BIOLOGY YEAR 12 - MODULE 8 - LESSON 17

Genetic Disorders

Use this worksheet after reading the lesson to practise the key ideas and prove you can meet the success criteria.

Name _____

Date _____

Class _____

1. Key Ideas

When errors in DNA sequence or chromosome number cause non-infectious disease — from conception onwards

- Types of genetic disorders: chromosomal abnormalities and single-gene (Mendelian) disorders
- How non-disjunction produces chromosomal abnormalities

2. Success Criteria

By the end, you should be able to:

- Types of genetic disorders: chromosomal abnormalities and single-gene (Mendelian) disorders
- Examples: Down syndrome, Turner, Klinefelter, CF, PKU, Huntington's, haemophilia
- Diagnostic tools: karyotyping, NIPT, amniocentesis, newborn screening

3. Key Terms

Genetic disorder
A disease caused by an abnormality in an individual's DNA, either in chromosome number/structure or in a single gene sequence.

Chromosomal abnormality
Disorder caused by the wrong number or structural rearrangement of chromosomes (e.g. trisomy, monosomy, translocation).

Non-disjunction
Failure of homologous chromosomes or sister chromatids to separate during meiosis, producing gametes with extra or missing chromosomes.

Single-gene disorder
Disease caused by a mutation in one specific gene; follows Mendelian inheritance patterns (autosomal recessive/dominant, X-linked).

Penetrance
The proportion of individuals with a given genotype who actually show the associated phenotype. A gene with 80% penetrance means 20% of carriers will not develop the condition despite having the genotype.

Carrier
An individual who carries one copy of a recessive disease allele but does not show symptoms. Two carriers have a 25% chance of producing an affected child.

4. Activity: Build the Lesson Map

Use the lesson to complete the table. Keep answers brief but specific.

Prompt	Your answer
Main concept	
Important example	
Common mistake to avoid	
How this links to the next lesson	

5. Short Answer Questions

1. Explain this lesson goal in your own words: "Types of genetic disorders: chromosomal abnormalities and single-gene (Mendelian) disorders". Use one specific example from the lesson.

BAND 3 2 MARKS

2. Apply this idea to a new example: "Examples: Down syndrome, Turner, Klinefelter, CF, PKU, Huntington's, haemophilia". Show your reasoning clearly.

BAND 4 3 MARKS

3. Analyse why this idea matters for understanding Genetic Disorders: "Diagnostic tools: karyotyping, NIPT, amniocentesis, newborn screening".

BAND 5 4 MARKS

6. Extend: Apply the Idea

BAND 5/6 5 MARKS

A student gives a memorised answer about Genetic Disorders but does not use evidence or reasoning.

Improve the answer by writing a stronger response that uses accurate terminology, a relevant example and a clear explanation.

7. Multiple Choice

1. What is the best first step when answering a question about Genetic Disorders?

☐ A. Identify the key concept being tested

☐ B. Write every fact from memory

☐ C. Ignore the command word

☐ D. Skip examples and evidence

2. Which answer would show stronger understanding of Genetic Disorders?

☐ A. An answer with accurate terms and reasoning

☐ B. A copied definition only

☐ C. A single-word response

☐ D. An answer with no example

3. What should you do if a question asks you to explain?

☐ A. Link the idea to a reason or cause

☐ B. List unrelated facts

☐ C. Only draw a diagram

☐ D. Write the shortest possible answer

8. Success Criteria Proof

Finish with evidence that you can do each success criterion.

SUCCESS CRITERION 1

Prove that you can: Types of genetic disorders: chromosomal abnormalities and single-gene (Mendelian) disorders

BAND 3 2 MARKS

SUCCESS CRITERION 2

Prove that you can: Examples: Down syndrome, Turner, Klinefelter, CF, PKU, Huntington's, haemophilia

BAND 4 3 MARKS

SUCCESS CRITERION 3

Prove that you can: Diagnostic tools: karyotyping, NIPT, amniocentesis, newborn screening

BAND 5 4 MARKS

One thing I still need help with: