1.12 Protein synthesis, mutations and screening

Name: _____ Class: _____ Date: _____ **Total: 27 marks**

KEY WORDS transcription • translation • codon • anticodon • frameshift • recessive • pre-implantation genetic diagnosis

Objective

Practise statements 2.13–2.18: transcription and translation, mutation types and their consequences, cystic fibrosis as the model genetic disease, and genetic screening with its ethics.

1 Key knowledge and worked examples

■ Two stages, two molecules

Transcription : in the nucleus, RNA polymerase uses the template strand; the RNA copy (after intron splicing) is mature **mRNA**. **Translation** : a **ribosome** moves along the mRNA; each **codon** pairs with the **anticodon** of a **tRNA** RNA carrying its amino acid; peptide bonds join them into the polypeptide.

■ Mutations

Substitution : one amino acid changes – or none (degeneracy), or a stop codon truncates the protein. **Insertion/deletion** / : **frameshift** – everything after the change is a different protein.

■ Cystic fibrosis

ΔF508: a three-base deletion removes one phenylalanine from the CFTR protein → faulty chloride channel → thick mucus → lung infections and poor digestion. Autosomal **recessive** : carriers (Ff) are healthy; Ff × Ff gives 1 affected in 4.

■ Screening and its ethics

Carrier testing, **pre-implantation genetic diagnosis** (PGD) with IVF, amniocentesis and chorionic villus sampling for foetuses. For every benefit (informed choice, earlier treatment) there is a risk (miscarriage risk from invasive sampling, anxiety, insurance or employment discrimination, and what a society does with the results).

■ Worked example: a probability chain

Two carriers (Ff) have a child. P(child affected) = 1/4. If they use PGD and test two embryos, P(first embryo unaffected) = 3/4, P(both unaffected) = 9/16 ≈ 56%. Show the Punnett square first, then multiply only independent events.

2 Practice

2.1 Describe the role of mRNA and of tRNA in translation. [4]

2.2 Explain why a deletion of three bases is often less harmful than a deletion of one base. [3]

2.3 State two methods of foetal genetic testing and one risk of each. [4]

3 Exam-style questions

3.1 The mRNA sequence 5'–AUG UUU GGC UAA–3' is translated. Write the anticodons that pair with the first three codons, state what the final codon does, and name the bond that forms between the first two amino acids. [5]

3.2 A couple both have a family history of cystic fibrosis and neither is a carrier by testing (both FF). Another couple are both carriers. Compare the genetic risks for their children, and explain why the carrier couple may still choose to have children naturally rather than use PGD. [5]

3.3 Hunter syndrome is caused by a dominant allele; a heterozygous father and an unaffected mother have three children. Determine the probability that all three are unaffected, showing your working, and explain how the answer would change if the allele were recessive and the mother were a carrier. [6]

Solutions

Original-task marking: M1 method, A1 accuracy, B1 reasoning.

2.1 mRNA carries the base sequence of the gene (in codon triplets) from nucleus to ribosome and is read there; tRNA molecules each carry a specific amino acid and their anticodons pair with complementary codons, delivering amino acids in sequence.

2.2 Three bases = one whole triplet, so only one amino acid is lost and the reading frame is preserved; a single-base deletion shifts every subsequent triplet, changing the entire protein beyond the mutation.

2.3 Amniocentesis – small miscarriage risk; chorionic villus sampling – miscarriage risk and some results ambiguous for the couple.

3.1 Anticodons: UAC (pairs AUG), AAA (pairs UUU), CCG (pairs GGC). UAA is a stop codon: it ends translation and releases the polypeptide. A peptide bond forms between the two amino acids.

3.2 FF × FF couples can only have FF children – no CF risk. Carrier couple (Ff × Ff): each child has 1/4 chance affected, 1/2 carrier. Reasons to proceed naturally: PGD requires IVF, which is physically demanding, costly and mostly unsuccessful per cycle; 3/4 of natural conceptions are unaffected and prenatal or newborn care has improved; personal, religious or ethical views about embryo selection.

3.3 Dominant, father Hh × mother hh: each child independently has 1/2 chance unaffected. P(all three unaffected) = $(1/2)^3 = 1/8$. If the allele were recessive and the mother a carrier: father Hh (if heterozygous) × Hh – each child 1/2 unaffected homozygous dominant, 1/4 carrier, 1/4 affected; P(all three unaffected including carriers) = $(3/4)^3 = 27/64$ – much higher than 1/8; (accept 1/8 for "no allele at all" with clear working).

Note: if the recessive father were homozygous affected (HH), all children would be carriers and none affected – state the parental genotypes you assume.