CAMBRIDGE ADVANCED NATIONALS

OCR
Oxford Cambridge and RSA

Set assignment

DRAFT

LEVEL 3 CAMBRIDGE ADVANCED NATIONAL (AAQ) IN

HUMAN BIOLOGY

Certificate H049

Extended Certificate H149

For first teaching in 2025

F172: Genetics

Version 1.0 (September 2023)

Introduction

This is Sample Assessment Material (SAM). It is an example set assignment that we publish alongside a new specification to help illustrate the intended style and structure of our set assignments.

During the lifetime of the qualification, updates to the set assignment template may happen. We always recommend you look at the most recent set of past set assignments where available.

Tell us what you think

Your feedback plays an important role in how we develop, market, support and resource qualifications now and into the future. Here at OCR, we want teachers and students to enjoy and get the best out of our qualifications and resources, but to do that we need honest opinions to tell us whether we're on the right track or not. That's where you come in.

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Designed and tested with teachers and students

Helping young people develop an ethical view of the world

Equality, diversity, inclusion and belonging (EDIB) are part of everything we do

Summary of updates

Date	Version	Page number	Summary of change
July 2023	1 DRAFT	All	Creation of document

Teacher support

We have a range of support services to help you at every stage, from preparation to delivery.

Our teacher support is designed to make teaching our qualifications straightforward, whether you are an experienced teacher, new to teaching, new to OCR, or not a subject specialist of the qualification you are teaching.

We offer free access to services such as Access to Scripts, ExamBuilder and ActiveResults, and you'll find comprehensive teaching resources and a range of professional development courses on our teacher website, Teach Cambridge.

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Contact details are available on the **final page** of the SAM.

Work with us

As one of the major UK exam boards, OCR is responsible for creating and marking exams taken by over a million students each year. We work with over 10,000 professionals to create, check, mark and moderate our assessments.

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People and planet

OCR is part of Cambridge University Press & Assessment, which has clear commitments to champion sustainability, diversity, trust and respect for our people and planet.

We are committed to supporting a curriculum that helps young people develop an ethical view of the world. This enables them to take social responsibility, understand environmental issues and prepare them for the green jobs of the future.

Our equality, diversity, inclusion and belonging principles are that we:

- are respectful and considerate
- celebrate differences and promote positive attitudes to belonging
- include perspectives that reflect the diverse cultural and lifestyle backgrounds of our society
- challenge prejudicial views and unconscious biases
- promote a safe and supportive approach to learning
- are accessible and fair, creating positive experiences for all
- provide opportunities for everyone to perform at their best
- are contemporary, relevant and equip everyone to live and thrive in a global, diverse world
- create a shared sense of identity in a modern mixed society with one humanity.

To learn more, including our work on accessibility in our assessment materials, visit our People and planet page.

Oxford Cambridge and RSA

OCR-set Assignment

Sample Assessment Material

OCR Level 3 Cambridge Advanced National (AAQ) in Human Biology (Certificate)

OCR Level 3 Cambridge Advanced National (AAQ) in Human Biology (Extended Certificate)

F172: Genetics

Scenario Title: Haemophilia (Jane) and Retinitis pigmentosa (Hugo)

This is a sample OCR-set assignment which should only be used for practice**.**

This assignment **must not** be used for live assessment of students.

The live assignments will be available on our secure website, 'Teach Cambridge'.

The OCR administrative codes linked to this unit are:

- unit entry code F172
- certification code H049/H149

The regulated qualification numbers linked to this unit are:

TBC

Duration

About:

- 15 hours of supervised time (GLH)
  (work that **must** be completed under teacher supervised conditions)
- 12 hours of unsupervised time
  (work that students can complete independently without teacher supervision)

Contents

Information and instructions for Teachers

Using this assignment

This assignment provides a scenario and set of related tasks that reflect how to research a genetic disorder, and how a genetic counsellor helps patients understand and make informed decisions about a genetic disorder.

The assignment:

- Is written so that students have the opportunity to meet the requirements of all assessment criteria for the unit.
- Will tell students if their evidence must be in a specific format. If the task does not specify a format, students can choose the format to use.
- **Must** be completed under teacher supervision. Any exceptions to this will be stated in the assessment guidance.

You **must**:

- Use an OCR-set assignment for summative assessment of students.
- Familiarise yourself with the assessment criteria and assessment guidance for the tasks. These are given at the end of each student task. They are also with the unit content in **Section 4** of the Specification.
  Assessment guidance is only given where additional information is needed. There might not be assessment guidance for each criterion.
- Make sure students understand that the assessment criteria and assessment guidance tell them in detail what they need to do in each task.
- Read and understand **all** the rules and guidance in **Section 6** of the Specification **before** your students start the set assignments.
- Make sure that your students complete the tasks and that you assess the tasks fully in line with the rules and guidance in **Section 6** of the Specification.
- Give your students the Human Biology **Student guide to NEA assignments before** they start the assignments.

You **must not**:

- Use live OCR-set assignments for practice or formative assessment. This sample assessment material **can** be used for practice or formative assessment.
- Use this sample assessment material for live assessment of students.
- Allow group work for **any** task in this assignment.
- Change any part of the OCR-set assignments or assessment criteria.

Pages 1-4 are for teachers only. Please do **not** give **Pages 1-4** to your students.

You can give **any** or **all** of the pages **that follow** to your students.

Tasks for students and assessment criteria

Unit F172: Genetics

Scenario Title: Haemophilia (Jane) and Retinitis pigmentosa (Hugo)

Scenario

You are a genetic counsellor working as part of a multidisciplinary healthcare team. You have recently had two patients referred to you by their GPs with different genetic disorders. A case study for each patient is on the following pages.

You will need to choose **one** of the case studies for the patients below:

Jane – Haemophilia

OR

Hugo – Retinitis pigmentosa

You want to provide information for the rest of your healthcare team for **one** case study. In **Tasks 1 and 2** you will create information about the genetic disorder chosen, including:

- fundamental information about the disorder
- the inheritance of the disorder
- the potential for gene therapy and genetic engineering.

In **Task 3** you will provide key information and support for the patient you have chosen.

Case Study: Jane – Haemophilia

Jane is a healthy 24-year-old woman. During a consultation with her GP, Jane described her family history of haemophilia. The GP made notes of Jane's concerns and has referred Jane for genetic counselling.

Jane's family history:

- Haemophilia has been in Jane's family on her mother's side for three generations.
- Jane was 5 years old when one of her male cousins was diagnosed. Jane's cousin needed to be careful because he could bleed a lot if he injured himself.
- One of Jane's uncles died due to bleeding complications.
- Jane's grandmother's male cousins would spontaneously bleed from their eyes, nose, and mouth.
- Jane's brother was diagnosed with haemophilia when he was 2 years old. Jane's mother first became concerned when she noticed that he had a lot of bruises. One day he tripped over in the garden and had a big haematoma on one of his cheeks. A paediatrician tested Jane's brother for haemophilia, and it was confirmed that he had the bleeding disorder.
- Jane's sister had very heavy nosebleeds and was diagnosed with mild haemophilia when she was 13 years old.

Jane has never been assessed for haemophilia. Jane and her partner are now planning to have children. Jane has decided she wants to know about how haemophilia is inherited in her family and the impact it might have on her children. Jane's partner thinks he might also have some distant family who have haemophilia, but he doesn't think that he has it himself.

Jane says that she does not know anything about genes but is keen to understand how genes have contributed to her family's health problems. Jane wants to know more about genetic testing before she agrees to having a genetic test because she is worried that her private details may be shared with others. Jane has heard of gene therapy and genetic engineering, and wonders if these may be available to help her and her family.

Case Study: Hugo – Retinitis pigmentosa

Hugo is a 26-year-old man living with a genetic disorder called retinitis pigmentosa (RP). At a recent visit to his GP, Hugo expressed concern about his future. Hugo's GP referred him to a genetic counsellor.

Hugo's medical and family history:

- Hugo was diagnosed with RP when he was 10 years old and, at the time, he and his family did not know anything about the disease. There was no history of RP in Hugo's family.
- Hugo's diagnosis started with a routine eye test where the optician noticed pigmentation on his retina. Hugo was referred to the local hospital.
- During the hospital eye test, Hugo was asked to press a button when he saw flashes of light in his peripheral vision. Hugo remembers that he waited several minutes thinking that the test hadn't begun. He had not seen any of the flashes of light in his peripheral vision.
- Hugo was referred to a specialist hospital where, following more tests, he received his diagnosis of RP. Hugo has hospital eye tests every 12 months to monitor the deterioration in his eyesight.
- Hugo has central vision but no outer vision. He has reduced night vision and struggles to see in low light, such as a dimly lit room, and at dusk and dawn.

Talking about how the condition affects his everyday life, Hugo told his GP that he manages well because he has never known any different. He was concerned, however, that his vision was getting worse. Despite the challenges, Hugo recently completed his degree at university and now has a full-time job. Hugo and his partner have bought a house and are planning to have children.

Hugo would like to know more about how his RP is inherited and the risk of his children having the disorder. Hugo has read that recent advances in gene therapies and genetic engineering technologies may mean that there may soon be a treatment available for his RP to prevent further deterioration in his vision.

Task 1

How do the fundamentals of genetics relate to the genetic disorder?

Topic Areas 1, 2 and 3 are assessed in this task.

The task is:

To provide information for your healthcare team on the fundamentals of the genetic disorder in the case study you have chosen.

- Choose **one** of the two genetic disorders to provide information about for your healthcare team.
- Research the fundamentals of the genetic disorder and how genes and DNA are affected.
- Produce information on the genetic disorder for your healthcare team.

Your evidence **must** include:

- Written evidence

Use the assessment criteria below to tell you what you need to do in more detail.

Pass	Merit	Distinction
P1: Use research to **summarise** DNA function for someone with the genetic disorder.	**M1:** Use research to **compare** the functioning gene/chromosome to the malfunctioning gene/chromosome for the genetic disorder.	**D1: Assess** how physiological processes are affected by the genetic disorder.
P2: Use research to **explain** how genes determine the signs and symptoms of the genetic disorder.	**M2:** Use research to **describe** how gene expression and gene regulation contribute to the genetic disorder.	
P3: Use research to **describe** how the genetic disorder is caused by type(s) of variation.		
P4: Use research to **describe** the mode of inheritance of the genetic disorder.		

Assessment Guidance

This assessment guidance gives you information to meet the assessment criteria. There might not be additional assessment guidance for each criterion. It is only given where it is needed. You must read this guidance before you complete your evidence.

Assessment Criteria	Assessment guidance
Task 1	• The research element of the criteria in this Task does **not** need to be completed under teacher supervised conditions but is necessary in order for students to access the criteria.
P1	• Students must use research to summarise DNA function for someone with the genetic disorder. Students must consider the impact on different sexes and at different life stages.
M1	• Students need to compare the functioning gene or chromosome to the malfunctioning gene or chromosome for the genetic disorder. Whether the focus is on 'gene' or 'chromosome' will depend on the genetic disorder.

Advice:

- Remember to clearly reference any information used from books, websites or other sources to support your evidence.

Task 2

Are gene therapy and genetic engineering options available for this genetic disorder?

Topic Area 4 is assessed in this task.

For your chosen genetic disorder, it is important for your healthcare team to know whether gene therapy or genetic engineering are options for any future patients who contact them.

The task is:

To create documentation for your healthcare team about gene therapy and genetic engineering for the genetic disorder.

- Research how relevant gene therapy is for the genetic disorder.
- Research the most relevant gene therapy.
- Produce information on benefits, risks, and challenges of gene therapy for the genetic disorder.
- Produce information on the advantages and disadvantages of exploring genetic engineering for the genetic disorder.

Your evidence **must** include:

- Written evidence

Use the assessment criteria below to tell you what you need to do in more detail.

Pass	Merit	Distinction
P5: Use research to **describe** how relevant gene therapies are for the genetic disorder.	**M3**: Use research to **describe** the medical benefits and risks of gene therapy for the genetic disorder.	**D2**: **Discuss three** advantages and **three** disadvantages of the potential for genetic engineering for this genetic disorder.
P6: Use research to **describe** how genes are altered through the most relevant gene therapy for this genetic disorder.	**M4: Analyse** the challenges involved with gene therapy for the genetic disorder.	
P7: **Explain** the method of delivery for the most relevant gene therapy for this genetic disorder.		

Assessment Guidance

This assessment guidance gives you information to meet the assessment criteria. There might not be additional assessment guidance for each criterion. It is only given where it is needed. You must read this guidance before you complete your evidence.

Assessment Criteria	Assessment guidance
Task 2	• The research element of the criteria in this Task does **not** need to be completed under teacher supervised conditions but is necessary in order for students to access the criteria.
P5	• Students must use research to describe how relevant **at least two** gene therapies are for the genetic disorder. • If at least two gene therapies are **not** relevant then there must be a description of why.
M3	• **M3** is an extension of **P5**.
M4	• Students must analyse the challenges involved with gene therapy for the genetic disorder. • The challenges might be holistic, like financial, practical or ethical considerations, or specific, like the number of genes affecting the genetic disorder, the countries the gene therapy is offered or people's understanding of the gene therapy.
D2	• Students must discuss **three** advantages and **three** disadvantages of the potential for genetic engineering for this genetic disorder. • This discussion might include, for example, exploring whether genetic engineering would be financially viable, ethical concerns, the complications of research, the impact on those who have the genetic disorder, improvements to quality of life.

Advice:

- Remember to clearly reference any information used from books, websites or other sources to support your evidence.

Task 3

What support is available for people with this genetic disorder?

Topic Areas 1 to 4 are assessed in this task.

The task is:

Produce written materials detailing key information and support for the patient in the chosen case study.

- Having completed your research on the genetic disorder, you will now produce materials detailing key information and support for the patient.
- You will need to produce information on the support available, the inheritance of the genetic disorder and how relevant gene therapy would be for the case study context.

Your evidence **must** include:

- Written evidence

Use the assessment criteria below to tell you what you need to do in more detail.

Pass	Merit	Distinction
P8: Use research to **summarise** how a genetic counsellor may be able to assist the patient.	**M5: Explain** how genetic counselling would be beneficial in the case study context.	**D3: Discuss** the relevance of gene therapies in the case study context.
P9: Explain the potential impact of the genetic disorder on the mental health of the patient.		
P10: Explain how privacy and ethical issues can be addressed for the patient.		
P11: Create diagrammatic representation(s) to show the inheritance of the genetic disorder in the case study context.	**M6: Explain** what the diagrammatic representation(s) means for the patient.	**D4: Discuss** what the diagrammatic representation(s) show about the inheritance of the genetic disorder in the case study context.
P12: Explain the type of genetic test(s) that is appropriate to diagnose the genetic disorder.	**M7: Analyse** the role of genetic test(s) in the case study context.	**D5: Assess three** available options for managing the outcomes of the genetic disorder in the case study context.

Assessment Guidance

This assessment guidance gives you information to meet the assessment criteria. There might not be additional assessment guidance for each criterion. It is only given where it is needed. You must read this guidance before you complete your evidence.

Assessment Criteria	Assessment guidance
Task 3	• In Task 3, where a criterion focuses on 'the patient' then students must focus on the patient. There is no expectation that they discuss the rest of the case study context. • In Task 3, where a criterion focuses on 'in the case study context' then students must include the whole case study context, for example, other family members, potential children, partners.
P8	• The research element of this criterion does **not** need to be completed under teacher supervised conditions but is necessary in order for students to access the criterion.
P10	• Students explain how at least **two** privacy issues and at least **two** ethical issues can be addressed for the patient. • If at least **two** privacy issues and/or ethical issues are not relevant then there must be an explanation of why.
P11, M6, D4	• For **P11**, **M6** and **D4**, students should include all relevant diagrammatic representations from Topic Area 2.2 DNA mutations as appropriate for the genetic disorder.
M5	• **M5** is an extension of **P8**.
M6	• **M6** is an extension of **P11**.
M7	• **M7** is an extension of **P12**.
D3	• Students must discuss the relevance of gene therapies in the case study context, with part of the discussion potentially being whether gene therapy is the most appropriate option or if there are other treatments available.
D4	• **D4** is an extension of **M6**.
D5	• For **D5**, **three** different options should be assessed, but the number of available options may be more than three depending on the genetic disorder. • Students are **not** required to assess more than three available options. • Options might focus on a range of factors including patient care, patient well-being, treatments and cures.

Advice:

- Remember to clearly reference any information used from books, websites or other sources to support your evidence.

NEA Command Words

The table below shows the command words that may be used in the NEA assignments and/or assessment criteria.

Command Word	Meaning
Adapt	• Change to make suitable for a new use or purpose
Analyse	• Separate or break down information into parts and identify their characteristics or elements • Explain the different elements of a topic or argument and make reasoned comments • Explain the impacts of actions using a logical chain of reasoning
Assess	• Offer a reasoned judgement of the standard or quality of situations or skills. The reasoned judgement is informed by relevant facts
Calculate	• Work out the numerical value. Show your working unless otherwise stated
Classify	• Arrange in categories according to shared qualities or characteristics
Compare	• Give an account of the similarities and differences between two or more items, situations or actions
Conclude	• Judge or decide something
Describe	• Give an account that includes the relevant characteristics, qualities or events
Discuss (how/whether/etc)	• Present, analyse and evaluate relevant points (for example, for/against an argument) to make a reasoned judgement
Evaluate	• Make a reasoned qualitative judgement considering different factors and using available knowledge/experience
Examine	• To look at, inspect, or scrutinise carefully, or in detail
Explain	• Give reasons for and/or causes of something • Make something clear by describing and/or giving information
Interpret	• Translate information into recognisable form • Convey one's understanding to others, e.g. in a performance
Investigate	• Inquire into (a situation or problem)
Justify	• Give valid reasons for offering an opinion or reaching a conclusion
Research	• Do detailed study in order to discover (new) information or reach a (new) understanding
Summarise	• Express the most important facts or ideas about something in a short and clear form

We might also use other command words but these will be:

- commonly used words whose meaning will be made clear from the context in which they are used
- subject specific words drawn from the unit content.

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