

# Genetic Diseases — Cystic Fibrosis, PKU, Huntington's Disease, Type 1 Diabetes

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

A single deleted amino acid in the CFTR protein fills a child's lungs with mucus. An expanded CAG trinucleotide repeat in the HTT gene produces a toxic polyglutamine protein that kills neurons for decades before any symptom appears. Every genetic disease tells the same story: one mutation, one altered protein, cascading consequences throughout the body.

- The gene, protein, and mechanism of disease for CF, PKU, Huntington's, and Type 1 diabetes
- Why a single amino acid change can abolish or dramatically alter protein function

## 2. Success Criteria

By the end, you should be able to:

- The gene, protein, and mechanism of disease for CF, PKU, Huntington's, and Type 1 diabetes
- The inheritance pattern (autosomal recessive or dominant) for each disease
- The specific consequences of each protein dysfunction at the organ/system level

## 3. Key Terms

### Cystic Fibrosis

Genetic, Why Doesn't Everyone in an Affected Family Have It?

### disease

also a genetic disease, but it behaves very differently — children of an affected parent have a 50% chance of inheriting

### inheritance pattern

[dominant/recessive] because

### mutant gene

sufficient to cause the disease

**protein that aggregates and**  
toxic to neurons

### Explain why genetic diseases

non-infectious and why 'genetic' does not mean 'inevitable'

## 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: "The gene, protein, and mechanism of disease for CF, PKU, Huntington's, and Type 1 diabetes". Use one specific example from the lesson.

**BAND 3** **2 MARKS**

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2. Apply this idea to a new example: "The inheritance pattern (autosomal recessive or dominant) for each disease". Show your reasoning clearly.

**BAND 4** **3 MARKS**

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3. Analyse why this idea matters for understanding Genetic Diseases — Cystic Fibrosis, PKU, Huntington's Disease, Type 1 Diabetes: "The specific consequences of each protein dysfunction at the organ/system level".

**BAND 5** **4 MARKS**

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## 6. Extend: Apply the Idea

BAND 5/6

5 MARKS

**A student gives a memorised answer about Genetic Diseases — Cystic Fibrosis, PKU, Huntington's Disease, Type 1 Diabetes 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.

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## 7. Multiple Choice

1. What is the best first step when answering a question about Genetic Diseases — Cystic Fibrosis, PKU, Huntington's Disease, Type 1 Diabetes?

- 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 Diseases — Cystic Fibrosis, PKU, Huntington's Disease, Type 1 Diabetes?

- 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: The gene, protein, and mechanism of disease for CF, PKU, Huntington's, and Type 1 diabetes**

**BAND 3** **2 MARKS**

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### SUCCESS CRITERION 2

**Prove that you can: The inheritance pattern (autosomal recessive or dominant) for each disease**

**BAND 4** **3 MARKS**

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### SUCCESS CRITERION 3

**Prove that you can: The specific consequences of each protein dysfunction at the organ/system level**

**BAND 5** **4 MARKS**

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**One thing I still need help with:**

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