Genetics (1.3)

Cheat Sheet & Study Guide

NCEA Level 1

Chemistry & Biology

External

Updated for 2026

TRAJECTORY EDUCATION

Title

Demonstrate understanding of genetic variation in relation to an identified characteristic (AS 92022)

Credits

5 (external)

Topics covered

Genetic variation basics, Inheritance patterns, Population-level effects, Health/medical implications

Exam layout

Three questions, including 2 - 3 parts for each (aim to write between 200 - 300 words per question).

How to study for this assessment

1. **Learn the terminology** (page 3 of this Guide)
2. **Get familiar with the content** (pages 4 - 6 of this Guide)
3. **Practice** (pages 6 - 14 of this Guide)

But before we get into it...

Exam Tips & Tricks (1.3)

How to get the grade you want

- **Know your terminology:** Be fluent with key terms (gene, allele, genotype, phenotype, mutation, meiosis). These are the foundation for **Achieved** marks.
- **Use examples:** Always anchor explanations in a clear example (e.g., cystic fibrosis, sickle cell, CCR5 mutation, kurī dogs). Examiners ***love*** applied knowledge.
- **Link processes:** Don't just describe meiosis – explain how crossing over *creates variation*. This is the jump from **Achieved** → **Merit**.
- **Evaluate significance**: For **Excellence**, always ask: *Why does this matter?* Link variation to survival, population resilience, or whakapapa continuity.
- **Structure your answers as 'A/M/E' answers:**
    - Start with definitions (**Achieved**).
    - Build into explanations (**Merit**).
    - End with evaluation/significance (**Excellence**).
- **Integrate Māori context (whakapapa):** Show awareness of cultural significance - this is often rewarded in **Excellence**.

Use your time wisely

- **Time management:** If this is your only Biology/Chemistry external, you will get **3 hours** to complete this exam. If so, **for each question,** aim to spend **5 – 10 minutes planning**, then **30 – 35 minutes writing**.
- **Check against criteria:** Before leaving the exam, go back through your answers and ask yourself: Did I define (**Achieved**)? Did I explain (**Merit**)? Did I evaluate (**Excellence**)?

Quick 'Cheat codes'

- *Always* use a case study (kurī, sickle cell, CCR5).
- *Always* mention meiosis (crossing over + independent assortment).
- *Always* discuss significance (population survival, whakapapa, medical implications).

Marking schedule (overview)

Achieved	Merit	Excellence
• Define terms clearly, & describe processes simply. • Use at least one example.	• Explain how variation arises (mutation, meiosis, sexual reproduction). • Link to inheritance patterns and allele frequencies. • Show cause-and-effect (e.g., mutation → new allele → phenotype change).	• Evaluate significance: Why is variation important for survival, whakapapa, or medical implications? • Apply knowledge to unfamiliar contexts (e.g., rabbits developing virus resistance). • Integrate multiple processes (mutation + meiosis + selection).

1 Terminology

Here are the key words you should be comfortable defining for the exam.
Tip - Create flashcards to help you memorise!

Term	Definition
Gene	A segment of DNA that codes for a trait.
Allele	Different versions of a gene (e.g., dominant/recessive).
Genotype	The genetic makeup of an organism (allele combination).
Phenotype	The physical expression of the genotype (observable trait).
Chromosome	DNA packaged into thread-like structures in the nucleus.
DNA	Molecules carrying genetic instructions (double helix structure).
Mutation	A change in DNA sequence that can create new variation.
Meosis	Cell division that produces gametes, introducing variation.
Mitosis	Process of cell division.
Crossing over	Exchange of DNA between homologous chromosomes during meiosis.
Independent assortment	Random distribution of chromosomes into gametes.
Gamete	Sex cells (sperm or egg) carry half the genetic material.
Zygote	Fertilised eggs formed when gametes combine.
Population	Group of organisms of the same species in one area
Allele frequency	How common an allele is in a population.
Genetic variation	Differences in DNA among individuals in a population.
Pedigree Analysis	Chart showing inheritance of traits across generations.
DNA Profiling	Technique to identify individuals using DNA patterns.
DNA Sequencing	Determining the exact order of DNA bases.
Somatic Mutation	DNA change in body cells (not inherited).
Gametic Mutation	DNA change in sex cells (can be passed to offspring).

2 The Content

Here is a ‘cheat sheet’ style summary of the main concepts to know for your exam.

Use these to find out the topics you should be focussing more on, or for quick revision on the lead-up to the exam.

Genetic variation basics

1. **DNA & Genes**
    a. DNA is the molecule of inheritance, made of base pairs (A, T, C, G).
    b. Genes are specific sequences of DNA that code for proteins, which determine traits.
2. **Alleles**
    a. Variants of a gene. Can be dominant, recessive, or codominant.
    b. Example: Eye colour gene has alleles for brown or blue.
3. **Sources of Variation**
    a. **Mutation**: Random changes in DNA; can introduce new alleles.
    b. **Meiosis**:
        i. *Crossing over*: Homologous chromosomes swap DNA segments.
        ii. *Independent assortment*: Chromosomes randomly distributed into gametes.
    c. **Sexual reproduction**: Random fusion of gametes creates unique offspring.
4. **Significance**
    a. Variation ensures populations can adapt to environmental changes (e.g., disease resistance).

Inheritance patterns

- **Genotype vs Phenotype**
    - Genotype = allele combination (e.g., Bb).
    - Phenotype = physical trait expressed (e.g., brown eyes).
- **Punnett Squares**
    - Tool to predict offspring ratios.
    - Example: Bb × Bb → 75% brown eyes, 25% blue eyes.
- **Pedigrees**
    - Diagrams showing inheritance across generations. Useful for identifying carriers.
- **Examples of Inheritance**
    - **Recessive**: Cystic fibrosis (need two faulty alleles).
    - **Dominant**: Huntington’s disease (only one allele needed).
    - **Codominance**: Sickle cell trait (both alleles expressed).

Population-level effects

- **Allele Frequency**
    - Proportion of a specific allele in a population.
    - Can change due to selection, drift, or migration.
- **Genetic Drift**
    - Random changes in allele frequency, especially in small populations.
- **Natural Selection**
    - Advantageous alleles increase survival and reproduction.
    - Example: Peppered moth colour changed during the industrial revolution.
- **Population Bottlenecks**
    - Sudden reduction in population size → loss of variation.
- **Founder Effect**
    - Small groups colonise new areas → limited variation carried forward.
- **Significance**
    - Populations with more variation are more resilient to change.

Gene tracking & technology

- **DNA Profiling**
  - Uses repeating DNA sequences to identify individuals.
  - Applications: Forensics, paternity testing.
- **DNA Sequencing**
  - Determines exact order of bases in DNA.
  - Applications: Identifying mutations, personalised medicine.
- **Medical Uses**
  - Detects carriers of genetic diseases.
  - Diagnose conditions early.
- **Case Study: CCR5 Allele**
  - Mutation in CCR5 gene provides resistance to HIV infection.
  - Shows how variation can have major health implications.

Mutations

- **Somatic Mutations**
  - Occur in body cells.
  - Not inherited, but can cause diseases like cancer.
- **Gametic Mutations**
  - Occur in sex cells.
  - Passed to offspring, affecting future generations.
- **Effects of Mutations**
  - **Neutral**: No effect on survival (e.g., silent mutations).
  - **Beneficial**: Increase survival (e.g., CCR5 mutation).
  - **Harmful**: Cause disease (e.g., cystic fibrosis).
- **Examples**
  - Huntington's disease (dominant harmful mutation).
  - Sickle cell anaemia (codominant mutation with mixed effects).

Understanding Punnett Squares

Punnett squares are a crucial part of 1.3. Practice drawing them up, using them, and make sure that you have a strong grasp on them!

What is a Punnett Square?

- A Punnett square is a simple grid used to predict the possible genetic outcomes of offspring.
- It shows how alleles from each parent combine during fertilisation.
- Useful for visualising genotype ratios (allele combinations) and phenotype ratios (observable traits).

How to use them: step-by-step guide:

1. Identify parent genotypes
   - Example: Brown eyes (B, dominant) and blue eyes (b, recessive).
   - Parent 1: Bb (heterozygous), Parent 2: bb (homozygous recessive).
2. Draw the grid
   - A 2×2 square for a single gene cross.
   - Write one parent's alleles across the top, the other down the side.
3. Fill in the boxes
   - Combine alleles from each parent into each square.
4. Analyse results
   - Count genotypes (e.g., Bb, bb).
   - Translate into phenotypes (e.g., brown eyes vs blue eyes).

Example;

Cross a Bb individual with a bb individual;

	B	**b**
b	Bb	bb
b	Bb	bb

- Genotypes: 2 Bb, 2 bb → 50% Bb, 50% bb.
- Phenotypes: 50% brown eyes, 50% blue eyes.

Common mistakes:

- Mixing up genotype vs phenotype.
- Forgetting that dominant alleles mask recessive ones.
- Not reducing ratios to simplest form (e.g., 2:2 → 1:1).

3 Practice

Test your understanding

If you think you are comfortable with the topics, test yourself - write your best answer, and check the answer key to find your expected grade.

In addition to these practice questions, past exam questions are a great tool. Check our website for copies of past exams & answers.

Genetic Variation Basics

Q1: Explain how meiosis contributes to genetic variation in a population.

- **Achieved:**
  - Define meiosis.
  - Describe crossing over and independent assortment.
- **Merit:**
  - Explain how these processes create unique gametes.
  - Link to increased variation in offspring.
- **Excellence:**

- Evaluate why variation is important for population survival in changing environments.

Q2: Describe one way mutation can introduce genetic variation.

- **Achieved:**
  - Define mutation and give a simple example.
- **Merit:**
  - Explain how mutation creates new alleles and affects genotype/phenotype.
- **Excellence:**
  - Discuss the potential impact of mutations on population survival (beneficial, harmful, neutral).

Inheritance Patterns

Q1: A heterozygous brown-eyed parent (Bb) and a blue-eyed parent (bb) have children. Use a Punnett square to show the possible offspring.

- **Achieved:** Correct Punnett square, identify genotypes and phenotypes.
- **Merit:** Explain genotype vs phenotype relationship.
- **Excellence:** Evaluate the significance of carriers in maintaining recessive alleles in populations.

Q2: Explain the difference between genotype and phenotype using an example.

- **Achieved:** Define both terms.
- **Merit:** Use a clear example (e.g., cystic fibrosis).
- **Excellence:** Discuss how the environment can also influence phenotype expression.

Population-Level Effects

Q1: Explain how a population bottleneck can affect genetic variation.

- **Achieved:** Define bottleneck and describe reduced variation.
- **Merit:** Explain how allele frequencies change after a bottleneck.
- **Excellence:** Evaluate long-term consequences for population survival and adaptability.

Q2: Describe how natural selection can change allele frequencies in a population.

- **Achieved:** Define natural selection.
- **Merit:** Explain how advantageous alleles become more common.
- **Excellence:** Evaluate with a case study (e.g., peppered moths, sickle cell trait).

Gene Tracking & Technology

Q1: Describe how DNA profiling can be used in forensics.

- **Achieved:** Define DNA profiling.
- **Merit:** Explain how it distinguishes individuals.
- **Excellence:** Evaluate reliability and ethical considerations.

Q2: Explain how DNA sequencing can be used to identify carriers of genetic conditions.

- **Achieved:** Define DNA sequencing.
- **Merit:** Explain how sequencing reveals mutations.
- **Excellence:** Discuss implications for medical treatment and family planning.

Mutations

Q1: Compare somatic and gametic mutations.

- **Achieved:** Define both types.
- **Merit:** Explain inheritance differences (somatic not passed on, gametic passed on).
- **Excellence:** Evaluate impacts on individuals vs populations.

Q2: Using an example, explain how a mutation can be beneficial.

- **Achieved:** Give example (e.g., CCR5 mutation).
- **Merit:** Explain how mutation changes protein function.
- **Excellence:** Evaluate significance for population survival and medical applications.

Long-answer, exam-style practice questions

Question 1 – Genetic Variation in a Population

A population of birds has two alleles for feather colour: B (blue, dominant) and b (brown, recessive). Scientists notice that after a disease outbreak, the frequency of the b allele increases in the population.

Question: Discuss how genetic variation arises in this population, and explain the significance of this variation for the survival of the species.

Marking Guide:

- **Achieved:**
  - Define key terms (gene, allele, genotype, phenotype).
  - Describe processes that create variation (mutation, meiosis, sexual reproduction).
  - Identify that variation exists in feather colour.
- **Merit:**
  - Explain how meiosis and sexual reproduction create unique offspring.
  - Explain how allele frequency can change due to selection.
  - Link feather colour variation to survival in the disease outbreak.
- **Excellence:**
  - Evaluate the significance of genetic variation for long-term survival of the population.
  - Integrate multiple processes (mutation + meiosis + natural selection).
  - Discuss how variation increases resilience to environmental change.

Question 2 – Inheritance and Medical Implications

Cystic fibrosis (CF) is caused by a recessive allele. A couple are both carriers (genotype Ff). They want to understand the chances of their child inheriting CF.

Question: Explain how inheritance patterns contribute to genetic variation, and evaluate the significance of this variation for individuals and populations.

Marking Guide:

- **Achieved:**
  - Define genotype and phenotype.
  - Use a Punnett square to show possible offspring (FF, Ff, ff).
  - Describe that CF occurs when both recessive alleles are inherited.
- **Merit:**
  - Explain how carriers maintain recessive alleles in populations.
  - Link inheritance patterns to genetic variation.
  - Explain medical implications for individuals (CF symptoms, treatment).
- **Excellence:**
  - Evaluate the significance of carriers for population survival (hidden variation).
  - Discuss broader implications (genetic counselling, medical technology).
  - Integrate inheritance with population-level effects.

Practice Question 3 – Population Bottleneck

A population of rabbits is reduced to only 50 individuals after a flood. Scientists notice reduced genetic variation in the population.

Question: Discuss how genetic variation is affected by population bottlenecks, and evaluate the consequences for the survival of the species.

Marking Guide:

- **Achievement:**
  - Define genetic variation and allele frequency.
  - Describe what a bottleneck is.
- **Merit:**
  - Explain how allele frequencies change after a bottleneck.
  - Link reduced variation to increased risk of disease or extinction.
- **Excellence:**
  - Evaluate long-term consequences for population survival and adaptability.
  - Integrate concepts of genetic drift, natural selection, and resilience.
  - Discuss significance for conservation efforts.

Practice Question 4 – Gene Tracking Technology

Scientists use DNA profiling to compare extinct kurī dogs with modern breeds. They find two distinct genetic markers in kurī DNA.

Question: Explain how gene-tracking technologies can be used to study genetic variation, and evaluate their significance for understanding whakapapa and population survival.

Marking Guide:

- **Achievement:**
  - Define DNA profiling and genetic markers.
  - Describe how markers differ between individuals.
- **Merit:**
  - Explain how profiling shows relatedness and ancestry.
  - Link to understanding whakapapa.
- **Excellence:**
  - Evaluate the significance of gene tracking for cultural
  - and biological knowledge.
  - Discuss implications for conservation, medicine, and whakapapa continuity.
  - Integrate technology with population-level effects.