4.1 Chromosomes, genes, alleles and mutations – *summary of mark schemes*

4.1.2	Define *gene*, *allele* and *genome*. Mark Scheme A. gene is a heritable factor / unit of inheritance; B. gene / sequence of nucleotides that controls a (specific) characteristic / trait; C. gene is composed of DNA; D. gene controls a specific characteristic / codes for a polypeptide / protein; E. allele is one specific / variant / alternative form of a gene; F. alleles of a gene occupy the same gene locus / same position on chromosome; G. alleles differ (from each other) by one / a small number of base(s) / base pair(s);
4.1.3	Define *gene mutation*. Mark Scheme A. gene mutation is change in a gene; B. change of base sequence; C. examples of gene mutation such as substitution;
4.1.4	Explain the consequence of a base substitution mutation in relation to the processes of transcription and translation, using the example of sickle-cell anemia. Mark Scheme A. mutation is a change in the genetic make-up; B. base substitution mutation occurs when one (nitrogenous) base in DNA chain is replaced by another; C. this is a gene mutation / change in the base sequence of a gene; D. changes triplet code / codon; E. changes the mRNA during transcription; F. different amino acid (may be) coded for / inserted; G. effect of mutation ranges from no effect / no change in amino acid sequence to drastic changes; H. (may) change protein / polypeptide / primary structure / sequence of amino acids / may code for a different protein; I. may cause sickle cell anemia / other correctly named disease / form mutation is a change in DNA sequence; J. sickle-cell anaemia involves change in gene for one of polypeptides in hemoglobin / Hb / HBA; K. GAG has mutated to GTG (on DNA); L. adenine replaced by thymine in DNA; M. transcription of DNA produces the triplet GUG instead of GAG on mRNA; N. one codon is different in mRNA; O. new codon is for valine rather than glutamic acid; P. tRNA brings amino acid to ribosome during translation; Q. different amino acid placed in polypeptide chain being formed by translation; R. the two amino acids differ in solubility / have different properties / valine causes HBS to be less soluble; S. changes the shape of hemoglobin / hemoglobin becomes less soluble and crystallizes out; T. cannot carry oxygen as well; U. red blood cells sickle / impairs blood flow; V. causes other health problems / anemia / tiredness; W. sickle cell anemia caused by two mutated recessive alleles; X. HBS allele causes sickle-cell anaemia but gives resistance to malaria;