Lecture 1. Course introduction.

Genetic terminology

DNA = deoxyribonucleic acid, two strands form a double-helix

four letters = nucleotides A, C, G, T

A binds to T and G binds to C

purines A,G and pyrimidines T,C

Human nuclear genome 3 000 000 000 base pairs

mitochondrial genome 16 000 base pairs

RNA = ribonucleic acid

one strand looped, letters A, C, G, U

Proteins

twenty letters = twenty amino acids

Protein synthesis, transcription and translation:

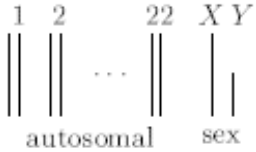
gene (a piece of DNA) $\rightarrow$ RNA $\rightarrow$ protein

Genetic code is degenerate, Table 1.1, p. 7

one codon (3 nucleotides) $\rightarrow$ one amino acid

61 codons $\rightarrow$ 20 amino acids, 3 codons $\rightarrow$ stop, $4^3 = 64$

Human nuclear DNA is packed in 23 pairs of chromosomes



XX = female

XY = male

Chromosome assortment

mother $(M_1^1F_1^1|M_2^1F_2^1|\dots|M_{22}^1F_{22}^1|M_X^1F_X^1)$

father $(M_1^2F_1^2|M_2^2F_2^2|\dots|M_{22}^2F_{22}^2|M_X^2Y)$

after meiosis and recombination

gametes $(M_1|M_2|\dots|M_{22}|M_X)$ and $(F_1|F_2|\dots|F_{22}|F_X)$

after mating

daughter $(M_1F_1|M_2F_2|\dots|M_{22}F_{22}|M_XF_X)$

Alleles: different variants of a gene

gene A with alleles (A, a), gene B with alleles (B, b)

One locus genotypes

homozygous AA, aa; heterozygous Aa

Two loci genotypes

$\frac{AB}{AB}, \frac{AB}{Ab}, \frac{AB}{aB}, \frac{AB}{ab}, \frac{Ab}{Ab}, \frac{Ab}{aB}, \frac{Ab}{ab}, \frac{aB}{aB}, \frac{aB}{ab}, \frac{ab}{ab}$

Phenotype = an observable trait of an organism

codominant alleles: AA, Aa, aa look different

Dominant allele A, recessive a

if AA and Aa look similar, while aa look different

Literature:

1. D.L.Hartl, A.G.Clarc. Principle of population genetics. Sinauer Associates, 2007.

2. R.Nielson, M. Statkin. An introduction to population genetics: theory and applications, Sinauer Associates. 2013.