

Chromosomes, Genes, Alleles and Mutations (4.1)

Define the following terms (4.1.2 / 4.1.3 / 4.2.2 / 4.3.1 / 4.4.11)

Gene: A heritable factor that controls a specific characteristic

Allele: One specific form of a gene, occupying the same gene locus as other alleles of the gene

Genome: The whole of the genetic information of an organism

Gene Mutation: A change in the nucleotide sequence of a section of DNA coding for a polypeptide

Homologous Chromosomes: Chromosomes that have the same structural features and the same genes

Genotype: The alleles of an organism

Phenotype: The characteristics of an organism (combination of genotype and environmental factors)

Dominant Allele: An allele that is expressed in the phenotype in a homozygous or heterozygous state

Recessive Allele: An allele that is only expressed in the phenotype in a homozygous state

Codominant Alleles: A pair of alleles which both affect the phenotype in a heterozygote

Locus: The particular position on homologous chromosomes of a gene

Homozygous: Having two identical alleles of a gene

Heterozygous: Having two different alleles of a gene

Carrier: A heterozygote who has one copy of a recessive allele that causes a genetic disease

Test Cross: Testing a suspected heterozygote by crossing with a known homozygous recessive

Clone: A group of genetically identical organisms or a group of cells derived from a single parent cell

Organisation of chromosomes (4.1.1)

- Eukaryotic chromosomes consist of DNA wrapped around histone proteins
- This forms the basic structure of the nucleosome, which are packaged into chromatin
- Chromatin containing active genes remains in an expanded form (euchromatin)
- Chromatin with genes not expressed by the cell becomes condensed (heterochromatin)
- Chromatin only supercoils into chromosomes during cell division (mitosis / meiosis)



Consequences of a base substitution mutation in sickle cell anaemia (4.1.4)

Cause:

- Base substitution: GAG → GTG
- Amino acid change: Glu → Val
- Location of change: Haemoglobin beta chain (chromosome 11)
- Mode of inheritance: Autosomal recessive

Consequence:

- Structural effect: Haemoglobin forms strands; red blood cells to become sickle-shaped
- Functional effect: Causes anaemia (low RBC count) as sickle-cells destroyed faster
Blood is less effective at transporting oxygen, causing fatigue
- Complications: Altered shape causes clotting, leading to organ failure (e.g. kidneys)

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Rachel S Tattersall



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the feed wire which connects one of the battery terminal posts to the switch the switch wire which connects the switch to the component the return wire which connects the components to the second battery terminal post either directly or indirectly through the frame of the vehicle earth return the vehicle with a metal

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oct 23 2017 master basic projects any skilled homeowner should have a basic understand of how to strip splice and join wires stripping wire is simply the process of removing insulation from the end of a wire before making a connection use a lineman s pliers to firmly strip the wire being careful not to gouge it splicing a wire is also very simple

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a general term including fittings devices appliances luminaires apparatus machinery and the like used as a part of or in connection with an electrical installation cmp 1 equipotential plane accessible conductive parts bonded together to reduce voltage gradients in a designated area

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electrical wire and circuit breakers are designed to work in tandem with one another and each must be of a proper corresponding size for example 14 2 gauge electrical wire is rated to a maximum of 15 amps and should not be used with any circuit breaker larger than 15 amps 12 2 gauge wiring is rated to a maximum of 20 amps

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