Genetic Variation
ANSWERS

Evolution and Gene Pools

Evolution
STOP AND CHECK (PAGE 5)

- Evolution is the development of populations over generations, due to changes in their DNA. It occurs through reproduction within populations and results in large-scale gradual and cumulative changes in the allele frequencies of the populations' DNA.
- Evolution involves changes to the DNA of a living organism. Changes to DNA occur through reproduction, therefore evolution can only occur in new offspring through the process of reproduction.

Defining a Population
STOP AND CHECK (PAGE 6)

- Within biology, a population is a group of the same species who live in the same area and breed with each other.

Genes and Alleles: The Gene Pool
STOP AND CHECK (PAGE 7)

- A gene is a section of DNA that codes for a particular trait, such as eye colour. An allele is an expression of a gene, describing the appearance of the trait, such as having blue or green eyes.
- A gene pool is the collection of different genes available in an interbreeding population and describes the number and nature of all the alleles.

Evolution and Gene Pools

QUICK QUESTIONS (PAGE 9)

- Evolution is the process that allows changes and developments to occur within a population over time, due to reproduction between members of the population. Each population has a gene pool that is a collection of genes available when breeding between members in the population occurs. The frequency of certain alleles within the gene pool of a population will determine how often certain genes are present in members of the population.

Sources of Variation

Sexual Reproduction

STOP AND CHECK (PAGE 10)

- Through meiosis, a cell divides to create four different cells called gametes, which each have half a set of chromosomes. Meiosis is important in sexual reproduction as it ensures the gametes from each parent contain half the number of chromosomes, and have a random combination of alleles, ensuring genetic variation between offspring.
- Before meiosis can occur in the cell, the DNA in the cell must replicate itself. This ensures there are two identical copies of each chromosome in the cell, before crossing over occurs.

Crossing Over

STOP AND CHECK (PAGE 11)

- During crossing over, sections of DNA are swapped between the homologous chromosomes inside a cell. The two copies of the maternal chromosome will swap sections of DNA with the two copies of the paternal chromosome, creating new chromosomes with different combinations of alleles.

- During the process of crossing over, sections of the DNA within maternal and paternal chromosomes are switched over to create new combinations of chromosomes. This then creates new combinations of alleles, contributing to genetic variation within the population.

Independent Assortment

STOP AND CHECK (PAGE 13)

- After the crossing over of chromosomes, the chromosomes undergo independent assortment before the cell splits into two daughter cells. Segregation of the chromatids in the two daughter cells then occurs, splitting these in two and results in four non-identical daughter cells.
- During independent assortment, the homologous maternal and paternal chromosomes line up randomly at the centre of the cell.
- During independent assortment, the alignment of chromosome pairs in the centre of the cell occurs independently of each other. This ensures that when the cells then split during segregation, a random combination of maternal and paternal chromosomes end up in each cell. This creates variation between each daughter cell, so offspring are non-identical to their parents, therefore creating variation within the population.

Summary

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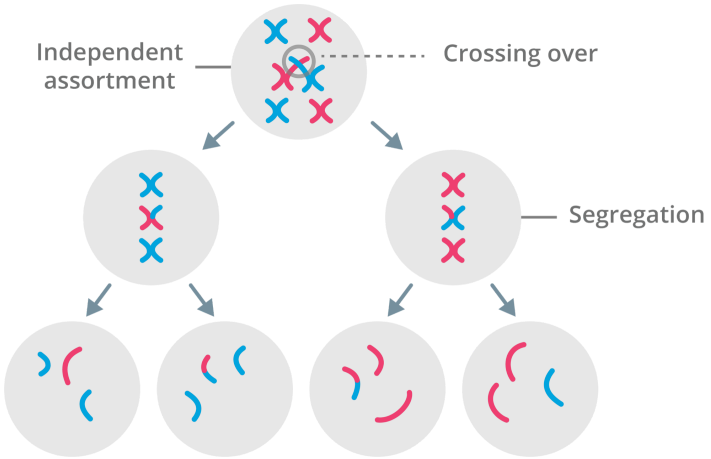

Mutations

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- Mutations are random, irreversible changes that occur in the base sequences of DNA and can create new alleles. Mutations can be both somatic, occurring in the body cells and gametic, occurring in the sex cells or gametes.
- When a mutation occurs in a cell that is about to become a gamete, it can be passed onto the offspring. If the individual is able to reproduce, this mutation allows a new allele to be added to the gene pool, which then increases the variation within the gene pool.

Sources of Variation

QUICK QUESTIONS (PAGE 16)

- New alleles can be produced in the gene pool through mutations occurring in the gametes, after the process of meiosis.
- Due to the process of meiosis, including the crossing over of sections of DNA, independent assortment of chromosomes and segregation of the cells into our daughter cells, it is almost impossible for identical cells to be produced. As each cell produced through meiosis has a random combination of DNA, it is very unlikely that two siblings will ever have the exact same DNA.
- Meiosis is the process of cell division within gametes that produces four daughter cells with a random combination of half the chromosomes of the parent cell. When two gametes fuse to form a zygote, they combine a random combination of chromosomes from both parents, resulting in an offspring that has a random combination of alleles from each parent, making them non-identical to both parents and others in the population. Gametic mutations occur when random, irreversible changes affect the base sequence of DNA in a cell before the process of meiosis begins. This also increases the genetic diversity of the population as it adds a new allele to the gene pool.
- Very little genetic diversity occurs in populations who reproduce asexually through mitosis. Genetic diversity only occurs through the introduction of mutations to the parent cell, before the division of cells occurs.

Monohybrid Inheritance

Dominance

STOP AND CHECK (PAGE 18)

- The 3 main genotypes that can be expressed are homozygous dominant, heterozygous or homozygous recessive. Both homozygous dominant and heterozygous genotypes produce a dominant phenotype, while a homozygous recessive genotype will produce a recessive phenotype.
- A dominant allele will mask the recessive allele if an individual is heterozygous for a trait. In other words, the resulting phenotype is a reflection of the dominant allele, while the recessive phenotype is only expressed phenotypically if the organism is homozygous recessive.
- The genotype for an allele is represented by using a letter. A dominant allele is expressed using a capital letter (i.e. G), while a recessive allele is expressed using a lowercase letter (i.e. g).

Genotype and Phenotype Ratios

STOP AND CHECK (PAGE 19)

- Genotype ratio = 1(BB): 1(Bb)
    - Phenotype ratio = 1(brown eyes)
- Genotype ratio = 1(HH): 2(Hh): 1(hh)
    - Phenotype ratio = 3(brown hair): 1(blonde hair)
- The real ratios don't always match the predicted ratios, as each offspring is treated as its own statistical event. Therefore, the probability of specific traits being present within each new offspring is not affected by the traits of the offspring prior. The smaller the number of offspring, the less likely the real phenotype ratio is to match the predicted one.

Codominance

STOP AND CHECK (PAGE 20)

- In a full dominant-recessive relationship there are three possible genotypes, but only two possible phenotypes. This means only the dominant allele is expressed. Whereas in a co-dominant relationship there are three possible genotypes and three possible phenotypes. In this type of relationship, both dominant alleles are equally dominant so both are expressed.

Incomplete Dominance

STOP AND CHECK (PAGE 21)

- In a full dominant-recessive relationship only the dominant allele is expressed in the individual, as this masks the recessive allele. Whereas in an incomplete dominant-recessive relationship no allele is able to mask the other allele, which results in a blended phenotype and allele.

Lethal Alleles

STOP AND CHECK (PAGE 23)

- Lethal alleles cause death in an individual when they are in their homozygous form. Whereas when they are in their heterozygous form they allow the individual to survive.
- When a lethal allele is present, there are three genotypes but only two phenotypes, as the recessive phenotype results in non-survival so is not considered in the phenotype ratio.

Multiple Alleles

STOP AND CHECK (PAGE 23)

- Multiple alleles refer to a gene that has more than two possible alleles. Although multiple alleles may be present for a gene, an individual can only ever have two alleles, one from each parent.

Monohybrid Inheritance

QUICK QUESTIONS (PAGE 24)

- If the alleles expressed complete dominance, one allele would completely mask the other. If red was dominant to white, the organism would exhibit a red hair colour.
- If the alleles were incompletely dominant, the combination of the red and white hair colour would mix. This would result in a pink phenotype being expressed.
- If the alleles were co-dominant, both the red and white hair colour would be equally expressed. This would result in an organism having both red and white hair in different patches or speckles.
- Genotype ratio = 1(FF): 2(Ff): 1(ff),
    - Phenotype ratio = 2(achondroplasia): 1(unaffected)

  As this is dealing with a lethal gene (F) the phenotype (FF) is not included as this genotype is fatal and will result in inviable offspring.

Dihybrid Inheritance

Punnett Squares

STOP AND CHECK (PAGE 26)

- Genotype ratio = 1(RRYY): 1(RRYy): 1(RrYY): 1(RrYy)
    - Phenotype ratio = 1(round and yellow)
- Genotype ratio = 1(RRYy): 1(RRyy): 1(RrYy): 1(Rryy)
    - Phenotype ratio = 1(round and yellow): 1(round and green)
- Genotype ratio = 1(RrYy): 1(rrYy): 1(Rryy): 1(rryy)
    - Phenotype ratio = 1(round and yellow): 1(wrinkled and yellow): 1(round and green): 1(wrinkled and green)
- Genotype ratio = 1(RrYy)
    - Phenotype ratio = 1(round and yellow)

Linked Genes

STOP AND CHECK (PAGE 29)

- Linked genes are those that are very unlikely to cross over during meiosis.
- Linked genes are unlikely to be separated during crossing over. This can reduce the amount of genetic variation possible in a population.

Test Cross

STOP AND CHECK (PAGE 30)

- Test crosses are used to determine the genotype of an individual who expresses a dominant trait. A test is carried out between the individual with the dominant phenotype and an individual with a recessive phenotype and can be carried out through monohybrid or dihybrid tests.
- The genotype of an individual is homozygous dominant cannot be determined to be 100% correct unless their DNA is sequenced. In a test cross, the likelihood of an individual showing as homozygous dominant can be increased by testing a very large number of offspring, all of whom need to have the dominant phenotype.

Dihybrid Inheritance

QUICK QUESTIONS (PAGE 30)

- If you wished to test an individual with brown eyes (BB) for their genotype, a monohybrid test could be conducted alongside an individual with the recessive genotype for blue eyes (bb).
- To test whether a plant is pure-breeding for round and yellow seeds (RRYY) a dihybrid test can be carried out with a plant who has the recessive genotype (rryy) and phenotype wrinkly and green.

Factors Causing Change in a Gene Pool

Natural Selection

STOP AND CHECK (PAGE 35)

- The ability of an individual to survive and reproduce.
- Populations evolve due to natural selection, so individuals who have more environmentally favourable alleles are more likely to survive and produce more offspring. Whereas individuals with less advantageous alleles are less likely to survive and produce less offspring. The term “survival of the fittest” is used due to the idea that more ‘fit’ individuals are more likely to survive and reproduce, due to their more favourable alleles.

Genetic Drift

STOP AND CHECK (PAGE 36)

- Genetic drift is the change in frequency of certain allele’s within a gene pool between generations and is completely due to chance.
- Genetic drift results in some alleles increasing in frequency within a gene pool while others decrease. When natural selection occurs, advantageous alleles tend to increase in frequency while disadvantageous alleles tend to decrease. This change in allele frequency is generally due to chance, due to the random processes of crossing over, independent assortment and segregation in meiosis.

Migration

STOP AND CHECK (PAGE 37)

- Migration involves the movement of individuals from one population to another. Emigration is the process of leaving a population while immigration is the process of joining a new population.
- When individuals immigrate and join a new population, their alleles are added to a new gene pool, which can change the allele frequency within the gene

pool. Consequently, when they leave a population they remove their alleles from its gene pool, reducing the allele frequency of the original population.

Founder Effect

STOP AND CHECK (PAGE 38)

- During the founder effect members of an original population leave to form a new population. There can then be a difference in the gene pool of the two populations or similarities due to the small group of founders originating from the larger group.
- As the founding population has fewer members than the original population, it will have a smaller gene pool which will most likely be different to the larger gene pool of the original population. This is because the small population is only a small of the larger original population, so it is likely the ratio of alleles will be different.

Bottleneck Effect

STOP AND CHECK (PAGE 39)

- During the bottleneck effect the gene pool of a population changes due to a large population loss. Due to the population loss, the gene pool will become smaller, there will be fewer alleles and the frequency of alleles may randomly change.
- As there is less population and less of a range of alleles present, there will be less genetic variation. The population will become more prone to genetic drift and it is likely that inbreeding may occur, which can reduce genetic diversity further.
- Genetic drift will have a greater effect on the population, and their generations of offspring, after the bottleneck effect occurs, as with a drastically reduced population, there will be a sudden decrease in allele frequencies.

Factors Causing Change in a Gene Pool

QUICK QUESTIONS (PAGE 39)

- Evolution is the change in the genetic makeup of a population over time, specifically the inherited traits from generation to generation. This can be affected by many events such as migration, the founder effect and the bottleneck effect. These three events may result in genetic drift within the population, causing a change in allele frequencies. With migration there may be an increase in allele frequencies in the new population through immigration, increasing the chance of evolution. Alternatively, in cases where a population is affected by emigration or the bottleneck effect, there may be fewer alleles, decreasing the chance of evolution as there may be less genetic diversity.
- Through natural selection, the population can be altered by individuals having more environmentally favourable alleles, making them more likely to survive and reproduce. These more favourable alleles would be passed on to later generations, increasing the frequency of these alleles. Alternatively in migration, an individual or group chooses to move to a more ideal population, removing their allele frequency from the original population and adding it to the new population. If they have more favourable alleles this can be beneficial for the immigrant population and detrimental to the emigrate population or vice versa if they have less favourable alleles.
- Due to natural selection, more environmentally favourable alleles may increase in frequency in a population over time, as individuals with these traits have a higher chance of survival and reproduction. Subsequently less favourable alleles may decrease in frequency as individuals carry these may have less chance of survival and reproduction, reducing their frequency in passing on to later generations. This process is generally affected by the environment in which the population exists. Whereas genetic drift is the change in frequency of alleles in a population due to chance, not an underlying factor.
- Bottleneck effect occurs when a population rapidly reduces in size, often due to an outside event. This leaves the small remaining population with little genetic diversity and less allele frequency, of which the population have not chosen therefore these alleles may not be specifically advantageous or disadvantageous. The founder effect occurs when a group decides to leave a population and form their own smaller population. As they are also a small

group, it is likely they will also be affected by little genetic diversity and less allele frequency. Both populations affected by the bottleneck and founders effect may be prone to genetic drift.

