6.3 Non-Mendelian inheritance

Name: _____ Class: _____ Date: _____ **Total: 23 marks**

KEY WORDS genomic imprinting

Objective

Recognise the non-Mendelian signatures and reason about parent-of-origin and repeat instability.

1 Key knowledge and worked examples

■ **The three layers**

mtDNA: maternal-only, heteroplasmy lottery. Imprinting (15q): paternal deletion → Prader–Willi, maternal → Angelman ✓. Repeat expansion: anticipation (CAG/CGG grow per meiosis).

■ **Worked example: one region, two syndromes**

Only one parental copy of 15q is active (methylation): delete the WORKING copy → disease, so the parent of origin decides the syndrome ✓.

2 Practice

2.1 Which non-Mendelian pattern affects all children of an affected mother? [3]

2.2 What parental-origin deletion gives Angelman? [3]

2.3 Define anticipation. [3]

3 Exam-style questions

3.1 A family has affected mothers, affected children of both sexes, and no male transmission over three generations (fictional).

Identify the pattern. Explain the severity spread (heteroplasmy and the meiotic bottleneck). Predict the risk to an affected woman's next child. [7]

3.2 A fragile-X family: the grandmother has a 70-CGG premutation; her daughter 150; her grandson >200 with the full syndrome (fictional).

Explain the expansion trend (premutation range 55–200; full >200). Explain why daughters of premutation carriers are at expanding risk. State the Huntington contrast (paternal CAG expansions → juvenile onset). [7]