## **Evolution and Medicine** Glossary for lectures and practicals

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## 1 Preface

Items below are selected to provide quick reference to students. Definitions and explanations have been selected from several textbooks (notably Stearns and Hoekstra, Evolution, an introduction, Oxford, 2000 as well as Freeman and Herron, Evolutionary analysis, Pearson, 2004), dictionaries and other sources, and edited at points by JDL. Extensive listings and explanation of evolutionary terms and further reading can be found at http://www.evolutionandmedicine.org.

## Nota bene

\* For non-native English speakers (and others), pointers below can help to prevent confusion:

- do distinguish between trait versus trade (in trade-off).
- and 'the constraint' versus 'is constrained'
- and reproduction versus reproducibility
- guard against loose use of terms such as species, organism, individual
- \* A selection of terms is listed in bold, for use in some of the exams.
- \* A few further explanations in Dutch are maintained in the text.

## 2 Glossary

Adaptation	A trait that increases the ability of an individual to survive or reproduce compared to individuals without that trait. Also the process that produces that state.
Aging	Late-life decline in an individual's fertility and probability of survival. See also: Senescence.
Allele	One of the different homologous forms of a single gene. So, a different DNA sequence at the same place (locus) in the chromosome.
Antagonistic pleiotropy	One gene has positive effects on fitness through its impact on one trait, but negative effects on fitness through its impact on another trait.
Antibiotic	Chemical or natural product with antibacterial action (bacteriocidic or bacteriostatic).

Appendix	Dutch: 'Blinde darm, wormvormig aanhangsel': dead-end extension in the gut, prone to life-threatening inflammation (appendicitis).
Atherosclerosis	Vascular disease resulting from interplay between lipid metabolism and chronic inflammation. Complications include reduced blood flow, and life threatening events such as myocardial infarction and stroke.
Bad genes	A useless term since bad genes or good genes do not exist, there are only 'quirks'. Antagonistic pleiotropy (see above) explains how genes can be beneficial in one condition (e.g. development) and deleterious in another (old age).
Bipedalism	Walking on two (hind) legs.
Coevolution	Evolutionary changes in one thing (genes, sexes, species) induce evolutionary changes in another, and vice versa (hence reciprocal adaptation). Prime example is the coevolutionary arms race between pathogens and hosts.
Convergent evolutior	Similarity between species caused by a similar, but evolutionarily independent, response to a common environmental problem.
Cystic fibrosis (CF)	Taaislijmziekte. Genetic disease leading to reduced mucus function, with a broad spectrum of subsequent disease symptoms.
EID	Emerging infectious diseases. Famous examples are AIDS, SARS and ZIKA.
Epigenetic inheritance	Somatic inheritance of the differentiated state of the cell through cycles of cell division. In other words, inheriting properties by mechanisms other than encoded by the DNA bases, for instance by methylation of DNA. See also: Genetic imprinting.
Exon	Part of a eucaryotic gene whose DNA sequence is translated into a protein (hence, no exons in procaryotes). See also: Intron.
Darwinian medicine	A basic science (not a method of practice) asking why the body is vulnerable (quote R.M. Nesse-G.C. Williams)
Darwinian fitness	The extent to which an individual contributes genes to future generations. Also: an individual's score on a measure of performance correlating with genetic contribution to future generations.
Deletion	Loss of a DNA stretch, reducing the genome size by the size of the sequence lost.
Drift	The random walk of gene frequencies that occurs in both large and small populations when variation in genes is not correlated with variation in reproductive success.
Duplication	Copying of a DNA sequence without loss of the original, increase the size of the genome by the size of the gene copied.
Evolution	Originally defined as descent with modification, or change in the characteristics of populations over time. Currently defined as changes in allele frequencies over time.

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Fitness	The extent to which an individual contributes genes to future generations. Also: an individual's score on a measure of performance correlating with genetic contribution to future generations.
Founder mutation/effect	Major changes in gene frequencies that occur in a population founded with a small sample of a larger population. Due to genetic drift in the form of sampling error in drawing founders from the source population.
Gene	Region of DNA that controls a discrete characteristic, usually corresponding to a single protein or RNA. This definition includes the entire functional unit, encompassing coding DNA sequences, noncoding regulatory DNA sequences, and introns.
Genetic bottleneck	A reduction in population size to a low-enough level for long enough that many alleles are lost and others are fixed.
Genetic drift	Random chance in allele frequencies due to chance factors.
Genetic imprinting	Genes marked by methylation of DNA in the germ line of parents. Methylated genes are not expressed in the early development of the offspring. Hence, this is a means to control DNA expression by a mechanism other than the nucleic acid sequence of DNA itself.
Genotype	In evolutionary biology, this is the information stored in the genes of one individual. In population genetics, genotype is the diploid combination of alleles at one locus present in an adult prior to meiosis. See also: Phenotype.
Genotype-by- environment interaction	Differences in the effect of the environment on the phenotype displayed by different genotypes. For instance, some people living in the same location change their skin color with the seasons and others do not.
Grandmother hypothesis of	Menopause is a life history adaptation associated with the contribution grandmothers make to feeding their grandchildren.
Group selection	Selection generated by variation in the reproductive success of groups.
Haplotype	Genotype for a collection of linked loci on a chromosome. Typically used for mitochondrial genotypes, because mitochondria are haploid and all loci are linked.
Hardy-Weinberg equilibrium	A situation in which allele and genotype frequencies in an ideal population do not change over generations, because the population does not experience selection, mutation, migration, genetic drift and random mating.
Heterozygosity	The proportion of a population that is heterozygous at a locus.
Heuristic	Helping to learn, as by a method of self-teaching. Dutch: De leer van het vinden, de wetenschap die langs methodische weg tot ontdekking of uitvindingen laat komen.
Hitch-hiking	Changes in the frequencies of neutral traits that are pleiotropically linked to other traits which are under selection. Or changes in the frequencies of neutral genes that are linked on chromosomes to changes in other genes that are under selection.

Homozygosity The proportion of a population that is homozygous at a locus.

HLA (similar to MHCHuman LeukocyteAntigens. These are the molecules that define 'self-ness'. in mice) For instance, they control acceptance or acceptance of grafts such as kidneys or skin.

- Intermediate host Species in which a microbe survives (and may reproduce and mutate), prior to transmission to a new host species. For instance, influenza toggles between birds, pigs and man.
- Intron A sequence within a gene that is removed after transcription and before translation by gene splicing. Hence, its DNA sequence is not represented in the RNA sequence of the spliced mRNA or the amino acid sequence of the protein. Introns occur in eucaryotes but not procaryotes.

Kin recognition The ability to discern the degree of relatedness of other individuals.

Kin selection (Older) relatives promote reproductive success of the (younger) relatives. In other words, adaptive evolution of genes caused by relatedness. An allele causing an individual to benefit its relative will increase in frequency if that allele is also found in the relatives, and if the benefit of the relatives more than compensates the cost to the individual.

MHC (similar to HLAMajor Histocompatibility Complex in animals. These are the molecules that define 'self-ness'. Literally: compatibility of tissues (histo). For instance, these molecules control acceptance or rejection of grafts such as kidneys or skin.

- Level of selection The level at which selection acts: species, individual, gene. This is the source of lots of confusion and lots of debate (e.g. the influential book "The Selfish Gene" by Richard Dawkins).
- Menarche Menstrual cycle (implying fertility).
- Mixing vessel A species in which different strains of a microbe can interact to form (dangerous) new variants. E.g. pigs in which pathogenic influenza variants may emerge by gene exchange.
- Myopia Near-sightedness, bijziendheid

Mutation Any change in the nucleic acid sequence of an organism, either a point mutation, a deletion, an insertion, or a chromosomal rearrangement.

Natural selection A non-zero correlation of trait variation with variation in reproductive success. In other words: A difference, on average, between the survival or reproductive success of individuals with certain phenotypes compared to individuals with other phenotypes.

Negative selection Selection against deleterious mutations (also called purifying selection).

Neutral Variation in a state is not correlated with variation in reproductive success: states are equally fit.

- Neutral evolution (neutral theory) The change and occasional fixation of alleles caused by the drift of alleles is not correlated with reproductive success. Also associated with the claim that the vast majority of observed base substitutions are neutral with respect to fitness.
- Orthology DNA sequence homology. See also: Paralogy.
- Obesitas State of being (severely) overweight.
- Paralogy DNA sequence homology plus conserved functions. See also: Orthology.
- Paleontology The study of fossil organisms.

Path dependence A certain body design has evolved along a gradual cumulative path, and there is no way back. An example is the human eye with its 'blind spot'.

- Phage A virus that infects bacteria.
- Phenotype The actual material organism (or part of it) as opposed to the information in the genotype that provides the blueprint.
- Phenotypic plasticity Sensitivity of the phenotype to differences in the environment.
- Phylogenetic trait A comparative method in which one constructs a phylogenetic tree and plots traits on the tree. One can then infer transition in traits from reading their position on the tree. For instance, one can identify the original location of ancestors by plotting geographical locations.
- Phylogeny The evolutionary history of a group. In other words, the history of a group of taxa (groups of related organisms) described as an evolutionary tree, with a common ancestor as the base and descendent taxa as branch tips.
- Pleiotropy One gene has effects on two or more traits. See also: Antagonistic pleiotropy.
- Population For sexual species, a group of interbreeding individuals and their offspring. For asexual species, a group of individuals living in the same area.
- Positive selection Selection in favor of advantageous mutations.
- Procaryotes Organisms that lack a nucleus and organelles such as mitochondria or chloroplasts. Hence, these are the Eubacteria and Archaea.
- Proximate causation The mechanical influence of traits during the lifetime of an organism, including biochemistry, development, and physiology. In other words, molecular and physiological explanations for how traits actually function.
- Proximate How does a trait work (compare to: evolutionary explanations).
- Punctuated A pattern seen in many but not all lineages in the fossil record, in which a long period of stasis is broken by a short period of rapid change. In some cases the rapid change is associated with speciation.
- Purifying selection Selection against deleterious mutations (also called negative selection)

Quirk Oddity, peculiarity. Dutch: Kronkel, spitsvondigheid, nuk, gril.

explanations

Recessive allele	An allele is recessive if it is not expressed in the phenotype in the heterozygous diploid state.
Reproductive success (RS)	The number of viable, fertile offspring produced per lifetime. It can be extended through several generations, and one can define it as the number of grandchildren that survive to reproduce.
Senescence	A decline with age in reproductive performance, physiological function, or probability of survival.
Sexual selection	The component of natural selection that is associated with success in mating.
SNP	Single nucleotide polymorphism.
Soma/somatic	Body/of the body, bodily.
Species	Biological species concept: set of organisms that could share grandchildren. Pylogenetic species concept: smallest diagnosable cluster of individual organisms with a parental pattern of ancestry and descent.
Species jump	The event that an infectious pathogen infects a new host species. A notorious example is the aids virus HIV, which is likely to originate from monkeys.
Sympatry	Occurring in the same geographic area.
Taxon	A named group of organisms (plural form is taxa).
Teleology	Teleologic - With a (final) goal or purpose.
Theory of evolution by natural selection	The hypothesis that descent with modification is caused in large part by the action of natural selection.
Trade-off	An inescapable compromise between one trait and another. Letterlijk: uitruil.
Truncation selection	Artificial selection in which only individuals with a value of a trait above or below a threshold are allowed to breed.
Ultimate causation	Explanations for why traits evolved, in terms of fitness benefits. Compare to proximate causation and proximate explanation.
Virulence	The damage inflicted by a pathogen on its host. Damage occurs because the pathogen extracts energy and nutrients from the host and because the pathogen produces toxic metabolic wastes.
Vulnerability	Kwetsbaarheid. Susceptibility to negative events.
Wild type	A term used in classical genetics to designate the standard genotype in the population from which mutations formed rare deviations. In other words, a phenotype or allele common in nature. Modern molecular data have destroyed the concept by demonstrating so much variation that the concept became meaningless.
Zoonosis	A human infectious disease in which an animal reservoir of the pathogen is

critical, e.g. influenza (birds).