

Things to remember in the last hour before the exam: Level 1 Genetics

(This is not a revision sheet – you’ve done that by now – it’s a list of things you might want to remind yourself about ...)

1. DNA = heredity material of cell – contains genetic instructions used in development / functioning of proteins.
2. DNA structure – double helix – like a twisted ladder. Made of nucleotides (sugar + phosphate + base). Backbone = alternating sugar & phosphate groups; rungs = bases that pair A-T & C-G.
3. DNA replication (copying) – must be accurate – occurs before any cell division; DNA unwinds, strands separate, new nucleotides come in and pair with exposed bases by complementary base-pairing rule. Nucleotides joined by enzymes and DNA coils up; half the chromosome is original DNA, half new DNA: known as semi-conservative replication.
4. Chromosome – an (organised) structure of DNA (found in the nucleus of a cell).
5. Gene – a segment of DNA found in a small section of the chromosome that codes for a particular protein / feature / characteristic. The base sequence provides the code for building the different proteins. (Triplet code determines amino acid sequence and therefore the protein made).
6. Alleles – alternative forms of a gene: slight differences in the base sequence cause the variations in phenotypes which lead to genetic variation between individuals, e.g. red or white flowers.
7. Genetic variation – differences in phenotypes between individuals arising because of the variety of different genotypes for traits within a population.
8. Phenotype – physical expression of genotype / alleles, eg brown or blond hair.
9. Genotype – combination of alleles – for each gene there are 2 alleles, one inherited from each parent.
10. Dominant allele – allele that masks / hides the recessive allele. A dominant allele is always expressed when the individual has one or two copies of that allele. Shown using a capital letter e.g. F.
11. Recessive allele – allele that is masked by the dominant allele / only expressed if no dominant allele is present. Only expressed if two recessive alleles are present. Shown using lower case letter e.g. f.
12. 3 possible genotypes e.g. FF homozygous dominant, Ff heterozygous, & ff homozygous recessive. The 3 genotypes give 2 possible phenotypes as both FF and Ff contain a dominant allele, which masks any recessive allele.
13. Punnett square gives theoretical probabilities – actual outcomes will not necessarily match predicted outcomes, especially with small population samples. Random fertilisation of eggs by sperm means the offspring will not always match the probability predicted, unless the number of offspring is quite large.
14. Phenotype ratios – can be expressed as ratios, fractions or percentages e.g. 3 black : 1 white, OR $\frac{3}{4}$ black & $\frac{1}{4}$ white, OR 75% black, 25% white.
15. Genotype ratios – can be expressed as ratios, fractions or percentages e.g. 1 FF : 2 Ff : 1 ff, OR $\frac{1}{4}$ FF & $\frac{1}{2}$ Ff & $\frac{1}{4}$ ff, OR 25% FF, 50% Ff and 25% ff.
16. Humans have 46 chromosomes: 23 pairs.
17. Inheritance of sex: EACH fertilisation has equal chance of producing a boy or a girl. Male XY, female XX. Half the male gametes will carry X chromosome, half will carry Y. Each fertilisation is a separate event and previous offspring do NOT affect the chance of subsequent offspring being XX or XY.
18. Mitosis – cell division for growth & repair (by replacing damaged cells) – produces 2 identical daughter cells.

19. Meiosis - cell division producing gametes/sex cells which have $\frac{1}{2}$ the normal number of chromosomes (haploid) as body (somatic) cells (diploid). Produces 4 genetically different daughter cells.
20. Meiosis shuffles existing alleles because of crossing over of homologous chromosomes and independent assortment of chromosomes. Crossing over happens during the first stage of meiosis when the homologous pairs of chromosomes line up in the middle of the cell and a small pieces of DNA are swapped between them. This results in a mixing of alleles between daughter cells.
21. A gamete (egg or sperm) has half the normal number of chromosomes as body cells to ensure that when a sperm fuses with an egg the resulting zygote has the correct number of chromosomes.
22. Fertilisation - is random which male gamete fertilises the female gamete - resulting in new combinations of alleles - producing a unique zygote.
23. Sexual reproduction - produces variation between individuals due to random assortment of chromosomes in meiosis and random fertilisation.
24. Advantage of variation to a species - some individuals may survive if some change to environment / threatening event occurs, e.g. drought, disease (selection pressures). If a new disease arrives, not all individuals will be wiped out. Individuals best suited to an environment will survive to reproduce and pass on their genes to future generations.
25. Selection pressures in a changing environment (drought, temperature change, pollution, disease etc.) will affect the gene pool of species over time.
26. An individual plant/animal does NOT adapt to change; the members of the species that are most able to withstand the change survive and may pass on their beneficial genes. The species - over time - will become adapted. NOTE: plants don't become IMMUNE to disease! "Resistant" is a better word.
27. Moth example - white moths more visible than dark moths on dark backgrounds - easily preyed upon. Pollution increased - tree trunks darkened. Light coloured moths stood out - were more preyed upon, - so reduced in number. Dark moths had a beneficial characteristic (better camouflaged on dark trees) and passed this on to their offspring. Result = more dark than light moths over time.
28. Dominant or recessive trait? If two normal individuals have normal and affected offspring, the only way this is possible is for the parents to be heterozygous, e.g. Ff. When two f alleles come together, a homozygous recessive ff (affected) offspring forms. If normal was recessive, the individuals would both have to be ff and then there is no way of forming an affected individual with F in its genotype.
29. Test cross (back cross) - reproduction with a homozygous recessive mate e.g. ff to establish the genotype of an individual as FF or Ff (which have the same phenotype). FF x ff - all offspring Ff: Ff x ff - expect approx. 50 / 50 mix of phenotypes as half offspring Ff and half ff.
30. Mutation - a change to the base sequence of a gene / change in the genetic code & therefore a new protein / characteristic / trait being made. Mutations result in new alleles being created within a population. (Crossing over & independent assortment in meiosis cause a mixing of existing alleles).
31. A mutation occurring in a gamete won't affect the individual but can be passed on / inherited. The offspring have this mutation in every cell and it can be passed on in their gametes. Mutations in a somatic (body) cells e.g. a skin cell, cannot be inherited.
32. Sexual vs asexual reproduction: Sexual repro. uses meiosis - offspring genetically different - takes longer time - needs 2 parents - but offspring have better disease survivability. Asexual repro - uses mitosis - all offspring are the same - produces identical offspring, in large numbers, much more quickly - but offspring have lack of disease survivability.