Meiosis

Zygote

- A cell formed from the fusion of the nuclei of two gametes

Meiosis

- Nuclear division
- Production of haploid cells from diploid cells
- Four daughter cells with half the chromosome number of the parent cell and reshuffled alleles
- 1 ref. to reduction division ; 2 halves the chromosome number ; 3 (so) fertilisation / fusion of gametes, gives, diploid / 2n / 40 ; 4 prevents chromosome number doubling (each generation) ;

Prophase I

- Chromosomes condense and become visible
- Arrange themselves in homologous pairs - bivalent
- Chromatid of one intertwines with other
- Crossing point called chiasma
- Centrioles migrate to opposite sides and form spindle fibres
- Nuclear envelope breaks down
- Nucleolus dissappears

Metaphase I

- Bivalents move to equator
- Held together at chiasma

Anaphase I

- Spindle fibres pull on centromeres
- Two whole chromosomes in each bivalent
- Centromeres remain intact

Telophase I

- Opposite ends of dividing cells - chromosomes
- Fibres break down
- Haploid cells formed

Cytokinesis

- Cytoplasm splits in two

Gene Determining

Explain the term F1 generation

- generation of offspring (from parental cross)
- parents are homozygous recessive and dominant
- F1 generation will be heterozygous

F2- offspring resulting from cross between two F1

Loci

- Position of a gene on a chromosome
- locus – the position of a, gene / allele, on a chromosome ;

Alleles

- different forms / variations of a gene due to different DNA base, sequence resulting in different polypeptide / protein produced

Independent Assortment

- Production of different combos of alleles in daughter cells
- Result of random alignment of bivalents during metaphase I

Gene

- Length of dna
- Codes for production of polypeptide

State three features visible on Fig. 7.1 that identify the chromosomes as a homologous pair

- have same genes
- genes at same loci
- same position of, S / centromere
- same length
- forms bivalent

Allele-alternative / different, form / version, of a gene

Crossing Over

- between non-sister chromatid
- non-sister chromatids have different combinations of alleles
- exchange alleles
- chromatids have new combinations of alleles
- linkage groups broken

- idea that gametes have unique combination of alleles
- random fusion of gametes during fertilisation

Explain how random assortment of homologous chromosomes also results in genetic variation

- homologous chromosomes / bivalents align independently of each other at the equator / metaphase plate
- this leads to different combinations of chromosomes in the daughter cells
- results in new combinations of alleles

Suggest two reasons why phenotypic ratios in the F2 generation do not always match the expected ratios.

- Epistasis
- Linkage
- Chance deviations
- Environmental effects
- Mutations
- Small sample size

Genotype - alleles possessed by organism

Phenotype - observable features of an organism

Dominant - same effect on a phenotye, regardless of if another allele is present

Recessive- only affects phenotype if no dominant is present

Epistasis- interaction of two genes at different loci, one gene may affect the expression of the other

Autosomal linkage - presence of two genes on the same autosome, do not assort independently

Codominant both alleles affect phenotype in heterozygote

Gene Control

TYR

- Albinism - missing melanin, red/pink eyes, poor vision
- DOP A / dopaquinone / melanin
- Found on long arm of chromosome 11
- Recessive gene for TYR results in absence or inactive
- Mark Scheme Response:

    - 1 (base), substitution / deletion / insertion or frame shift ; 2 ref. to change in, primary / secondary / tertiary, structure (of polypeptide / protein) or change in, 3D / active site, shape ; 3 ref. to stop codon ; 4 (so) no / inactive, tyrosinase produced ; 5 tyrosine not converted to, DOPA / dopaquinone ; 6 melanin not formed ;
    - 1 TYR codes for tyrosinase ; 2 converts tyrosine into DOPA ; 3 converts DOPA into dopaquinone ; 4 dopaquinone converted to melanin ;

HBB

- Codes for amino acid in bete globin polypeptide
- CTT replaced wih CAT
- Less soluble - molecules stick to each other - long fibres in RBC
- Sickle shape
- Useless at transporting oxygen
- Stuck in small capillaries

Outline the phenotypic effects of having abnormal haemoglobin in a person with sickle cell anaemia.

- haemoglobin less soluble
- if oxygen concentration decreases haemoglobin molecules stick together / form long fibres
- red blood cells, pulled out of shape / become sickle shaped
- blood poor at transporting oxygen so less oxygen getting to, cells / tissues / organs
- red blood cells may block vessels
- pain / sickle cell crisis / fatigue
- less oxygen for (aerobic) respiration
- cells / tissues / organs, fail to function

Suggest why a person with sickle cell trait may not show the symptoms of sickle cell anaemia

- smaller quantity of abnormal haemoglobin
- fewer red blood cells become sickled / sickling less severe

H8, factor VIII

- F8 synthesises coagulation factor VIII in liver cells
- Blood clotting
- Abnormal, less , none
- Excessive bleeding
- Non homologous region of X chromosome, sex linked

Explain why a promoter also needs to be transferred into the mammalian cells so that human factor VIII can be synthesised

- required, for gene expression to start transcription
- transcription factor can bind to promoter
- RNA polymerase can bind (to promoter) ;

HTT
- Chromosome 4
- Huntingtin protein
- Development of neurons
- Dominant allele
- Heterozygote will have disease
- Repeat of CAG triplet greater than 40
- Brain cells die more rapidly (than normal) / brain degeneration
- Involuntary movements / mental deterioration
- Mood changes
- Onset in middle ages

Explain how gibberellin acts on DELLA proteins to stimulate the production of amylase in a germinating seed
- DELLA proteins inhibit transcription factor / PIF
- Gibberellin binds to receptor in aleurone layer
- DELLA proteins broken down
- PIF, binds to promoter region (of DNA)
- Transcription of gene coding for amylase

Le gene, gibberellin and stem elongation
- Dominant Le - tall plant
- Regulates enzyme tht produces giberellin
- Stimulates cell division and elongation
- Recessive codes for alanine instead of threoine
- Non functional enzyme

Structural gene- codes for protein that has a function within a cell
Regulatory gene - codes for protein that controls expression of other genes
Operon- functional unit of transcription

Lac operon- production of lactase
LacZ- bete galactosidase
LacY- permease
LacA- Transacetylase
Contolled by same promoter, transcribed at same time

No lactose
- Regulatory gene codes for repressor

- Binds to operator region, close to gene for beta galactosidase
- Because repressor attached to operator, RNA cant bind to DNA at promoter region
- No transcription of structural genes

Lactose
- Taken up by bacterium
- Binds to repressor protein, distorting its shape, prevents binding to DNA at operator
- mRNA produced

Inducible enzyme- synthesised only when a substrate is present
- not made all the time / constitutive
- gene switched on / protein made, (only) when needed

Repressible enzyme - synthesis prevented by presence of an effector
- end-product inhibition / negative feedback / feedback inhibition

Transcription factor - molecule that affects whether or not a gene is transcribed. Expressed in correct cell at correct time and correct extent

Exam Questions and Mark Scheme responses

Using named examples, describe the differences between structural and regulatory genes and the differences between repressible and inducible enzymes.
- structural genes 1 code for, non-regulatory / structural, proteins / polypeptides ; 2 named example of structural gene ; e.g. lac Z / lac Y / lac A 3 (proteins associated with) rRNA / tRNA ; 4 (proteins such as) enzyme / named (structural) protein ; regulatory genes 5 code for, regulatory / non-structural, proteins / polypeptides ; 6 named example ; e.g. gene coding for repressor protein / lac I / PIF / correct ref. DELLA protein / gene for transcription factors 7 detail ; e.g. switches genes on or off / ref. gene expression / ref. transcription ; repressible enzymes 8 (generally) produced continuously ; 9 synthesis can be prevented by binding of repressor protein to, specific site / promoter / operator ; 10 named example ; e.g. enzyme involved in tryptophan synthesis inducible enzymes 11 synthesis only occurs when, substrate / inducer, is present ; 12 idea that transcription of the gene only occurs when, substrate / inducer, binds to, transcription factor / repressor protein; 13 named example ; e.g. β galactosidase / lactose permease / transacetylase

Explain the function of transcription factors in gene expression in eukaryotes. [
- 1 (TF) can form part of protein complex ; 2 (TF) bind to, DNA / promoter / enhancer ; 3 (so) RNA polymerase binds to promoter ; 4 (so) transcription begins / mRNA synthesised / gene expressed / gene switched on ; 5 (or TF binds to DNA) no transcription / no mRNA synthesised / gene not expressed / gene switched off ; 6 can activate genes in correct, order / time / cells / amount ; 7 ref. to correct (pattern of) development ; 8

described example ; e.g. homeobox genes / hox genes / determine sex 9 allow responses to environmental stimuli ; 10 described example ; e.g. correct genes expressed in response to, very high temperatures / light exposure 11 ref. to regulate cell cycle ; e.g. role in cell cycle checkpoints / apoptosis 12 ref. to cell signalling ; e.g. response to hormones

Describe how different types of gene mutation can cause changes in the protein that is synthesised.

- 1 base substitution / mis-sense mutation ; 2 changes, triplet / codon ; 3 base, deletion / insertion ; 4 (results in) frame shift / description ; 5 change in, protein primary structure / amino acid sequence ; 6 protein folds incorrectly / changes tertiary or 3-D structure ; 7 changes protein function / prevents protein function / makes protein unstable; 8 idea of new STOP codon ; 9 only, short / first part of / no, protein is produced ;