Summary
Medical applications of genetic technology are increasingly important in twenty-first century healthcare. DNA sequencing is becoming an affordable tool for doctors, and gene-based therapies continue to emerge.

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. Students choose a genetic disease and research the use of genetic testing or another genetic technology to diagnose, treat or prevent the 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. Findings could be presented as a structured report.

Stimulus materials, resources and activities are also provided that cover the background and develop the necessary skills. Activities align to a wide range of syllabus outcomes from modules 5, 6 and 8 as well as Working Scientifically skills.

In this teacher guide...
Resources and Activities ................................................................. 2
About the Investigation .................................................................. 3
Syllabus links .................................................................................. 3
Scaffolding the Depth Study ............................................................. 4
Evidence Plan .................................................................................. 5
Student Logbook ............................................................................ 5
Report format .................................................................................. 6
Teacher background notes ............................................................... 7
Avoiding plagiarism ......................................................................... 8
Notes on disease selection and inquiry questions ......................... 8
Acknowledgements ......................................................................... 9
Other teaching resources ................................................................ 10
Appendix 1: Evidence Plan Template .............................................. 11
Appendix 2: Sample Student Outline .............................................. 14
Appendix 3- Sample Marking Rubric .............................................. 16
Resources and Activities

We have developed resources and activities that could be used to support this depth study or incorporated into teaching programs or other depth studies. The activities are designed to cover the context and background knowledge needed to undertake an investigation about medical applications of genetic technology and align with the syllabus (modules 5, 6 and 8). Resource guides are also provided for students as starting points for their research.

The table below outlines the activities. More information, including a full syllabus map, can be found on our website [www.garvan.org.au/kccg-teachers](http://www.garvan.org.au/kccg-teachers). Full teacher guides are available by emailing [kccgeducation@garvan.org.au](mailto:kccgeducation@garvan.org.au).

<table>
<thead>
<tr>
<th>Stimulus materials</th>
<th>Videos and articles that can be used to engage students with the context of medical genetics.</th>
</tr>
</thead>
</table>
| Activity: Research Matrix for Genetic Diseases | -"Systems thinking" matrix for concept-mapping inherited disease  
-Case studies provide scaffolded practice  
-Students use matrix to research a chosen genetic disease  
-Skills in synthesis and referencing of multiple sources  
-Includes resource guide on a selection of genetic diseases  
-Students produce summary of disease for formative feedback |
| Activity: Tiny Genome | -Paper based practical  
-Whole genome sequencing of a hypothetical creature  
-Types and effects of mutations  
-Extension activities introduce population genetics databases and problem solving. |
| Activity: Six Ws of Genetic Testing | -Covers the “what/who/how/when/why/whether” of genetic testing.  
-Problem solving and critical thinking based on case studies of real life applications  
-Role play activity for considering implications of genetic tests |
| Activity: Technology explainer <coming soon> | -Literacy activity  
-Article about medical applications of genetic technologies  
-Students extract information to answer inquiry question: “Could DNA manipulation for medicine change the human population forever?” |
| Activity: Genetic Testing - Issue Scan | -Ethics activity (introduces bioethical principles)  
-Students consider a range of perspectives regarding possible ethical, legal and social implications of genetic and genomic testing  
-Develop skills in evidence-based argument. |
| Student resource guides | A list of references relating to the benefits and limitations of genetic technology and the associated social and ethical considerations to give students a starting point in their research |
About the Investigation

Before a new genetic technology is implemented within a healthcare system, it must first be evaluated. For example, before a new genetic test is approved in Australia, the Medical Services Advisory Committee creates a “Clinical Utility Card”, which reports on the benefits and limitations of the test as well as social, ethical and economic considerations surrounding its use.

For this depth study, students develop an inquiry question to evaluate the use of a genetic technology in the context of a chosen genetic disease. They then examine the risks and benefits of the technology, using evidence to support their claims.

Examples of inquiry questions

- 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?

(see “Notes on disease selection and inquiry questions” below for more information)

Syllabus links

This unit of work has links to multiple content and skills outcomes. A full syllabus map for the depth study and supporting activities can be found at www.garvan.org.au/kccg-teachers. The following outcomes are assessed in the suggested marking rubric for the report (appendix 3):

Working Scientifically outcomes:

Questioning and Predicting

- develop and evaluate inquiry questions and hypotheses to identify a concept that can be investigated scientifically, involving primary and secondary data

Conducting Investigations

- select and extract information from a wide range of reliable secondary sources and acknowledge them using an accepted referencing style

Problem Solving

- use scientific evidence and critical thinking skills to solve problems

Communicating

- communicates scientific understanding using suitable language and terminology for a specific audience or purpose

Knowledge and Understanding outcomes:

Module 5: Heredity

- model the process of polypeptide synthesis, including:
  - assessing how genes and environment affect phenotypic expression
- investigate the use of technologies to determine inheritance patterns in a population using, for example:
  - DNA sequencing and profiling

Module 6: Genetic Change

- evaluate the benefits of using genetic technologies in agricultural, medical and industrial applications
- interpret a range of secondary sources to assess the influence of social, economic and cultural contexts on a range of biotechnologies

Module 8: Non-infectious Disease and Disorders

- investigate the causes and effects of non-infectious diseases in humans, including but not limited to:
  - genetic diseases
Scaffolding the Depth Study

The requirement for 15 hours of class time for the depth study means that students can be guided through the investigation one step at a time. A progression such as the following is suggested. This suggested program is based on twenty 45min lessons.

- **Student and teacher reflection (1 lesson)**
- **Writing and summative assessment of a structured report (3 lessons)**
- **Investigation of risks and benefits of genetic test/technology (4 lessons)**
  - Student Resource Guide
  - Evidence plan template
  - Peer Review
- **Investigation of a chosen genetic disease (3 lessons)**
  - Research matrix
  - Genetic disease resource guide
- **Class activities - genetic technologies and social and ethical issues (3 lessons)**
  - Genetic technologies explainer
  - Issues scan
- **Students propose and evaluate inquiry question (1 lesson)**
- **Class activities - engage and explore context of genetic medicine (4 lessons)**
  - Stimulus materials
  - Tiny genome
  - Six Ws
Evidence Plan

Students can use the template in appendix 1 to plan their report, listing the benefits and risks of their chosen approach to genetic testing or technology. Students summarise their evidence and evaluate their sources, similar to an annotated bibliography. The plan can also be used to practice formatting of references.

This template should be peer reviewed, focusing on the claims, evidence and quality of sources. A suggested rubric is included that uses the “what worked well/event better if” (WWW/EBI) framework for peer assessment.

This activity addresses the following Working Scientifically skills outcomes, which can be assessed by requiring the template and peer review to be included with the final report:

Conducting Investigations
- select and extract information from a wide range of reliable secondary sources and acknowledge them using an accepted referencing style

Analysing Data and Information
- assess the relevance, accuracy, validity and reliability of primary and secondary data and suggest improvements to investigations

Communicating
- construct evidence-based arguments and engage in peer feedback to evaluate an argument or conclusion

Student Logbook

Students could keep a logbook (either paper or electronic) to record progress throughout the depth study. This can be attached as an appendix to the student’s reports. For assessment, this should include:
- Selection and evaluation of the inquiry question
- Justification for any changes to the inquiry question
- Feedback on disease summary produced during the “Genetic disease research matrix” activity
- Evidence plan for the risk benefit evaluation, which includes evaluation of resources and peer feedback on evidence-based argument.

It could also be used for other activities completed in class and/or notes on independent research.
Report format
The exact format for the report should be modified from school-to-school and year-to-year. The requirements will depend on the needs of the student group. The marking rubric should be adapted to match the specifications given to students. We have based our suggestions around using a written report, however this could be adapted for other formats.

Basic report format
Introduction with inquiry question
About the genetic disease
About the genetic test or technology
Risks and benefits (evaluation of the inquiry question)
Conclusion

Options for expanding on simple format
(NB: not all of these aspects could be covered in a single report):

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

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 (could include data from a population genetics study)

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 methods of diagnosis/treatment
  - Social, economic and/or and ethical implications
  - Data on clinical validity and utility (advanced students, refer to NWABR Genetic Testing lesson plan in other teaching resources section)

Conclusions and future implications
- 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) based on this information.

Appendix: Student logbook
Teacher background notes

In the 15 years since the human genome project, the cost of DNA sequencing has reduced to the point where the genome of an individual – their entire set of genetic information – can be made available for use in healthcare. This has immediate impact on the way that rare genetic diseases and cancer are diagnosed and treated. It also suggests a future where genetic information can be used to make everyday health decisions. Advances in genetic engineering mean that genetic therapies will also be increasingly common.

Tomorrow’s citizens will not only need to understand DNA and the uses of genomic data, but they also need to have the skills to find and critically analyse information, and make well-informed and reasonable decisions about their own health and the implications for society.

The new HSC syllabus contains a lot of genetics-related content. This depth study addresses multiple content topics and working scientifically skills. Students will gain a systematic understanding of a genetic disease on multiple levels: from the way the change in the DNA effects phenotype, its inheritance and population genetics, its diagnosis, treatment and management, through to the ethical and social implications of using genetic information or technology to prevent or manage the disease. This deep understanding may then act as a scaffold for understanding other diseases and biological processes throughout the HSC course. The depth study also develops numerous “learning across the curriculum” capabilities including critical thinking, ethical understanding, ICT capability, literacy, personal and social capability, and an appreciation of difference and diversity.

The investigation undertaken in this depth study is secondary-sourced, however it is not simply an informational report. Students should be encouraged to think scientifically about gathering evidence of risks and benefits to solve a real-world problem in medical genetics. The evaluation framework used in this depth study is based on real-world decision-making strategies in science and medicine. You can read more about such approaches in:

How is genetic testing evaluated? A systematic review of the literature
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5945588/

About Us
The Kinghorn Centre for Clinical Genomics (KCCG) is a research centre within the Garvan Institute of Medical Research, located in Darlinghurst, NSW. The focus of KCCG is realising the clinical value of the genome for precision healthcare. We have world-class researchers, clinicians and bioinformaticians studying a wide range of diseases and applications of genomic information. The Education and Communication team delivers targeted education and community engagement about genomics for healthcare professionals, teachers and communities. We’re committed to supporting teachers with resources and strategies for teaching genetics. If you have not already joined our mailing list, visit our website and subscribe for updates on resources and professional development opportunities
Avoiding plagiarism
Unfortunately, the availability of completed research reports on the internet is a risk that needs addressing even at the secondary level. We suggest taking the following measures to reduce the risk of your students accessing completed reports that fulfil the marking criteria for your depth study:

Timing
Requiring students to choose their genetic disease and their inquiry questions during lesson times rather than at home prevents them from seeking completed assignments. Inquiry questions can be modified throughout with justification in student logbooks.

Adapt and modify
The guidance given about the report requirements in this teacher guide is intentionally broad. You may choose to narrow the scope of the task, assign diseases or technologies, or include specific additional requirements to create a unique assessment task.

Supervise, scaffold, and support
We suggest providing class time for independent work during which student research can be supervised. Ensuring that students are well supported and have the required skills to complete the assessment task has also been shown to reduce the risk of plagiarism. This is the goal of the activities suggested in this depth study.

Give explicit instructions
Giving students clear instruction on academic integrity can reduce accidental plagiarism. School-wide policies, NESA’s “All my own work” modules, and the input of school librarians can provide valuable guidance for students.

Feed-forward and student logbooks.
This depth study provides numerous points where formative assessment can be provided to students. Student logbooks could also be used to record teacher advice and show evidence of edits and reflection. This “feed-forward” for the final report increases the quality of the work produced and is also evidence of academic integrity.

Further reading on avoiding student plagiarism can be found here

Notes on disease selection and inquiry questions
The research matrix activity should be used to introduce the exploration of a genetic disease. It includes a guide for students with a selection of genetic diseases and some links to websites, articles and papers to begin their research. These diseases were chosen because of links with other parts of the syllabus or connections with Garvan research. However, almost any genetic disease could be used if teachers would prefer to give their students a wider selection of choices.

If students are choosing their own diseases, the following guidelines should be considered
• The disease has a well-established genetic component (ie at least one causative gene has been found). Multifactorial diseases could be studied if there is a risk variant with a well-defined mechanism.
• The student has capacity to find resources and possibly adapt assignment guidelines as appropriate
• The disease is not covered extensively elsewhere in the HSC course. Well known genetic diseases such as Huntington’s disease, PKU, cystic fibrosis and BRCA1/2 mutation have numerous resources available, which may prove either overwhelming or unfairly favourable for students.

To prompt students to choose an inquiry question, the following questions might be used (or adapted according to individual requirements).
• When researching your chosen disease, what challenges or controversies did you come across when it comes to diagnosis, prevention or treatment?
• How might DNA sequencing, genetic testing or other genetic technologies be applied to the prevention, diagnosis or treatment of this disease?
• Can you use the Six Ws (from the activity in section 1) to propose an approach to genetic testing?
• What ethical or social issues are important for these applications?
• What questions need to be asked?

We recommend advising students to consider current or ‘near-future’ applications so they can evaluate in the context of contemporary society. For example, rather than “should embryo editing be allowed for <genetic disease>?” students may evaluate the question “should parents undertake preimplantation genetic diagnosis…”, with the future possibility of embryo editing being included as a risk or a benefit.

Allowing students to develop and evaluate their own inquiry question fulfils the requirement of the depth study to assess the working scientifically skill “Questioning and Predicting”. A student’s choice of question can be assessed as part of the final report (See Appendix 3). One of the outcomes involves developing a question that is suited to the investigation that is being performed. Asking students to being their question with the word “should” will help them to phrase an inquiry suitable for a risk benefit evaluation.

Evaluation of enquiry question
Students could be asked to evaluate their inquiry question in their logbooks using prompts such as:
• Why have you chosen this application of genetic technology?
• Do you know whether this approach is currently used?
• What initial ideas do you have regarding the risks and benefits?
• What are two important questions you will need to research?

Acknowledgements

We thank the genomics researchers and education experts at the Kinghorn Centre for Clinical Genomics, as well as the gifted and enthusiastic biology teachers that have collaborated with us on these resources. Your time and effort is greatly appreciated.
Other teaching resources

The following freely available resources may also be useful to support the depth study:

**Genetics Home Reference** - from National Library of Medicine
This is a database from the National Library of Medicine that contains easy-to-read descriptions of genetic diseases and genes. The “help me understand genetics” section contains a wealth of information on genetic disease and technologies.

**Genetic and genomic testing** – Fact sheet from Centre for Genetics Education
This fact sheet on genetic and genomic testing contains a lot of the background information needed for the depth study. Authentic resource used for patients.

**Personal Genetics Education Project (pgEd)** from Harvard University
http://pged.org/lesson-plans/
Lesson plans including: genetics and reproduction, consumer genetics, genome editing and CRISPR, personalised medicine, and scientific themes in personal genetics. The “Introduction to personal genetics” activity includes a good overview PowerPoint presentation. Other lessons could be used for greater depth on a particular topic and to introduce some of the ethical considerations.

**Introducing genomics** - from Nowgen Schools Genomic Programme
http://www.genomicsforschools.org/resource/pos/
This advanced resource provides additional ideas for extending students.

‘What do you think’ from Unlocking Life’s Code (Smithsonian and NHGRI)
https://unlockinglifescode.org/wdyt/
Recommended introduction- a brief, interactive website where students answer questions about genomic information and compare their answers to others.

‘Bioethics 101’ from Northwest Association for Biomedical Research
https://www.nwabr.org/teacher-center/bioethics-101
The Issue Scan activity on our website includes material adapted from NWABR. The full series of six lesson plans includes many other activities that could be useful.

**Genetic Testing** – Lesson Plan from Northwest Association for Biomedical Research
https://www.nwabr.org/sites/default/files/Genetic_Testing_Lesson6_NWABR.pdf
This advanced lesson plan can be used to expand on evaluating genetic tests. It introduces the concept of clinical validity and treatment availability. It includes prompts for critical evaluation of texts, which may be useful to extend students.

‘Highschool Bioethics Project: Genetic Testing’ from NYU School of Medicine
https://med.nyu.edu/highschoolbioethics/modules/genetic-testing
This advanced lesson plan includes a number of relevant ethical considerations as well as some research paper extracts to help students consider the impact of genetic testing.

**Improving your scientific writing style** - from Monash University
A useful set of tips for students on writing in the field of science.
Appendix 1: Evidence Plan Template

This analysis framework doubles as a plan for your report.

Begin by brainstorming the risks and benefits associated with your inquiry question. Consider the potential outcomes for the patient, their families and the wider society. Include ethical, legal, social and economic considerations.

You cannot cover all of the benefits and risks in your report so choose two or three that are most important or interesting to you. You can copy and paste the table to expand the template to include more claims. Find at least one reliable reference to support each point. This plan will be peer assessed and then go on to form the basis of your report.

For each main point you will include in your report, you should complete the following:

<table>
<thead>
<tr>
<th>Benefit/risk: Briefly describe the claim you are making. What is the benefit or risk?</th>
</tr>
</thead>
<tbody>
<tr>
<td>Evidence: Why is this a benefit/risk? How significant or likely is it? What evidence from your reference supports your claim? How would you convince someone who disagrees with you?</td>
</tr>
<tr>
<td>Reference: Author, year of publication, title of article or chapter, title of publication (eg website, newspaper, journal), url</td>
</tr>
<tr>
<td>Evaluation of reference: Consider the author, publication, funding sources and audience. How reliable/valid/appropriate is the reference for your report? Is it peer-reviewed? What biases might be involved?</td>
</tr>
</tbody>
</table>

**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?

<table>
<thead>
<tr>
<th>Benefit 1:</th>
</tr>
</thead>
<tbody>
<tr>
<td>Evidence</td>
</tr>
<tr>
<td>Reference</td>
</tr>
<tr>
<td>Evaluation</td>
</tr>
</tbody>
</table>
### Benefit 2:

**Evidence**

**Reference**

**Evaluation**

### 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?

#### Risk 1:

**Evidence**

**Reference**

**Evaluation**

#### Risk 2:

**Evidence**

**Reference**

**Evaluation**
## Peer evaluation of evidence plan

<table>
<thead>
<tr>
<th>Outcome</th>
<th>What worked well?</th>
<th>Even better if…</th>
</tr>
</thead>
<tbody>
<tr>
<td>The risks, benefits and implications chosen are clear and make sense for the inquiry question</td>
<td></td>
<td></td>
</tr>
<tr>
<td>The evidence logically supports the claims</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Counterpoints have been considered</td>
<td></td>
<td></td>
</tr>
<tr>
<td>The references are valid and reliable</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Limitations to the references have been identified in the evaluation</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
Appendix 2: Sample Student Outline

Year 12 Biology Depth Study
GENETIC TECHNOLOGIES, DNA & DISEASE

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 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 we sequence the genomes of newborns to diagnose inherited heart diseases”
“Should gene editing be used to treat sickle cell anaemia”

Class time for this depth study will be divided between activities that develop your background understanding of medical genetics, and time for independent research and report writing. Resource guides will be provided to support your research.

Nature of Assessment task
You will present your evaluation as a report with the following sections:
- Introduction outlining inquiry question
- About the genetic disease
- About the genetic test or technology
- Risks and benefits (evaluation of the inquiry question)
- Conclusion

Your report should include in-text citations and reference list according to school guidelines.

You will also hand in a logbook that includes
- Evaluation of your inquiry question and justification of any changes you make
- Formative feedback on disease summary produced during week #
- Peer evaluation of an evidence plan for your report produced during week #
  - See attached template
Outcomes Assessed

Working Scientifically
A student:
BIO11/12-1: develops and evaluates questions and hypotheses for scientific investigation
BIO11/12-3: conducts investigations to collect valid and reliable primary and secondary data and information
BIO11/12-5: analyses and evaluates primary and secondary data and information
BIO11/12-6: solves scientific problems using primary and secondary data, critical thinking skills and scientific processes
BIO11/12-7: communicates scientific understanding using suitable language and terminology for a specific audience or purpose

Knowledge and Understanding
A student:
BIO12-12: explains the structures of DNA and analyses the mechanisms of inheritance and how processes of reproduction ensure continuity of species
BIO12-13: explains natural genetic change and the use of genetic technologies to induce genetic change
BIO12-15: explains non-infectious disease and disorders and a range of technologies and methods used to assist, control, prevent and treat non-infectious disease

General Capabilities
- Critical and creative thinking
- Ethical understanding
- Information and communication technology capability
- Literacy
- Personal and social capability
### Appendix 3 - Sample Marking Rubric

This table provides examples of outcomes and descriptors that could be adapted depending on the specific requirements of the report. Weightings and word limits should be chosen based on the focus of the depth study and the capacity of the students.

<table>
<thead>
<tr>
<th>Section</th>
<th>Outcome</th>
<th>Bands 5-6</th>
<th>Bands 3-4</th>
<th>Bands 1-2</th>
<th>Comments</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Introduction</strong></td>
<td>A student develops and evaluates questions and hypotheses for scientific investigation BIO11/12-1</td>
<td>Effectively uses the background information to develop a highly relevant inquiry question with limited guidance.</td>
<td>Uses the background information to develop a relevant inquiry question with some guidance.</td>
<td>Inquiry question is present but may be weak or not relevant to the background information.</td>
<td>The report names a genetic disease, or a non-infectious disease with some genetic component, though this is not well explained</td>
</tr>
<tr>
<td><strong>Questioning and predicting</strong></td>
<td></td>
<td>The report clearly identifies a genetic disease, or identifies a non-infectious disease with a strong explanation of its genetic component.</td>
<td>The report identifies a genetic disease, or identifies a non-infectious disease with a clear genetic component.</td>
<td>Inquiry question includes a medical application of genetic technology, though appropriateness for the disease is not clear.</td>
<td>Inquiry question identifies a concept that can be investigated, but it may not suit the scope of the report or an evaluation of risks and benefits</td>
</tr>
<tr>
<td></td>
<td></td>
<td>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).</td>
<td>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).</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>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.</td>
<td>Inquiry question identifies a concept that can be investigated within the scope of the report and allows for some evaluation of risks and benefits</td>
<td></td>
<td></td>
</tr>
<tr>
<td>About the disease</td>
<td>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.</td>
<td>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.</td>
<td>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.</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>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.</td>
<td>Explains the link between a genetic mutation, changes to a protein, and how these cause the disease phenotype.</td>
<td>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 missing or incorrect aspects.</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Includes data from a population genetics study to describe the prevalence and/or inheritance pattern of the disease.</td>
<td>Includes secondary data to describe the prevalence and/or inheritance pattern of the disease.</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>About the genetic technology</td>
<td>N/A</td>
<td>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).</td>
<td>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).</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>There is a concise and clear explanation of how the technique works, covering aspects that are relevant for the analysis to follow.</td>
<td>Description of the technique and application in the context of the chosen disease is attempted but may be weak.</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
## Risk and benefit analysis

*explains natural genetic change and the use of genetic technologies to induce genetic change BIO-12-13*

**conducts investigations to collect valid and reliable primary and secondary data and information BIO11/12-3**

<table>
<thead>
<tr>
<th>Level</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Excellent</td>
<td>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). 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.</td>
</tr>
<tr>
<td>Good</td>
<td>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). 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.</td>
</tr>
<tr>
<td>Fair</td>
<td>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). Evidence of limited wider reading in conducting the investigation.</td>
</tr>
</tbody>
</table>

## Conclusion

*solves scientific problems using primary and secondary data, critical thinking skills and scientific processes BIO11/12-6*

**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**

<table>
<thead>
<tr>
<th>Level</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Excellent</td>
<td>There is an insightful conclusion that provides a meaningful answer to the inquiry question, distilling key information from prior sections. Provides a very strong suggestion of the implications of the approach for the prevention or treatment of the disease, AND future directions for research.</td>
</tr>
<tr>
<td>Good</td>
<td>There is a conclusion that provides some answer to the inquiry question, making some reference to information from prior sections. Provides a good suggestion of the implications of the approach for the prevention or treatment of the disease, OR future directions for research.</td>
</tr>
<tr>
<td>Fair</td>
<td>There are some statements that link back to the to answer to the inquiry question. Provides a limited suggestion of the implications of an aspect of the approach for the prevention OR treatment of the disease OR future directions for research.</td>
</tr>
<tr>
<td><strong>Communication and referencing (across whole report)</strong></td>
<td><strong>Uses appropriate language proficiently for a specific audience and displays very high quality written communication which is clear and concise.</strong></td>
</tr>
<tr>
<td>---</td>
<td>---</td>
</tr>
<tr>
<td>communicates scientific understanding using suitable language and terminology for a specific audience or purpose BIO11/12-7</td>
<td>Spelling, grammar, and formatting of report is to a very high standard and adds to clarity of writing. There are very minor errors.</td>
</tr>
<tr>
<td>conducts investigations to collect valid and reliable primary and secondary data and information BIO11/12-3</td>
<td>Sources are reference in text consistently (following school guidelines)</td>
</tr>
<tr>
<td></td>
<td>There is a reference list that contains all sources used in text. The sources are consistently and correctly referenced with almost no mistakes.</td>
</tr>
</tbody>
</table>

[Optional: could incorporate these above]

| **Student Logbook** (attached as appendix) | There is strong evidence of how the inquiry question has been thoughtfully evaluated, revised or modified after initial research and evaluation. | There is a discussion of how the inquire question have been evaluated, revised or modified after initial research and evaluation. | There is some discussion of how the inquire question have been evaluated, revised or modified after initial research and evaluation. But it may be brief or flawed. |
| | There is a very well-developed evaluation of secondary sources for relevance, accuracy, validity and reliability. | There is an evaluation of secondary sources including relevance, accuracy, validity and reliability. Some aspects may be missing. | There are some comments as to the relevance OR accuracy OR validity OR reliability of secondary sources. This may be weak. |
| | There is peer evaluation of evidence-based argument | There is some evaluation of evidence-based argument | |