Genetica 2 Hoofdstuk 25 samenvatting

Niek Scholten

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Chapter 25

25.1 Genetic variaton

Begrippenlijst:

- Micro-evolution: Evolutionary change within populations of a species
- Macro-evolution: Evolutionary change leading to emergence of new species and other taxonomic groups
- Population: A group of individuals belonging to the same species that live in the same geographical area
- Gene pool: The collection of all the genetic information carried by individuals of a particular population or species
- Genetic variation: The diversity of alleles and genotypes that exist within and among populations of a species
- Genetic drift: The random fluctuation of allele frequencies in a population, which can lead to the loss of genetic diversity over time

Genetic variation dan be detected using artificial selection. But the most direct way to estimate genetic variation is to compare nucleotide sequences of inividuals.

Alcohol dehydrogenase (Adh) is an example of genetic variaton that can be estimated using the comparison of nucleotide sequences. The CF gene is another good example of genetic variation, more then 1900 mutations have been identified. Causes cystic fibrosis.

The neutral theory of molecular evolution proposes that mutations leading to amino acis substitutions are usually detrimental, with only a small fraction being favorable. Some mutations are neutral; that is, they are functionally equivalent to the allele they replace.

The frequency of these alleles are determined by mutation rates and genetic drift. Another explpanation for high genetic variation is natural selection. A good example is sickle cell anemia, which is a favourable mutation in geographical areas with malaria.

25.2 Hardy-Weinberg

Begrippenlijst:

- Two-allele system: A genetic system in which a particular trait is controlled by two different alleles, often referred to as dominant and recessive
- Genetic variability: The range of genetic differences that exist within and among populations of a species
- Locus: A specific position on a chromosome where a particular gene or DNA sequence is located

The Hardy-Weinberg law describes what happens to allele and genotype frequencies in "ideal" populations. The following assumptions are made about "ideal" populations:

1. Individuals of all genotypes have equal rates of survival and reproductive succes
2. No mutation occurs
3. No migrations occurs

There are 2 main predictions from the outcome of the law: 1. Allele frequencies do not change from one generation to the next 2. Afterone generation of random mating, genotype frequencies can be predictred from the allele frequencies

The following formula is used:

p^2 + 2pq + q^2 = 1

Where: - p equals frequency of allele A - q is frequency of allele a

There are 3 additional important consequences:

1. Dominant traits to not neccessarily increase from one generation to the next
2. Genetic variability can be maintained because in the "ideal" population allele frequencies remain unchanged
3. Under the predefined assumtions, knowing the frequency of one genotype enables us to calculate the frequencies of all other genotypes on that locus

25.3 Hardy-Weinberg in humans

Begrippenlijst:

- PCR (Polymerase Chain Reaction): Laboratory technique used to amplify a specific segment of DNA through a series of temperature-dependent reactions
- Restriction-enzyme digest analysis: Laboratory technique used to cut DNA molecules into fragments at specific sites using restriction enzymes

An example of Hardy-Weinberg in humans is the analysis of the CCR5 gene. CCR5 encodes for the protein of the same name, which is a receptor for strains of HIV-1. Different combinations result in different effects:

- Homozygous: resistant to HIV-1
- Heterozygous: susceptible to infection but progress more slowly to AIDS

Calculating Hardy-Weinberg with 3 alleles is done as follows:

p^2 + 2pq + q^2 + 2pr + 2qr + r^2 = 1

Where p, q, and r represent the frequencies of the three alleles in the population, and p^2, 2pq, q^2, 2pr, 2qr, and r^2 represent the expected frequencies of the three possible genotypes (AA, AB, and BB) and their combinations (AC, BC, and CC) in the population.

To calculate the frequencies of X-linked traits using the Hardy-Weinberg equation, you need to take into account the differences in inheritance patterns between X-linked and autosomal traits.

For X-linked traits, the allele frequencies are expressed in terms of their frequencies in females, since males only have one X chromosome and will always express the allele on that chromosome.

25.4 Natural selection

Begrippenlijst:

- Natural selection: The process by which certain heritable traits become more or less common in a population over time, depending on their fitness and adaptiveness to the environment
- Fitness: The relative ability of an individual or genotype to survive, reproduce, and pass on its genes to the next generation
- Degrees of selection: The different levels at which natural selection can act, including at the level of the gene, individual, group, or species
- Directional selection: Favors individuals with a trait at one extreme of the distribution, leading to a shift in the mean of the population over time
- Stabilizing selection: Favors individuals with an intermediate trait value, leading to a reduction in the variation of the population over time
- Disruptive selection: Favors individuals with extreme trait values at both ends of the distribution, leading to a bimodal distribution and an increase in the variation of the population over time

25.5 Mutation

Begrippenlijst:

- Mutation rate: The frequency at which new mutations occur in a given gene or genome over a specified period of time

Mutation is the only process that creates new alleles in gene pool. If mutation rate is known, the extent of change to allele frequency from one generation to next can be estimated.

25.6 Migration

Begrippenlijst:

- Migration: Occurs when individuals move between populations

25.7 Genetic drift

Begrippenlijst:

- Bottleneck: A sharp reduction in the size of a population, which can result in a loss of genetic diversity and an increase in genetic drift

The founder effect is the phenomenon in which a new population is established by a small number of individuals who carry only a subset of the genetic variation present in the larger source population, leading to reduced genetic diversity in the new population.

25.8 Nonrandom mating

There are 3 types:

1. Assortative mating: Assortative mating occurs when individuals preferentially mate with partners who are similar to themselves in some characteristic, such as phenotype or genotype. For example, individuals may prefer to mate with partners who have the same eye color or blood type.
2. Disassortative mating: Disassortative mating occurs when individuals preferentially mate with partners who are dissimilar to themselves in some characteristic. This can lead to an increase in heterozygosity in the population, as individuals with different alleles for a gene are more likely to mate.
3. Inbreeding: Inbreeding occurs when individuals mate with close relatives, such as siblings or cousins. This can lead to an increase in homozygosity in the population, as recessive alleles that were previously masked by dominant alleles become expressed. Inbreeding can also increase the frequency of deleterious alleles, leading to a reduction in fitness and an increased risk of genetic disorders.

To calculate the coefficient of inbreeding (F), you need to know the pedigree of the individual and the relatedness of its parents or ancestors. F can be calculated using the following formula:

F = 1/2ˆN

Where N is the number of meioses (generations) back to the common ancestor. For example, if two individuals share a common grandparent, N would be 2.

25.9 Speciation

Begrippenlijst:

- Reproductive isolating mechanisms: Biological barriers that prevent or reduce interbreeding between populations
- Prezygotic isolating mechanisms: Barriers to reproduction that prevent individuals of different species from mating or producing viable offspring
- Postzygotic isolating mechanisms: Barriers that occur after fertilization and reduce the fitness of hybrid offspring, such as hybrid inviability (death at an early stage), hybrid sterility (inability to produce viable gametes), or hybrid breakdown (reduced fitness or fertility in later generations)

A species is a group of interbreeding individuals that are reproductively isolated from other groups. This means that individuals within a species can mate and produce viable and fertile offspring, while individuals from different species cannot.

Average time for speciation is 100,000–10 million years.

25.10 Phylogeny

Begrippenlijst:

- Monophyletic groups: An ancestral species and all its descendants
- Genetic equidistance: Differences in amino acid sequence between species are proportional to evolutionary distance
- Minimal mutational distances: The minimum number of genetic mutations that are required to transform one allele or DNA sequence into another

The molecular clock hypothesis proposes that the rate of evolution for a given DNA sequence or gene is relatively constant over time, allowing for the estimation of evolutionary time scales based on the number of mutations that have accumulated since the divergence of two lineages.