InkBiology

ANSWER BOOK

Genomes, screening and gene therapy: what a sequence is good for

Gene regulation, genomics and biotechnology · Unit B17

AQA 3.8.3, 3.8.4.2, 3.8.4.3 · OCR A 6.1.3 · Edexcel A Topic 2, Topic 6 · CAIE 19.2

5 questions · 16 marks

NAVY EDITION

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SECTION A · Short, 1 to 2 marks

A1 **B1** a sequence of bases for the organism, coding and non-coding alike. **B1** annotation locates the genes within that sequence and works out which non-coding regions do something, and it is a separate and slower job done afterwards. [2]

SECTION B · Standard, 3 to 5 marks

B1 **B1** bacterial genes are continuous, with no introns, so one gene gives one polypeptide and the coding sequence can be read straight off the DNA. **B1** human genes contain introns, which have to be identified and removed before the coding sequence is known. **B1** alternative splicing lets one human gene give several different polypeptides. **B1** large stretches of the human genome are regulatory rather than coding, and post-translational modification changes the polypeptide again after translation. [4]

B2 **B1** somatic therapy alters body cells such as bone marrow, retina or airway epithelium, whereas germ line therapy alters a gamete, a zygote or an early embryo. **B1** a somatic change is not inherited, because the cells that become sperm or eggs are untouched, whereas a germ line change is present in every cell of the resulting person, including those cells, and passes to every descendant. **B1** somatic treatment often has to be repeated, because cells that are not stem cells are replaced within weeks, whereas a germ line change is permanent and cannot be withdrawn from the family line. **B1** somatic therapy is licensed for several conditions, whereas germ line therapy in a pregnancy is prohibited by law in every country that has legislated on the question. [4]

B3 **B1** the probe is a short single strand of DNA whose base sequence is complementary to the sequence being looked for, carrying a radioactive or a fluorescent label. **B1** the sample DNA is made single-stranded by heating and the probe is added, and hybridisation occurs wherever the probe meets a complementary sequence, held by hydrogen bonds between complementary bases. **B1** unbound probe is washed away, and the label is then detected, on photographic film or under ultraviolet light, only where the probe has bound. [3]

B4 **B1** the quoted figure is the probability of obtaining a matching profile given that the person is not the source of the sample. **B1** the lawyer has reversed it into the probability that the person is not the source given that the profile matches, which is a different quantity and is not what the figure measures; reversing the two is the prosecutor's fallacy. **B1** the figure also assumes an unrelated person, and relatives share far more alleles than strangers, while a degraded sample gives a partial profile at fewer loci and the match probability weakens by orders of magnitude. [3]