

Task 1: Depth Study

Due Date: Monday 9th December 2024 by 3:00pm

Task Distributed: 18/10/2024

Unit: Genetics/Disease

Task Type: Depth Study

Task Weighting: 30%

Outcomes: 11/12-1, 11/12-3, 11/12-5, 11/12-6, 11/12-7, BIO 12-12, BIO12-13, BIO12-15

Task Description

This assessment addresses the syllabus requirement for students to complete a Depth Study.

A depth study is any type of investigation that allows the student to gain knowledge in an area of their individual interest. It includes secondary-source research, data analysis and a written report. Students will be examined on their ability to gather relevant secondary-sourced data and how they communicate their findings to a particular audience.

This depth study uses medical genetics as a context for an investigation into the uses of genetic technologies to diagnose genetic conditions and treat disease. The investigation takes the form of an evidence-based evaluation of the application of the technology, looking at risks and benefits, including the limitations and social and ethical issues involved.

NESA Glossary of Key Words

Understand the verb associated with the task. The verb will provide an understanding of the detail needed to successfully answer the question.

- **DISCUSS:** Identify issues and provide points for and/or against.
- **EVALUATE:** Make a judgement based on criteria; determine the value of.

Check the NESA Glossary of Key Words for further guidance <https://educationstandards.nsw.edu.au/wps/portal/nesa/11-12/hsc/hsc-student-guide/glossary-keywords>

Details of Submission

Medical applications of genetic technology are important for the syllabus and your future lives as informed citizens and consumers. All medical technologies have limitations, benefits and risks, so they must be carefully evaluated before they are adopted.

During this depth study, you will become an expert in a disease you choose. You will also select a genetic technology that could be used to diagnose, prevent or treat that disease. You will develop an inquiry question that can be addressed with an evaluation of the benefits and risks of the genetic test or technology in the context of your chosen disease. This should include the potential benefits and limitations of the technology as well as associated social, ethical, legal or economic issues.

Examples of inquiry questions

- “Should gene editing be used to treat sickle cell anaemia?”
- “Should all newborns be DNA tested for inherited heart disease?”
- “Should the government pay for DNA sequencing when there is a family history of heritable cancer?”
- “Should carrier screening for spinal muscular atrophy be a requirement for all parents-to-be?”
- “Should consumers find out their risk for inherited high cholesterol using online genetic testing?”
- “Should genome editing be used to treat inherited hearing loss?”

Class time for this depth study will be set aside for independent research and report writing.

Nature of Assessment task

You will present your evaluation as a report with the following sections:

1. Introduction outlining inquiry question.
2. About the genetic disease
3. About the genetic test or technology
4. Conclusion
5. Appendix (evaluating reliable references used to gather data and information)
6. Reference List (Harvard Style)

Report format

1. Introduction

- Identifies the genetic disease and the genetic technology being investigated
- Outlines the inquiry question and justifies its importance

2. About the genetic disease

- Symptoms, age of onset, etc.
- Range of genes/mutations involved.
- What is known about the protein product of the gene(s) involved?
- How genotype contributes to disease phenotype (i.e., how do changes to the protein result in the symptoms).
- Inheritance pattern.
- Current methods of diagnosis and treatment.
- Prevalence (i.e., how commonly it occurs). This could include data from a population genetics study.

3. About the genetic test or technology

- Scientific methods used.
- How it could be (or currently is) applied in the context of the chosen disease.

Risk/benefit analysis of inquiry question (should form main section of report)

- Risks and benefits of chosen application of DNA testing or genetic technology in the context of chosen disease
This could include:
 - Uses, advantages and limitations
 - Comparison with current or prior methods of diagnosis/treatment

- Social, economic and/or and ethical implications (including a definition of what these terms mean).
- Data on clinical validity

Benefits

How effective is the test or technology? How useful? How is it better than current approaches to diagnosis/prevention of the disease?

What are the potential positive outcomes: For the individual? For society? What are the cost savings? How is the test or technology ethical?

Risks

What are the limitations of the test or technology? What are the potential harms: For the individual? For society?

What are the costs? Could its application be considered unethical? Can these risks be reduced?

4. Conclusion

- Meaningful answer to the inquiry question based on evaluation.
- What might the future hold for the prevention or treatment of this disease?
- What other genetic technologies might be used (either now or in the future).

5. Appendix (evaluating reliable references used)

Select three references that you have used within your depth study and for each analyse and evaluate the data and information found within these references. Consider the author, publication, funding sources and audience. How reliable/valid/appropriate is the reference for your report? Is it peer reviewed? Provide evidence to explain the reliability, validity, and accuracy of each of these three sources.

6. Reference List

A reference list, citing all the resources you have used throughout your entire depth study should be written in Harvard Style.

Submission Information

As per NESAs requirements, the depth study will be conducted during class time in your Biology lessons (15 hours) and at home. Once you start your assessment you are to share your GOOGLE DOC with your teacher who will track your progress and provide feedback where necessary.

The report must be submitted on or before the due date as a **GOOGLE DOC** submitted on google classroom.

Teacher Feedback and Student Self-Reflection

- The task will typically be returned to students within 14 days of the due date.
- At this time feedback including information on how to improve will be provided through marking criteria and written feedback.
- Students can clarify or seek further feedback by speaking with their teacher or the assessment marker.
- You will also receive feedback on your literacy performance based on the criteria in the school's literacy marking rubric. The marks achieved for literacy will account for between 10% – 20% of the maximum task value.

Upon return of the task, students will also be expected to complete a self-reflection.

How does this link to my learning?

- This task will enable students to develop their research and presentation skills.
- They will also further their understanding in an area of genetics and technology of their choice.
- This information will be relevant when studying the Genetics and Disease topics covered in class.

Assessment Procedures

All students should be fully aware of the School Assessment Procedures for their year group. These were provided at the beginning of the school year and are available off the school website under the Learning Tab for each year group.

4. HOW WILL MARKS BE AWARDED TO MEASURE MY LEARNING?

Section <i>Outcomes</i>	0 marks	1 mark	2 marks	3 marks	Marks
Introduction Questioning and predicting <i>A student: develops and evaluates questions and hypotheses for scientific investigation BIO11/12-1</i>	No inquiry question present.	Inquiry question is present but may be weak or not relevant to the background information.	Uses the background information to develop a relevant inquiry question with some guidance.	Effectively uses the background information to develop a highly relevant inquiry question with limited guidance.	
	Genetic disease named but no genetic component.	The report names a genetic disease, or a non-infectious disease with some genetic component, though this is not well explained.	The report identifies a genetic disease or identifies a non-infectious disease with a clear genetic component.	The report clearly identifies a genetic disease or identifies a non-infectious disease with a strong explanation of its genetic component.	
	Inquiry question does not include a medical application of genetic technology.	Inquiry question includes a medical application of genetic technology, though appropriateness for the disease is not clear.	Inquiry question identifies a medical application of genetic technology that is appropriate and relevant for the chosen disease (an approach to genetic testing or a genetic therapy).	Inquiry question identifies a medical application of genetic technology that is highly appropriate and relevant for the chosen disease (an approach to genetic testing or a genetic therapy).	
	Inquiry question does not identify a concept.	Inquiry question identifies a concept that can be investigated, but it may not suit the scope of the report or an evaluation or risks and benefits.	Inquiry question identifies a concept that can be investigated within the scope of the report and allows for some evaluation of risks and benefits.	Inquiry question identifies a concept that can be investigated within the scope of the report and allows for in-depth evaluation of risks and benefits.	
	No discussion.	There is some discussion of how the inquiry question has been evaluated, revised or modified after initial research and	There is a discussion of how the inquiry question has been evaluated, revised or modified	There is strong evidence of how the inquiry question has been thoughtfully evaluated, revised or modified after initial research and evaluation.	

		evaluation. But it may be brief or flawed.	after initial research and evaluation.		
About the disease <i>Explains non-infectious disease and disorders and a range of technologies and methods used to assist, control, prevent and treat non-infectious disease BIO12-15</i> <i>explains the structures of DNA and analyses the mechanisms of inheritance and how processes of reproduction ensure continuity of species BIO12-12</i>	No description of the disease.	Describes the disease and its symptoms, as well as current approaches to diagnosis, treatment/management, and prevention. Some sections may have been missed or poorly covered. Technical terms may have not been used or used incorrectly.	Describes the disease and its symptoms, as well as current approaches to diagnosis, treatment/management, and prevention. This includes good use of technical terms/scientific language.	Accurately and concisely describes the disease and its symptoms, as well as current approaches to diagnosis, treatment/management, and prevention. This includes high-level use of technical terms/scientific language.	
	No attempt is made to explain the link.	An attempt is made to explain the link between a genetic mutation, changes to a protein, and how these cause the disease phenotype. This may have missed or incorrect aspects.	Explains the link between a genetic mutation, changes to a protein, and how these cause the disease phenotype.	Clearly explains the link between a genetic mutation, changes to a protein, and how these cause the disease phenotype, using well synthesised concepts and evidence of systems thinking.	
	No data included.	Includes incomplete data.	Includes secondary data to describe the prevalence and/or inheritance pattern of the disease.	Includes data from a population genetics study to describe the prevalence and/or inheritance pattern of the disease.	
About the genetic technology <i>explains the structures of DNA and analyses the mechanisms of inheritance and how processes of reproduction ensure continuity of species BIO12-12</i>	No description of the way in which the chosen medical application of genetic technology could be employed to detect, treat, or prevent the chosen disease.	There is some description of the way in which the chosen medical application of genetic technology could be employed to detect, treat, or prevent the chosen disease (DNA sequencing/profiling or recombinant DNA technology).	There is a well-developed description of the way in which the chosen medical application of genetic technology could be employed to detect, treat, or prevent the chosen disease (DNA sequencing/profiling or recombinant DNA technology).	N/A	
	No description of the technique and application of the chosen disease is attempted.	Description of the technique and application in the context of the chosen disease is attempted but may be weak.	There is a concise and clear explanation of how the technique works, covering aspects that are relevant for the analysis to follow.	N/A	

<i>explains natural genetic change and the use of genetic technologies to induce genetic change BIO12-13</i>					
Risk and benefit analysis <i>explains natural genetic change and the use of genetic technologies to induce genetic change BIO-12-13</i> <i>conducts investigations to collect valid and reliable primary and secondary data and information BIO11/12-3</i>	There is no description of benefits or risks of the genetic technology.	There is a description of some benefits OR limitations of the genetic technology. Evaluation includes some limited considerations of the clinical benefits/limitations of the approach OR socio-scientific issues relevant to the approach (legal/ethical/economic etc).	There is an evaluation of the benefits and limitations of the genetic technology, but some may be covered insufficiently. Evaluation includes thoughtful considerations of the clinical benefits/limitations of the approach OR socio-scientific issues relevant to the approach (legal/ethical/economic etc).	There is a very strong evaluation with well-chosen benefits and limitations of the genetic technology. Evaluation includes thoughtful considerations of the clinical benefits/limitations of the approach AND socio-scientific issues relevant to the approach (legal/ethical/economic etc).	
	No evidence of wider reading in conducting the investigation.	Evidence of limited wider reading in conducting the investigation.	Evidence of some wide reading in conducting the investigation. Selection of a range of sources and extraction of information and/or data. Information/data is used in some aspects to support arguments.	Evidence of much wider reading in conducting the investigation. Selection of a wide range of reliable sources and extraction of valid and reliable information and/or data. Information/data is well integrated to support arguments.	
Conclusion <i>solves scientific problems using primary and secondary data, critical thinking skills and scientific processes BIO11/12-6</i> <i>explains non-infectious disease</i>	There are no statements to link back to the answer to the inquiry question.	There are some statements that link back to the answer to the inquiry question.	There is a conclusion that provides some answer to the inquiry question, making some reference to information from prior sections.	There is an insightful conclusion that provides a meaningful answer to the inquiry question, distilling key information from prior sections.	
	No suggestion of the implications of an aspect of the approach for the prevention or treatment of	Provides a limited suggestion of the implications of an aspect of the approach for the prevention	Provides a good suggestion of the implications of the approach for the prevention or treatment of the	Provides a very strong suggestion of the implications of the approach for the prevention or treatment of the	

<i>and disorders and a range of technologies and methods used to assist, control, prevent and treat non-infectious disease BIO12-15</i>	the disease or future directions for research.	OR treatment of the disease OR future directions for research.	disease, OR future directions for research.	disease, AND future directions for research.	
Communication and referencing (across whole report) <i>communicates scientific understanding using suitable language and terminology for a specific audience or purpose BIO11/12-7</i> <i>conducts investigations to collect valid and reliable primary and secondary data and information BIO11/12-3</i>	Report is very brief with sections missing and with incorrect information.	Report is brief AND/OR difficult to read. Sections miss key information or contain incorrect information.	Uses appropriate language overall for a specific audience and displays good written communication but it does contain areas that are unclear or hard to understand.	Uses appropriate language proficiently for a specific audience and displays very high-quality written communication which is clear and concise.	
	Most of the report is grammatically incorrect.	Spelling, grammar, and formatting have many errors.	Spelling, grammar, and formatting of report is to good standard but contains some errors.	Spelling, grammar, and formatting of report is to a very high standard and adds to clarity of writing. There are very minor or no errors.	
	No reference list	The reference list is missing or contains many mistakes and missing.	There is a reference list that contains all sources used in text. The sources are referenced with a few mistakes and inconsistencies.	There is a reference list that contains all sources used in text. The sources are consistently and correctly referenced with minor or no mistakes.	
Appendix (evaluating reliable resources used)	No evaluation of secondary sources	There are some comments as to the relevance OR accuracy OR validity OR reliability of secondary sources. This may be weak.	There is an evaluation of secondary sources including relevance, accuracy, validity and reliability. Some aspects may be missing.	There is a very well-developed evaluation of secondary sources for relevance, accuracy, validity and reliability.	

<p><i>develops and evaluates questions and hypotheses for scientific investigation BIO11/12-1</i></p> <p><i>analyses and evaluates primary and secondary data and information BIO11/12-5</i></p> <p><i>communicates scientific understanding using suitable language and terminology for a specific audience or purpose BIO11/12-7</i></p>					
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TOTAL

/52

