**Understandings, Applications and Skills** (This is what you maybe assessed on)

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|  | **Statement** | **Guidance** |
| 3.1.U1 | A gene is a heritable factor that consists of a length of DNA and influences a specific characteristic. |  |
| 3.1.U2 | A gene occupies a specific position on a chromosome. |  |
| 3.1.U3 | The various specific forms of a gene are alleles. |  |
| 3.1.U4 | Alleles differ from each other by one or only a few bases. |  |
| 3.1.U5 | New alleles are formed by mutation. | Deletions, insertions and frame shift mutations do not need to be included. |
| 3.1.U6 | The genome is the whole of the genetic information of an organism. |  |
| 3.1.U7 | The entire base sequence of human genes was sequenced in the Human Genome Project. |  |
| 3.1.A1 | The causes of sickle cell anemia, including a base substitution mutation, a change to the base sequence of mRNA transcribed from it and a change to the sequence of a polypeptide in hemoglobin. | Students should be able to recall one specific base substitution that causes glutamic acid to be substituted by valine as the sixth amino acid in the hemoglobin polypeptide. |
| 3.1.A2 | Comparison of the number of genes in humans with other species. | The number of genes in a species should not be referred to as genome size as this term is used for the total amount of DNA. At least one plant and one bacterium should be included in the comparison and at least one species with more genes and one with fewer genes than a human. |
| 3.1.S1 | Use of a database to determine differences in the base sequence of a gene in two species. | The Genbank® database can be used to search for DNA base sequences. The cytochrome C gene sequence is available for many different organisms and is of particular interest because of its use in reclassifying organisms into three domains. |

**Recommended resources:**

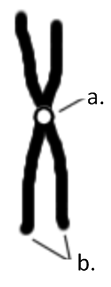
<http://bioknowledgy.weebly.com/31-genes.html>

Allott, Andrew. *Biology: Course Companion.* S.l.: Oxford UP, 2014. Print.

3.1.U1 A gene is a heritable factor that consists of a length of DNA and influences a specific characteristic. AND 3.1.U2 A gene occupies a specific position on a chromosome. AND 3.1.U3 The various specific forms of a gene are alleles. AND 3.1.U4 Alleles differ from each other by one or only a few bases.

1. State definitions of the following:  
   1. Chromosome\*
   2. Chromatid\*
   3. Gene
   4. Allele
   5. Gene locus

*\* These definitions are included for context and can be recalled from learning during 1.6 Cell division*



1. State the components of a chromosome.
2. State the number of chromosomes present in a single human diploid cell.
3. Identify the structures shown on the line drawing of a chromosome in prophase.
4. Alleles of a gene vary only slightly from each other, but can produce very different characteristics. Complete the table by using your general knowledge state two examples of genes and the possible alleles shown for each.

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| --- | --- |
| **Gene** | **Possible alleles** |
|  |  |
|  |  |

3.1.A2 Comparison of the number of genes in humans with other species.

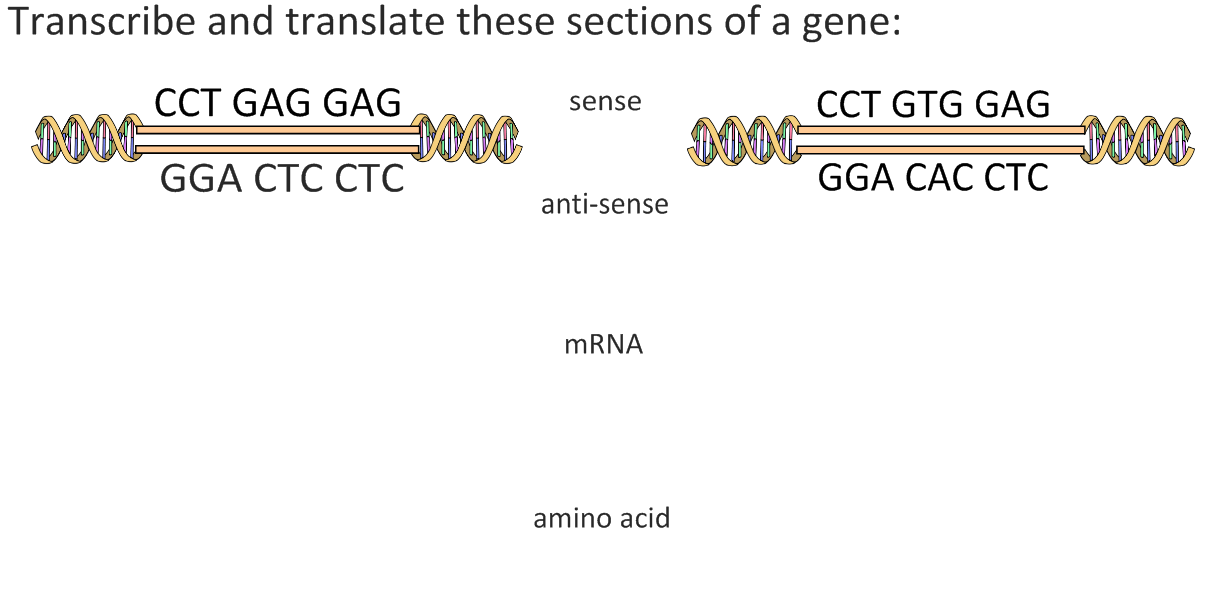
1. Although the genetic code is universal the number of genes held by different species varies greatly. State the **name** and approximate **number** of **genes** held by:  
   1. Humans
   2. A plant
   3. A Bacterium
   4. An organism that possesses more genes than humans
   5. An organism that possesses less genes than humans

3.1.U6 The genome is the whole of the genetic information of an organism. AND 3.1.U7 The entire base sequence of human genes was sequenced in the Human Genome Project. AND Nature of Science: Developments in scientific research follow improvements in technology - gene sequencers are used for the sequencing of genes. (1.8)

1. State the definition of the genome:
2. The Human Genome\* Project (HGP) was an international 13-year effort, 1990 to 2003. State it’s primary goals:

1. Key to the success of the Human Genome\* Project (HGP) was the use of gene sequencers. List the key advances in technology made their use possible.

3.1.A1 The causes of sickle cell anemia, including a base substitution mutation, a change to the base sequence of mRNA transcribed from it and a change to the sequence of a polypeptide in hemoglobin. AND 3.1.U5 New alleles are formed by mutation.

1. 

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| --- | --- | --- | --- | --- | --- | --- |
|  |  |  | mRNA |  |  |  |
|  |  |  | amino acid |  |  |  |

1. Distinguish between the two DNA strands above in terms of:
   1. DNA base sequence.
   2. Amino acid sequence in the resulting polypeptide.
2. Mutations can sometimes result in new alleles:
   1. State the definition of a mutation
   2. A base substitution mutation is a particular type of mutation. Distinguish between mutations in general and base substitution mutations.
   3. Outline how a base substitution mutation can result in a new allele.
3. Describe the effects of sickle cell disease on sufferers in terms of:
   1. Haemoglobin production
   2. Symptoms and mortality
4. Some people inherit both a normal allele (Hb A) and a sickle cell (Hb S) allele. Such people do show very few symptoms of sickle cell disease.
   1. Identify parts of the world this genotype could be beneficial.
   2. Explain your answer

3.1.S1 Use of a database to determine differences in the base sequence of a gene in two species.

One use of aligning base sequences is to determine the differences between species: this can be used to help determine evolutionary relationships.

GenBank is the NIH genetic sequence database, an annotated collection of all publicly available DNA sequences. Use it to find and extract base sequences by following the below steps:

* Go to GenBank website <http://www.ncbi.nlm.nih.gov/genbank>
* Select ‘Gene’ from the search bar
* Enter the name of a gene (e.g. AMY1A for salivary amylase 1A or COX1 for cytochrome oxidase 1) AND the organism (use the binomial) and press ‘Search’

n.b. if you are comparing species the gene chosen needs to be the same for each species

* Select the ‘Name/Gene ID’ to get a detailed view
* Scroll down to the ‘Genomic regions, transcripts, and products’ section and click on ‘FASTA’
* Copy the entire sequence from ‘>’ onwards
* Save the sequence – you will need to align with the other species next

1. Your task is to analyse the differences between three or more species (the skill asks for two species, but the online Clustal tool works better with a minimum of three). List the three species chosen and the gene you are choosing to work with:
   1. Species (give common names and the binomial)
   2. Gene (give both the name and the code)

To align the sequences:

* Go to the Clustal Omega website <http://www.ebi.ac.uk/Tools/msa/clustalo/>
* In STEP 1 Select ‘DNA’ under ‘a set of’
* Paste the chosen sequences into the box (each sequence must start on a new line)
* Press ‘Submit’ (and wait – depending on the size of the sequences you may have to wait for a couple of minutes)

Analysis:

* ‘Alignments’ allows you to visually check the results – this is easier if the chosen gene has a short base sequence
* Under ‘Results Summary’ use the ‘Percent Identity Matrix’ to quantify the overall similarity (0 = no similarity, 100 = identical)
* Under ‘Phylogenic Tree’ chose the ‘Real’ option for the Phylogram to get a visual representation of how similar the species are (based on the chosen gene).

1. State the gene and species analysed.
2. Align the sequences, analyse the results and outline your findings below:

**Citations:**

Allott, Andrew. *Biology: Course Companion.* S.l.: Oxford UP, 2014. Print.

Taylor, Stephen. " Essential Biology 4.1 Chromosomes, Genes, Alleles, Mutations.docx." Web. 16 Jul. 2015. < http://www.slideshare.net/gurustip/essential-biology-041-chromosomes-genes-alleles-mutations>.