

Winter 2023 ANBI 139 Evolution of Human Disease

Glossary of Terms

Running list, to be complemented with your questions

Anthropocene: The proposed geologic epoch defined by human influence on the Earth. There is yet to be consensus for when the anthropocene began with suggestions ranging from the start of the **agricultural revolution** to the first atomic explosion.

Holocene: The current geological epoch, from about 11.7 kya (after the end of the last Ice Age cycle) to the present that is marked by globally warmer and more stable climates.

Morphology: the shape of an animal, or one of its organs

Genetics: the study of inheritance, DNA and its functions

Holocene: The epoch of the last 11 thousand years

Heritable variation: Variation biology and behavior that is inherited from one or both parents

Differential reproductive success: The difference between one individual's lifetime reproductive success and that of the average individual in the population

Population bottleneck: The dramatic reduction in population size, which often results in a loss of **genetic diversity**.

Cell: biological unit consisting of a lipid membrane covered by a glycocalyx and containing DNA and cytoplasm.

Organism: Living entity that can reproduce and evolve.

Genome: The totality of the genetic material in an organism.

Nucleic acids: The molecules of inheritance, DNA and RNA

Proteins: "Building blocks of life" proteins consist of chains of connected amino acids and are directly encoded by genes in the DNA of organisms. Short proteins are called peptides. Most proteins fold and form complex 3-D structures.

Glycans: Mono- oligo – or polysaccharides, unlike proteins glycans can be branched.

Lipids: Fats consisting of fatty acids and diverse modifications

Soma: "Body" the cells that make the body of a sexually reproducing organism.

Germ line: The cells of an organism that give rise to gametes (sperm and eggs).

Phylogenetic tree: Graphic depiction of lineages (past populations) of organisms.

Phylogeny: The history of lineages or organisms

Convergent evolution: Independent evolution giving rise to similar morphology or adaptations.

Horizontal Genetic Exchange (Gene Flow): genetic exchange across distinct lineages of organisms: e.g. from bacteria to eukarya.

Disease: A disorder of structure or function in a human, animal, or plant, especially one that produces specific symptoms or that affects a specific location and is not simply a direct result of physical injury

Genetic cause: Disease caused by inherited DNA mutations

Environmental cause: Disease caused by environmental effects (exposure to UV light)

Byproducts of defense: Diseases resulting from immune responses

Disease of homeostasis: Diseases caused by breakdown of homeostasis

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Lack of maintenance: Diseases of “wear and tear”

Stochastic developmental problems: Diseases due to mishaps in development.

Reproductive disease: Diseases linked to mother-offspring or father-mother conflicts.

Parasite: Organism that benefits by harming its host.

Pathogen: Organism that causes disease in the host.

Symbiont: Organism that benefits from the host and also benefits the host.

Protozoan: One-celled organism that cannot photosynthesize.

Bacteria: One celled organisms without a nucleus and with complex glycan shields.

Helminth (worm): Multicellular invertebrate animal with tube like appearance.

Virus: Organism that cannot replicate without hijacking the services of a living cell.

Tuberculosis: Wide-spread disease caused by the bacterium *Mycobacterium tuberculosis* that infect about 1/3 of all humans but only causes disease in some hosts.

Achondroplasia: A developmental growth disorder leading to extremely short stature.

Malaria: Group of diseases caused by protozoa of the genus *Plasmodium*. The parasite reproduces in mosquitos and gets transmitted to humans and other mammals when mosquitos feed of mammalian blood.

Sickle cell anemia: A blood disorder caused by mutations that also convey resistance to malaria.

Prion: Glycoprotein of unknown function that can miss-fold. The miss-folded form can cause other prion proteins to miss-fold leading to brain disease.

Cannabis Use Disorder (CUD): A newly classified disease of substance abuse involving cannabis products.

Cystic Fibrosis: A genetic disease involving challenges in ion transport and causing mucus accumulation and secondary infections in the lungs.

Obesity: Disease of homeostasis defined as a BMI larger than 30.

Eating disorders: Mental disorder defined by abnormal eating behaviors that negatively affect a person's health.

Hysteria: A non-existing disease that was commonly used as diagnosis for a range of behavior in women.

Drapetomania: A fictitious disease of enslaved people escaping from plantations.

Lactase persistence: Continued expression of the digestive enzyme lactase, allowing adult humans to digest the disaccharide lactose in mammalian milk.

Poliomyelitis: Virus infecting humans and other primates through the gut and affecting the central nervous system, where host immune responses often cause damage leading to various forms of paralysis.

Haploid: Containing only a single copy of the genome (sperm and egg).

Diploid: Containing two copies of the genome (all somatic cells).

Chromatin: DNA wrapped around histone proteins.

Histone: Positively charged proteins that act as “spools” for genomic DNA.

Protamines: Positively charged proteins that help condense the DNA in sperm.

Chromosome: Segments of DNA wrapped around histones, each species of animal has a characteristic number of chromosomes (humans have 46, chimpanzees have 48).

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Karyotype: Characteristic number of chromosomes of a species.

Gene: Functional stretch of DNA.

Locus: Site/location on the DNA.

Allele: Variant of a stretch of DNA, diploid animals have two alleles at each locus.

Haplotype: Unique combination of alleles across neighboring loci

Protein coding gene: A stretch of DNA encoding a peptide or protein

RNA gene: Stretch of DNA encoding RNA with a function other than coding for protein.

Microbe: Organism that is not visible to the naked eye.

Culture: (from Latin *cultivare*=to plant/grow) behavior and/or beliefs that are transmitted and shared across individuals.

Cultural universals: Cultural traits that have been observed in all human populations

Genomic instability: Loss of stability of a genome as can happen after viral infection or during cancer.

Germ theory of disease: Theory stating that disease is caused by invisible microbes/germs that can infect their hosts.

Anthrax: Bacterium associated with animal carcasses that causes black scabs on the skin and often leads to fatal disease.

Antiseptic: Practices and methods that prevent the introduction of microbes

Pasteurization: Heat treatment that kills all microbes and extends the shelf life of food.

Immune surveillance: the processes by which immune cells “patrol the body and detect non-self molecules as potential danger signs of infection and/or cancer.

Memory B cells: Type of circulating white blood cell that can retain the memory for antigens (foreign molecules) it has encountered many years earlier during a prior infection or vaccination.

T cells: White blood cells that are educated in the Thymus and act as scouts and first line of defense by killing suspicious looking cells.

Neutrophils: White blood cells that patrol the periphery of the body and attack any suspicious looking entities.

Macrophages: White blood cells that can gobble up foreign cell, viruses and molecules or clean up debris from dead or sick cells.

Dendritic cells: White blood cells that act as sentinels and report back to the lymph nodes on what molecules they have encountered throughout the body.

Blood: Unique liquid tissue circulating through the blood vasculature of animals. It functions mainly in gas exchange but is “supercharged” as it will clot upon contact with air to prevent loss of blood and initiate healing after injury. It also allows immune cells to reach most parts of the body and transmits hormonal signals.

Clotting: the activation of a rapid cross-linking of liquid proteins resulting in goeey blood clots.

Serum: Yellowish liquid left after blood has been caused to clot in test tube, it still contains all the antibodies.

Plasma: Liquid portion of blood (minus all the cells), still containing unclotted clotting factor.

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Mucus: Various gel-like secretions lining most epithelia in the body. Mucus consists mainly of mucin glycoproteins, which consist mostly of sialic acid rich glycans and a long core protein that are highly hydrated (biogel).

Blood Brain Barrier: Barrier preventing large molecules from passing from the blood into the central nervous tissue. The barrier consists mostly of special tight junction sealing the space between the endothelial cells lining the blood vessels.

Innate immune system: the part of the immune system that is encoded in the genome.

Adaptive immune system: Part of the immune system that can “learn” about novel molecules and uses somatic recombination to generate molecular probes (antibodies) to recognize novel molecules.

Humoral immunity: Immunity mediated by blood and body fluids (antibodies and complement)

Cellular immunity: Immunity mediated by immune cells.

Hematopoietic stem cells: Stem cells that give rise to all cells of the blood and platelets.

Stem cells: cells that can generate any tissue type.

Innate immune receptors: Receptors that can recognize telltale molecules of self and non-self.

NETs (neutrophil extra cellular traps). Chromatin and anti-bacterial proteins ejected from neutrophils as defensive traps.

Clonal selection: The notion that a clone of immune cells gets selected upon recognizing an antigen.

Antibody: Protein (immunoglobulin) that recognizes foreign molecules and tags these for immune destruction.

Antigen: Molecule that can be recognized by the immune system.

Immune Organ: an organ contributing to the function of the immune system.

HLA/MHC: Group of genes encoding important self-identification proteins: these sample the content of a cell and present sampled molecules to T-cells for inspection.

Microbiome: Complete collection of microbes living in or on an animal.

Milk oligosaccharide: Modified lactose oligosaccharides that are made by the mammary glands and provide nutrition and protection to the infant.

Louse: an insect that lives on hair, clothing or pubic hair and sucks the blood of its host.

“Old Friends”: Parasites that have existed with humans for million of years.

Generation time: Time it takes for an organism to reach sexual maturity and reproduce.

Red queen Effect: Notion that hosts have to run just to stay in one place due to the rapidity of the evolution of their parasites.

Sexual recombination: Shuffling and halving of the two parental genomes during the production of sperm and eggs.

Two-fold cost of sex: Notion that obligate sexual reproduction requires males and females, and that good genotypes cannot persist fro one generation to the next.

Viral sex; Notion that viruses can recombine their DNA or RNA when more than one virus infect the same host cell.

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Sexually transmitted infection: Infections passed on during sexual behavior, they can be non-symptomatic but still be passed on.

Herd immunity: Phenomenon of protection of susceptible individuals if these are surrounded by a sufficient number of resistant individuals in a herd/population.

Glycan heterogeneity: Oligosaccharides attached to glycoproteins vary between proteins, cells, tissues, individuals, populations and species.

ABO histo-blood group: Stable system of glycan differences producing individuals with different composition and distribution of sugar chains in their blood and bodily secretion (milk, mucus, tears, semen).

Glycocalyx: "Sugar coat" complex layer of glycoconjugates, glycolipids and glycoproteins surrounding each living cell and most viruses.

Extra cellular matrix: secreted proteins, proteoglycans and glycans forming much of the connective tissues.

Virulence: Degree to which a pathogen causes disease/ harms its host.

Horizontal vs vertical transmission: Transmission between individuals other than from parent to child.

Superinfection: Multiple infection of the same host.

Emergence: sudden appearance of a pathogen in a new host.

Toxoplasma gondii. Protozoan cat parasite that can infect humans and can cause fetal damage and psychological changes.

Mode of transmission: Manner by which a parasite gets from one host to another.

Guinea worm (*Dracunculus medinensis*): Parasitic worm with complex life cycle and infecting humans via water contact.

Shistosome: Parasitic worm with complex life cycle and infecting snails and humans via water contact.

Home base: Stable reuse of the same location as sleeping and feeding quarters by a group of animals.

Neolithic: "New stone" the time period of the last ten thousand years, where humans settled and started farming and/ raising livestock.

COPD: Chronic obstructive pulmonary disease (caused by smoking and /or exposure to indoor or industrial smoke)

Animal domestication: Complete control of an animal's reproduction and selection for docility.

Pellagra: Disease as result of vitamin B (niacin) deficiency. Common in populations that rely on corn as a staple food but do not nixtamalize the corn.

Puerperal fever: Fever associated with bacterial infections after giving birth, a disease directly caused by doctors who were not washing their hands before delivering children.

Toxic shock syndrome: Very serious sequelae from bacterial infection after not changing a tampon.

Antibiotic resistance: resistance of bacteria to the killing by antibiotics.

DES: Synthetic steroid drug used to treat women in risk of miscarriage or chemical that caused major developmental defects in the daughters of treated mothers.

Thalidomide: Drug used to treat morning sickness that caused major deformities in infants.

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Autism: A wide spectrum of mental conditions, present from early childhood, characterized by difficulty in communicating and forming relationships with other people and in using language and abstract concepts.

Hygiene hypothesis: Idea that many modern medical problems derive from an excess of hygiene and lack of appropriate exposure to microbes or parasites.

Trypanosome: Protozoan parasite transmitted by insect that cause severe diseases such as sleeping sickness and Chagas disease.

Leishmania: protozoan parasite that cause severe tropical diseases.

Megafauna: Animals larger than 90 lbs.

Emerging disease: Disease that suddenly appears with humans as novel host.

Legionella: bacterial pathogen that can thrive in air conditioning systems and causes severe disease.

Pandemic: World-wide wave of infection (epidemic)

Influenza A virus: an RNA virus of water birds

Zika virus: Emerging virus from primates in Africa that can cause brain defects (microcephaly) in humans.

Hemagglutinin: Sugar (sialic acid) binding protein on the surface of Influenza viruses.

Neuraminidase: Sugar (sialic acid) cleaving protein on the surface of influenza A viruses

SIV: Simian Immunodeficiency Virus (a misnomer, as it does NOT cause immunodeficiency in most non-human primates)

HIV: Human immunodeficiency Virus, HIV1 infected humans from chimpanzees in central Africa, HIV2 infected humans from sooty mangabey monkeys in West Africa.

Patient zero: First patient of an emergent epidemic

Nipah virus: Asian virus carried by bats infecting other mammals including humans.

Ebola virus: African virus of bats infecting humans and other primates

Vaccination: Triggering of protective immunity in a host by exposure to parts of a pathogen or the whole inactivated or attenuated pathogen.

Small pox virus: Highly contagious virus eradicated by vaccination.

Monkey pox virus: A zoonotic virus from an African tree rodent (squirrel) that easily infects humans.

West Nile virus: Emerging virus carried by ravens and other birds of the corvid family, can be deadly when transmitted to humans by mosquitos.

Cancer: Collection of diseases that share the breakdown of cellular collaboration.

Hyperplasy: Overgrowth of a tissue or cell type.

Neoplasm: Novel growth of a tissue.

Tumor: Mass of cells or tissue

Oncogene: Gene associated with the causation of cancer.

Oncofetal antigen: Molecule often observed only of fetal (including placenta) and cancer tissue.

Tumor suppressor gene: Gene that controls cell division and growth or otherwise prevents cells from "rogue behavior".

Antagonistic pleiotropy: Genes with multiple effects, where early effects have opposite effects than later effects.

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Three germ layers: Layers of tissue that give rise to different organs and tissue types.

Carcinoma: Cancers of the epithelia

Sarcoma: Cancers of connective tissue.

Adenoma: Cancers of glands

Leukemia: Cancers of white blood cells

Glioma: Cancers of glial cells.

GOD: Generation Of Diversity: a key advantage of organisms, immune responses and cancers.

Metastasis: Dispersal of a cancer to a site other than its origin.

Tumor specific antigens: Molecules characteristic for certain cancers.

P53 (tumor protein 53): gate keeping protein that controls cell division and growth.

Peto's paradox: Observation that animals with 100 or 1000 more cells than humans do not have higher cancer rates.

Somatic selection: Natural selection of cancer cells over other somatic cells.

Cancer immune editing: Somatic evolution of cancer cells to evade and/or manipulate the immunity of the organism.

Striding bipedality: bipedal locomotion with regular long strides, including fast walking and running, requires rotation of torso.

Varicose veins: damage to veins in lower part of the body due to gravity increasing blood pressure.

Obstetric dilemma: The need for narrow hips for bipedality and the constraint this exercises on the birth canal for a species with large headed babies.

Williams syndrome: a genetic condition due to the loss of ~1.5 million base pairs of DNA from chromosome 7. Causes distinct appearance of children and leads to hyper verbal behavior, musical talent, and excessive levels of trust, but is also accompanied by several medical conditions (vasculature).

Dyslexia: Cognitive condition causing challenges in reading and orthography but associated with advantages in pattern recognition.

Anxiety disorder: a group of mental disorders characterized by feelings of anxiety and fear.

Cephalo-pelvic disproportion: the mismatch between baby head size and a mother's pelvic canal.

Population age structure: The distribution of ages in a population, often depicted as "pyramid" with younger ages at the bottom and older ages at top, males on the right and females on the left.

Hunter gatherer: human populations that forage for subsistence, collecting wild plants and hunting animals for food.

Nomophobia: suggested name for the pathological fear of losing mobile phone access.

Environment of evolutionary adaptation (EEA): the environment in which modern *Homo sapiens* has lived for the largest proportion of time (>90%), i.e. pre-agricultural hunter-gathering, at low population size and in small social groups.

Mismatch: A lack of fit between ancient adaptations, such as the tendency to love salty, sweet and fatty foods under environments where such foods are rare and modern life, where such food items are in our reach 24/7.

Non-communicable disease: disease that are not infectious

Epidemiological Transition: A drastic change in disease landscape such as the one between infectious disease and degenerative, non-communicable disease.

Double burden of disease: the condition currently encountered by populations in developing nations where the threat of many infectious diseases remains but the scourge of degenerative disease also exists.

Yaws: An infectious disease caused by the bacterial parasite *Treponema pertenuae*, a close relative of syphilis caused by *T. pallidum*.

The Black Death: recurrent epidemics of the plague caused by the flea transmitted bacterium *Yersinia pestis* that hit Europe and Asia over the last 2000 years. The 14th century epidemics killed about half of the European population.

The White Death: a name sometimes used for tuberculosis an infectious disease caused by the bacterium *Mycobacterium tuberculosis*.

Leprosy: An infectious disease caused by the bacterium *Mycobacterium leprae*.

Metagenomics: the study of the complete mixture of genomes in a given samples, e.g. metagenomics of human fecal samples from airline toilets will reveal all life form in these samples: viruses, phages, bacteria, archaea, protozoa, food plants and animals and human DNA.

Ovum: A female gamete, also known as an egg or oocyte. Ova (*pl*) are produced by the ovaries of the woman.

Spermatozoon: A male gamete. Sperm (or spermatozoa, *pl*) are produced in the testes of the man.

Ovulation: The release of an ovum from the ovary. In humans, ovulation usually involves the release of a single egg in each menstrual cycle.

Fallopian tube: A narrow tubular extension of the uterus, which opens out next to the ovary. It is also called the oviduct. Following ovulation, the ovum passes into the opening of the fallopian tube and travels towards the uterus.

Menstrual cycle: An interval of approximately 28 days, which commences with the onset of menstruation. Ovulation occurs mid-way though a menstrual cycle, approximately 14 days before the onset of the next cycle.

Amenorrhoea: The absence of menstruation. A missed menstrual period is often the first observable sign that pregnancy has commenced, although there are many other causes.

Fertile period: The time in a woman's menstrual cycle during which coitus may result in pregnancy. This period probably varies considerably between women. Coitus up to 6 days prior to and 1 day after ovulation may result in pregnancy although the most fertile days are the day of ovulation and the 2 days beforehand.

Fertilization: The fusion of a spermatozoon and an ovum, which usually takes place in the fallopian tube up to 24 hours after ovulation.

Conception: A biologically imprecise term meaning either 'the coming into existence of a new human being' or 'the beginning of a pregnancy'. It is often used synonymously with fertilization but may also refer to implantation.

Embryo: A newly fertilized ovum until the eighth week of development.

Zygote: The newly fertilized ovum: a one-cell embryo.

Blastocyst: An embryo approximately 5–6 days after fertilization.

Implantation: The biological process that begins when a blastocyst attaches to the lining of the uterus approximately 6–7 days after fertilization. The embryo subsequently becomes embedded within the uterine lining.

Human chorionic gonadotrophin (hCG): A protein produced by the embryo. It signals to the mother that an embryo is present and prevents menstruation and the loss of the embryo. Elevated levels of hCG can be detected in the serum or urine of a woman from around the time of implantation.

Fecundability: A measure of reproductive potential. It is the probability of becoming pregnant in a single menstrual cycle. **Fecundity** is often used to mean the probability of achieving a live birth in a single cycle. A **fecund** cycle is one in which fertilisation occurs.

Pregnancy: The condition of a woman harbouring an embryo, fetus or unborn child. When pregnancy begins is a matter of some confusion. Pregnancy may be considered to commence with fertilisation and lasts approximately 38 weeks. Clinicians often time the onset of pregnancy from day 1 of the last menstrual cycle, 2 weeks before fertilisation, and refer to subsequent time as a period of **gestation**. On this account, pregnancy or gestation lasts approximately 40 weeks. Some scientists and legal judgements define pregnancy as beginning with implantation, one week after fertilisation. This definition is of particular utility in the context of IVF treatment where evidence of implantation is the earliest sign that a transferred embryo has developed normally and that fertility treatment has, up to that point, been successful. For some women, the start of a pregnancy may be noted with the first missed menstrual period, approximately 2 weeks after fertilisation, or a positive pregnancy test.

Miscarriage: The premature termination of a pregnancy leading to loss of a developing embryo or fetus. Embryo loss may occur before a woman knows she is pregnant. Miscarriage late in pregnancy is often called abortion, with a cut-off of approximately 20 weeks gestation used to distinguish between miscarriage and abortion.

Early Pregnancy Loss: This usually refers to the loss of an embryo very early in pregnancy, even before a clinical diagnosis is made, when a woman would not be aware of the pregnancy. Such losses are also called **occult**, because they are hidden, or **biochemical**, because they can only be identified by detecting hCG. Pregnancy loss shortly after a clinical diagnosis may also be described as **early**.