2012; Boyle et al., 2012). This finding has significantly improved our understanding of how changes in gene regulation contribute to human health and disease.

Main Discoveries

Research conducted by the ENCODE Consortium has shown that a large proportion of the human genome is transcribed into RNA, even though only a small fraction of these transcripts encode proteins. This finding revealed that transcription is far more widespread than previously believed. Thousands of regulatory regions have been identified, including promoters, enhancers, silencers, and insulators. These elements influence when and where genes are expressed and often function in a cell-type-specific manner (Gerstein et al., 2012; Kundaje et al., 2012).

The ENCODE Project also highlighted the importance of chromatin structure in gene regulation. Chromatin organization, which is shaped by histone modifications and DNA accessibility, determines whether regulatory elements and genes can interact. Changes in chromatin state can activate or repress gene expression, allowing cells to respond to developmental cues and environmental signals.

Another major discovery of the ENCODE Project concerns noncoding RNAs. Researchers identified thousands of long noncoding RNAs that appear to have regulatory functions. These RNAs are often expressed in specific tissues and developmental stages and can influence transcription, chromatin remodeling, and RNA processing (Djebali et al., 2012).

In addition, ENCODE research demonstrated that many genetic variants associated with human diseases are located in noncoding regulatory regions rather than within protein-coding genes. By integrating ENCODE data with genome-wide association studies (GWAS), scientists were able to functionally annotate many disease-associated variants. This finding has significantly improved understanding of how changes in gene regulation contribute to disease risk (Schaub et al., 2012; Boyle et al., 2012).

Relevance of the ENCODE Project

Understanding the functional elements of the genome is essential for explaining how genes are regulated, how cells differentiate, and how complex organisms develop. The ENCODE Project provides open access to high-quality genomic data, making it a valuable resource for both research and education (Rosenbloom et al., 2010).

By integrating ENCODE data with other large-scale genomic datasets, researchers gain new insights into the molecular basis of complex diseases such as cancer, diabetes, and neurological disorders. Identifying functional variants in regulatory regions allows scientists to develop more accurate models of gene regulation and to identify potential targets for medical intervention.

Key Vocabulary

Term	Definition
Genome	The complete set of genetic material (DNA) in an organism.
Gene	A DNA sequence that contains instructions to produce a functional product, usually a protein.
Noncoding RNA	RNA molecules that do not code for proteins but have regulatory functions.
Promoter	A DNA sequence where transcription of a gene is initiated.
Enhancer	A DNA region that increases the transcription of specific genes.
Chromatin	The complex of DNA and proteins that forms chromosomes in the nucleus.
Transcription	The process of copying a segment of DNA into RNA.
Epigenetics	The study of changes in gene expression that do not involve changes in the DNA sequence.
GWAS	Genome-Wide Association Study; identifies genetic variants linked to traits or diseases.

Interactive Vocabulary Practice (Gimkit Activity)

Now that you have learned the key concepts related to the human genome and the ENCODE Project, it is time to **check your understanding in an interactive way**.

Student Instructions

1. Take your phone, tablet, or computer.
2. Open the link provided by your teacher to the Gimkit game.
3. Enter the game code and your first name.
4. Read each question carefully and choose the correct answer.
5. Use the feedback after each question to learn from your mistakes.
6. Try to improve your score by applying the vocabulary and concepts from this section.

2. Execute: AR Application Features and Educational Value (Assemblr EDU)

In this learning unit, Assemblr EDU is used as the primary augmented reality platform to support the exploration of functional elements of the human genome. Assemblr EDU allows students to interact with layered three-dimensional content in a structured and intuitive way, making it particularly suitable for visualizing complex biological systems such as DNA organization, gene regulation, and chromatin structure.

Layered visualization of genomic elements
Assemblr EDU enables the creation of AR scenes composed of multiple content layers. In this unit, students explore layered genomic maps that include DNA strands, protein-coding genes, regulatory elements, noncoding RNA regions, and chromatin modifications. This layered approach helps learners understand how different functional elements coexist and interact within the genome, rather than viewing them as isolated components.

Interactive exploration and user control
Students actively engage with AR objects by tapping, rotating, and selecting elements within the Assemblr environment. Interactive hotspots provide concise explanations, guiding questions, and vocabulary support. This hands-on interaction encourages active learning and allows students to explore genomic structures at their own pace, reinforcing understanding through direct manipulation.

Visualization of gene regulation mechanisms
Assemblr EDU supports the visualization of regulatory processes by allowing learners to activate or deactivate specific genomic elements within the AR scene. For example, students can observe how the presence or absence of enhancers or regulatory regions influences gene expression. This interactive representation supports conceptual understanding of regulatory logic and cause-and-effect relationships in genomics.

Contextual and comparative learning
Through carefully designed AR scenes, students can compare gene regulation in different cellular contexts. By switching between visual layers or scenarios, learners observe how chromatin structure and regulatory element activity differ across cell types. This feature supports understanding of cell-type-specific gene expression and the dynamic nature of genome regulation.

Embedded scaffolding and formative assessment
Assemblr EDU allows the integration of instructional text, guiding prompts, and short reflection questions directly within the AR experience. These embedded scaffolds support comprehension and help students monitor their own learning. The AR tasks function as formative assessments, enabling teachers to observe student interaction and identify areas that require further clarification.

Alignment with learning objectives
By using a single AR platform throughout the unit, students develop familiarity with the interface and focus on scientific content rather than technical complexity. Assemblr EDU supports the unit’s learning objectives by combining scientific accuracy, digital competence, and inquiry-based learning, ensuring that AR serves as a meaningful educational tool rather than a visual add-on.

3. Enhance:

Augmented Reality (AR) serves as a powerful enhancement tool for learning about complex biological systems such as the human genome. Genomics involves processes that are abstract, microscopic, and dynamic, making them difficult to understand through traditional text-based or two-dimensional visual resources. By integrating AR into this learning unit, these invisible mechanisms become visible, interactive, and conceptually accessible for students.

Within the context of this unit on the ENCODE Project, Assemblr EDU plays a central role in enhancing conceptual understanding and learner engagement. Assemblr enables students to explore three-dimensional representations of genomic structures, regulatory elements, and chromatin organization in a spatial and interactive environment. This approach supports deeper comprehension by allowing learners to observe how different functional elements of the genome interact within a shared biological context.

Through direct interaction with AR content, students strengthen their spatial reasoning and systems thinking skills. By manipulating virtual genomic elements, learners gain insight into how DNA is organized within the nucleus, how chromatin structure influences accessibility, and how regulatory regions control gene expression. These interactions encourage students to move beyond memorization and toward meaningful conceptual understanding.

How Assemblr EDU Enhances Learning

1. Enhanced Visualization of Abstract Genomic Concepts

Assemblr EDU allows students to rotate, zoom, and explore three-dimensional models of DNA, chromatin fibers, regulatory regions, and transcription-related structures. This visual access is particularly valuable for understanding spatial mechanisms such as enhancer–promoter interactions and chromatin looping, which are often difficult to grasp through static diagrams alone.

Color-coded layers and structured layouts help students distinguish between different genomic elements and recognize functional relationships. For example, regulatory regions can be visually separated from coding sequences, reinforcing the concept that noncoding DNA plays an active and essential role in gene regulation.

2. Active and Experiential Learning

Rather than passively receiving information, students actively engage with AR-based scenarios. In Assemblr EDU, learners interact with hotspots, layered models, and guided prompts that require decision-making and interpretation. For example, students may explore how activating or deactivating a regulatory element affects gene expression within the AR environment.

This experiential approach promotes inquiry-based learning and encourages students to test hypotheses, observe outcomes, and reflect on cause-and-effect relationships. Real-time interaction helps reinforce learning and supports long-term retention of complex concepts.

3. Differentiated Instruction and Accessibility

Assemblr EDU supports diverse learning needs by offering multiple entry points to content. Visual learners benefit from three-dimensional models, while learners who need additional support can rely on embedded explanations and structured prompts. The platform allows teachers to adjust the level of guidance provided, making it suitable for mixed-ability classrooms.

Students can progress through AR activities at their own pace, revisiting elements as needed. This flexibility supports inclusive, student-centered learning and helps reduce cognitive overload when working with challenging scientific material.

4. Increased Engagement and Knowledge Retention

The interactive and immersive nature of AR increases student motivation and attention. By engaging multiple cognitive channels—visual, spatial, and kinesthetic—Assemblr EDU supports deeper learning and improved retention of genomic concepts.

When students actively manipulate genomic elements rather than simply reading about them, they are more likely to remember key ideas such as gene regulation, chromatin accessibility, and the functional importance of noncoding DNA. This engagement is especially valuable for students who may find molecular biology abstract or intimidating.

5. Real-World Context and Scientific Relevance

Assemblr EDU enables students to connect genomic concepts to real-world applications. AR scenes can be designed to include disease-associated variants, regulatory mutations, or cell-type-specific gene expression patterns inspired by ENCODE data. This helps learners understand how genomic research contributes to personalized medicine, disease prevention, and biotechnological innovation.

By linking classroom learning to authentic scientific questions, AR supports scientific literacy and helps students recognize the relevance of genomics in modern society.

Practical Integration within the Learning Unit

In this unit, Assemblr EDU is used not as an isolated activity but as an integrated learning environment. Students first build foundational knowledge through reading and vocabulary work, then apply and deepen their understanding through AR exploration and interactive challenges. Reflection tasks embedded within or following AR activities encourage students to articulate their learning and connect visual experiences to scientific explanations.

Through this structured integration, AR becomes a tool for enhancing understanding, supporting inquiry, and fostering curiosity. Assemblr EDU transforms abstract genomic knowledge into an interactive learning experience, helping students develop both scientific competence and digital skills.

Conclusion:

The human genome is a highly complex and dynamic system that extends far beyond the protein-coding sequences traditionally associated with genes. Findings from the ENCODE Project have fundamentally transformed our understanding of genome function by demonstrating that noncoding DNA, regulatory elements, and chromatin organization play essential roles in controlling gene expression (ENCODE Project Consortium, 2007; Djebali et al., 2012). These discoveries have shown that biological complexity arises not only from genes themselves but from the sophisticated regulatory networks that govern when, where, and how genes are activated.

This learning unit has introduced students to the principles of functional genomics by combining scientific knowledge with innovative digital tools. Through the exploration of ENCODE research, learners have examined how regulatory regions, noncoding RNAs, and chromatin structure interact to fine-tune gene activity. By working with authentic scientific concepts and data, students gained insight into how modern genomics research is conducted and why large-scale collaborative projects are essential for advancing biological knowledge.

The integration of **Augmented Reality (AR)**, specifically through **Assemblr EDU**, has played a central role in enhancing student engagement and understanding. AR transformed abstract and microscopic genomic processes into interactive and visually accessible experiences. By manipulating three-dimensional models and exploring layered genomic structures, students were able to build deeper conceptual understanding and strengthen their spatial and systems thinking skills. This approach supports inquiry-based learning and encourages students to move beyond memorization toward meaningful interpretation of scientific phenomena.

In addition to developing subject-specific knowledge, this unit supported the acquisition of transversal skills. Students improved their digital competence through the use of AR technologies, strengthened collaboration and communication skills during group activities, and practiced critical thinking by analyzing regulatory mechanisms and disease-associated variants. Gamified elements and formative assessments further supported motivation and active participation throughout the learning process.

Overall, this learning unit demonstrates how the thoughtful integration of scientific research and digital innovation can create inclusive, engaging, and future-oriented educational experiences. By connecting genomic theory with interactive AR-based practice, students are better prepared to understand the relevance of genomics in medicine, biotechnology, and society. The unit encourages scientific curiosity, responsible use of digital technologies, and an appreciation for the complexity of the human genome—key competencies for learners navigating an increasingly science-driven world.

Phases of the Learning Unit and Description

Phase	Description
Explore	- Research and discovery: Students explore the Human Genome Project and the origins of the ENCODE Project using adapted scientific texts and guided reading activities.
	- Content development**:** Introduction to key genomic concepts, including DNA structure, genes, coding and noncoding regions, regulatory elements, chromatin, and functional genomics. Vocabulary tables and visual aids support conceptual understanding.
	- Conceptual scaffolding**:** Student difficulties in understanding complex gene regulation are identified through discussion and formative checks. Interactive tools, including a Gimkit vocabulary activity, are used to reinforce key terminology and prepare learners for AR-based exploration.
Execute	- Curriculum implementation: Core lessons are delivered through interactive presentations, scientific texts, and guided discussions. Assemblr EDU is introduced as the main AR platform to support understanding of chromatin organization, gene regulation, and regulatory interactions.
	- Interactive AR exercises: Students complete Assemblr-based AR activities, including exploration of layered genomic maps, identification of regulatory elements, and visualization of gene expression mechanisms inspired by ENCODE data.
	- Feedback and reflection: Teachers collect student reflections, peer feedback, and results from formative assessments (including AR tasks and digital quizzes) to monitor learning progress and adjust instruction.
Enhance	- AR integration: Assemblr EDU is used to model complex genomic structures and regulatory systems. Students interact with visual overlays showing gene expression patterns, chromatin organization, and enhancer–promoter interactions.
	- Inquiry-driven learning: Learners engage in structured AR activities, including hotspot-based exploration, guided simulations, and step-by-step AR walkthroughs of genome regulation processes.
	- Gamification elements: Students earn points or digital badges for completing AR tasks, accurate use of scientific vocabulary, and successful identification of genomic elements. - Quests and progression: Optional "Genomic Quests" allow students to solve increasingly complex ENCODE-based challenges in Assemblr, unlocking new AR scenes or content layers as mastery is demonstrated. - Collaborative AR tasks: Students work in teams to construct functional gene networks, chromatin models, or simplified transcription simulations within the AR environment, strengthening peer learning and communication skills.
	- AR-based assessment: Students complete structured assessments directly connected to AR activities, including labeling tasks, regulatory region identification, and cause–effect predictions based on genomic changes. Assessment is guided by rubrics focused on conceptual understanding, accuracy, and correct use of biological vocabulary.

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