IMPLICATIONS OF ANTHROPGENY FOR MEDICINE AND HEALTH

Glossary

MEDICINE & HEALTH

Atherosclerosis: Build-up of cholesterol and inflammation in the lining of blood vessels.

Bayesian: Methods in probability and statistics named after Thomas Bayes (1702-61) in which a quantity is assigned to represent a state of knowledge, or a state of belief.

Beckwith-Wiedemann Syndrome: An overgrowth disorder caused by an imbalance in sex-specific modification of chromosomes and characterized by higher risk of childhood cancer and certain congenital features.

Carcinoma: A cancer of the epithelial tissue of the lining of internal organs or the skin.

Cardiovascular Disease: Conditions of the heart that include diseased vessels, structural problems, and blood clots (sometimes used synonymously with Atherosclerosis).

Coronary Thrombosis: Blockage of blood flow to the heart, caused by atherosclerosis and blood clotting in a coronary artery. The most common kind of heart attack.

Cardiomyopathy: An acquired or hereditary disease of heart muscle resulting in weakening, enlargement, thickening, or rigidity of the heart.

Cholera: A bacterial disease causing severe diarrhea and dehydration, usually spread in sewage-contaminated water.

Chronic Mountain Sickness: A disease characterized loss of adaptation to high-altitude hypoxia. Signs include severe polycythemia (increased blood volume occupied by red blood cells) and hypoxemia (lack of oxygenation).

Commensal: A relationship between organisms where one derives food or other benefits from the other without hurting or helping it.

Comparative Method: A method of evolutionary analysis that uses comparisons across independently evolved species, as a means for studying historical and physical constraints.

Disease Phenotype: Outwardly apparent effects of a disease.

Evolutionary Medicine: The application of modern evolutionary theory to understanding health and disease.

Falciparum Malaria: Human-specific (malignant) malaria caused by the protozoan parasite, *Plasmodium falciparum*.

Health Disparity: Differences in the burden of disease, injury, violence, and health care as experienced by socially disadvantaged populations.

Homeostasis: A relatively stable equilibrium between interdependent elements, especially as maintained by physiological processes.

Hygiene Hypothesis: A lack of early childhood exposure to infectious agents, symbiotic microorganisms and parasites believed to increase susceptibility to allergies and autoimmune diseases.

Hypoxia: Less than the normal amount of oxygen reaching the tissues; also, low partial pressure of oxygen at high elevations (hypobaric hypoxia).

Intrauterine Life: The interval of life between conception and birth.

Maintenance and Defense: An organism's way of maintaining its body and physiological homeostasis while also defending against parasites, pathogens, and internal crises (e.g. cancer).

Meningococcus (Neisseria meningitidis): A bacterium that can cause meningitis and meningococcemia, a life-threatening infection in the bloodstream (sepsis).

Muscular Dystrophy: A group of genetic diseases that cause progressive weakness and loss of muscle mass.

Obstetric Dilema: A biological constraint of bipedalism and large fetal brains imposed on the human female pelvis.

Pathogen: A bacterium, virus, or other microorganism that can cause disease.

Pathophysiology: Disordered physiological processes associated with disease or injury.

Pneumococcus: A bacterium that infects the lungs and sometimes the blood stream.

Prader-Willi Syndrome: A genetic disorder usually caused by deletion of part of chromosome 15 inherited from the father, causing imbalance in sex-specific imprinting. Results in behavioral problems, intellectual disability, and short stature.

Reproductive Debut: Age at which an individual initiates their reproductive career, influences lifetime reproductive success.

Preeclampsia: A pregnancy associated disorder characterized by high blood pressure and large amounts of protein in urine, typically accelerating during the third trimester.

REM sleep: Rapid eye movement sleep, a phase of mammalian sleep characterized by random movement of eyes, low muscle tone, and vivid dreams.

Sialic Acids: Acidic molecules prominently found at the outermost fringes of the forest of sugar chains that cover all vertebrate cells.

Silver-Russell Syndrome: A complex genetic disorder affecting growth.

Typhoid fever: A bacterial infection of *Salmonella typhi* spread through contaminated food, water, or close contact.

Zoobiquity: A species-spanning approach to medicine that recognizes that animals and humans get many of the same diseases, yet physicians and veterinarians rarely consult one another.

EVOLUTION & ANTHROPOGENY

Acclimatization: A reversible change in a biological characteristic contributing to maintaining homeostasis during exposure to an environmental stress.

Admixture: Breeding between isolated populations.

Denisovans: An extinct hominin population contemporary with Neandertals that hybridized with ancient humans. Knowledge of Denisovan morphology is based on two small fossils found in Siberia. **Developmental adaptation:** An irreversible biological characteristic acquired during growth and development in a stressful environment.

Gene Flow: Movement of alleles between populations.

Genetic adaptation: A biological characteristic with a heritable basis that improves reproduction and/or survival and results from evolution by natural selection.

Genetic Drift: Loss of alleles by chance.

EVOLUTION & ANTHROPOGENY - CONTINUED

"Great Apes": A taxonomic family that was once incorrectly used to denote chimpanzees, bonobos, gorillas and orangutans, but not humans.

Hominid: A classification comprising all modern and extinct "Great Apes" and humans.

Hominin: A classification comprising modern and extinct human species, and all immediate ancestors (includes the genera *Homo, Australopithecus, Paranthropus* and *Ardipithecus*).

Homo: The genus that comprises the species *Homo sapiens*, which includes modern humans, as well as several extinct species classified as ancestral to or closely related to modern humans.

Homo erectus: An extinct hominin species with fossil evidence from 1.9 million (possibly earlier) to 70 thousand years ago and found from Africa to Indonesia. May have been the first hominin to leave Africa. *H. erectus* DNA may be retrievable from other species due to archaic admixture.

Hybridization: Breeding among recognized species.

Introgression: Transfer of alleles between species.

Middle Pleistocene: A period of geological time (781-126,000 years ago). An important time for the diversification of hominins, including the emergence of Neanderthals and *Homo sapiens*.

Morphology: Shape or form (outward appearance) of an organism.

Neanderthals: An extinct Eurasian hominin species that existed from 500,000 to 30,000 years ago and hybridized with ancient humans.

Phylogeny: Historical relationships of species or loci.

Polymorphism: An allelic difference observed in more than 1% of the population studied.

Population: A defined group of similar individuals among whom interbreeding occurs.

Selection: Allele frequency change over time caused by the different replication rate of specific alleles.

GENETICS & GENOMICS

Allele: Alternative variant gene forms at the same locus (location on the chromosome).

Allele Frequency: The proportion of all alleles within a population that are a particular type.

Allosomes: Chromosomes that determine sex (XY, with Y-Chromosome inherited paternally).

Autosomes: All other non-allosomal chromosomes. Do not differ between the sexes.

Chromatin: DNA wrapped around histone proteins.

Chromosomes: Discrete strands of packaged DNA.

Codon: A sequence of three nucleotides along a DNA or RNA chain encoding a single amino acid.

Derived Alleles: Variants arising since last common ancestor. **Diploid:** Two sets of paired chromosomes.

DNA: The molecule of inheritance, consisting of sequences of the four nucleotide building blocks (ATGC).

Enhancer: Short region of DNA that can be bound by proteins to alter transcription of a gene.

Epigenetic: Information not encoded directly in DNA.

Epigenome: Molecular modifications of the DNA and its associated histone proteins, affecting its function.

Euchromatin: Open chromatin, allowing DNA information to be read.

Fixed Alleles: Replaced all other alleles in a population. **Gene**: DNA whose information encodes a function.

Gene Regulation: Alterations of gene expression/activity. **Genetics:** The study of genes and their inheritance.

Genome: All DNA in a cell. Also refers to the DNA sequence that typifies an individual or species.

Genome Wide Association Study (GWAS): An approach for "gene mapping" in which hundreds of thousands of SNPs are tested statistically for genetic associations with a phenotype.

Genomics: The study of genome structure/function.

Genomic Imprinting: Modification of the genome at the level of

DNA (e.g. methylation) or its packaging into chromatin (histone tail modification via phosphorylation, acetylation, ubiquitination or glycosylation).

Genotype: The two alleles at one or more diploid loci.

Haploid: One set of unpaired chromosomes.

Haplotype: A set of alleles at distinct positions in the genome which are inherited together.

Heterochromatin: Tightly wrapped and inactive chromatin.

Heterozygotes: Have two different alleles at a locus.

Homozygotes: Have two identical alleles at a locus. Individuals in Haplogroups share a given haplotype.

Histones: Chief protein components of chromatin and can be chemically modified as part of epigenetics.

Karyotype: Chromosome number in the cell nucleus.

Mitochondrial DNA (mtDNA): Maternally inherited DNA found only in mitochondria (the energy generators of cells).

Mutation: Change of a DNA sequence.

Phenotype: Observable traits of an organism (result from interactions between genes and environment).

Polymorphism: An allelic difference observed in more than 1% of the population studied.

Post-translation Modifications: Alter mature protein.

Promotor: Region of DNA that initiates transcription of a particular gene.

Recombination: Exchanges between chromosomes that causes independent inheritance of alleles.

Sequence: The linear order of the building blocks, which encodes individual form and function.

Single Nucleotide Polymorphisms (SNPs): Single nucleotide differences (e.g. A vs. T).

Transcription: DNA sequence converted into RNA.

Transcription Factor: A factor (most often a protein) that influences the transcription of other coding sequences.

Translation: mRNA converted into a protein sequence. **Variant**: DNA that differs among groups studied.

This glossary is the product of the Anthropogeny Graduate Specialization students, CARTA faculty and staff, the symposium speakers, and Stevan Springer (UC San Diego).