Cellular and Molecular Explorations of Anthropogeny

Glossary

Genetic nomenclature in humans and other species
Human genes are written as all capitals and italicized (ex. *FOXP2*), whereas mouse genes (used as non-human models) are written as first letter only capitalized and all italicized (ex. *Foxp2*). Proteins, the products of genes, follow the same capitalization as the gene encoding them but are not italicized (ex. FOXP2 or Foxp2).

Admixture: Breeding between isolated populations.

Allele: Alternative DNA sequence at the same genetic locus (location on the chromosome).

Antagonistic Pleiotropy: A phenomenon whereby multiple influences of the same gene have opposite effects on the fitness of the organism

***ARHGAP11B*:** A human-specific protein coding gene that promotes amplification of basal progenitors in the subventricular zone, producing more neurons during fetal cortical development. It has been implicated in the evolutionary expansion of the human brain neocortex.

Basal Progenitor (PB): A cortical neural progenitor cell which undergoes replication and division. Basal progenitor cells are a subset that lie in the subventricular zone and lack contact with the neighboring ventricle—only contacting the outer, basal, surface—and contribute to the expansion of the outer cortex.

Basal Radial Glia (bRG): A primary progenitor cell capable of generating neurons, astrocytes, and oligodendrocytes. Basal radial glia and outer radial glia are defined by their position, morphology, and genetic phenotype.

Chromosomes: Discrete strands of packaged DNA.

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

Copy Number Variation: A mutation involving the repetition of sections of the genome. The number of repeats varies between individuals.

Cortical-Basal Ganglia: Subcortical nuclei in the base of the forebrains of vertebrates, including humans, which are involved with a variety of functions including control of voluntary motor movements, procedural learning, routine behaviors or "habits" such as teeth grinding, eye movements, cognition, and emotion.

Cranial Neural Crest Cells: Cells that become the structures of the endocranium and face.

Daughter Neuron: Resulting cell(s) formed when neural stem cells or progenitor cells undergo cellular division.

Denisovans: An extinct hominin population contemporary with Neanderthals **that hybridized with ancient humans**. Knowledge of Denisovan morphology is limited to two small fossils found in Siberia.

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

***FOXP2*:** A gene in humans that encodes for a transcription factor protein and is involved in the production of speech.

Gene: DNA whose information encodes a function.

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.

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

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

Germinal Zone: A region where cell division and proliferation occurs during vertebrate central nervous system development consisting of 2 layers lining the ventricles (ventricular zone and subventricular zone).

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

Hemideletion: One of two paired chromosomes is affected by a deletion. The other chromosome is intact.

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

Hominin: A classification of species comprising humans and extinct relatives (ex. *Australopithecus*, *Paranthropus* and *Ardipithecus*. Not all are ancestral to humans) from after the split with the common ancestor with chimpanzees.

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

Hybridization: Breeding among recognized species.

Indel: Insertions or deletions of DNA sequence.

Induced Pluripotent Stem Cells (iPSC): Somatic (body) cells artificially that are reprogrammed to an embryonic-like stem cell state and differentiated into other types of cells.

Introgression: Transfer of alleles between species.

Locus (pl. Loci): A unique physical position on a chromosome.

Mechanistic Target of Rapamycin (mTOR): A protein kinase, which in humans is encoded by the *MTOR* gene.

Medium Spiny Neurons: A special type of GABAergic inhibitory cell representing 95% of neurons within the human striatum, a basal ganglia structure.

Mutation: Change of a DNA sequence ranging from single base pair change to changes involving multi-million base pairs due to deletions and duplications.

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

Neocortex: A part of the cerebral cortex concerned with sight, hearing, and touch in mammals, regarded as the most recently evolved part of the cortex.

Nerve: A bundle of fibers that transmits impulses of sensation to the brain or spinal cord, and impulses from these to the muscles and organs.

Neural Stem Cell: A self-renewing, multipotent cell that generates the neurons and glia of the nervous system of all animals during embryonic development. Some persist in the adult vertebrate brain and continue to produce neurons throughout life.

Neural Progenitor Cell (NPC): Cells that are capable of dividing a limited number of times and have the capacity to differentiate into a restricted repertoire of neuronal and glial cell types.

Nervous System: The network of nerve cells and fibers that transmits nerve impulses between parts of the body.

Neuron: A specialized cell that transmits nerve impulses.

Neural circuit: A neural circuit is a functional entity of interconnected neurons that is able to regulate its own activity using a feedback loop.

Neurobiology: The study of the morphology, behavior, and other qualities of the nervous system.

Neurogenetics: The study of the role of genetics in the development and function of the nervous system.

Nuclear Pore Complexes (NPCs): Protein and ribonucleoprotein transport channels in the nuclear envelop of eukaryotic cells. Evolved ~ 1.5 billion years ago. While the primary role of NPCs is to regulate nucleo–cytoplasmic transport, recent research suggests that certain NPC proteins have additionally acquired the role of affecting gene expression at the nuclear periphery and in the nucleoplasm in metazoans.

Nucleoporin 98 (Nup98): A protein coding gene that plays a role in the nuclear pore complex assembly and/or maintenance. Associated diseases range from Myelodysplastic Syndrome and Acute Monocytic Leukemia.

Organoid: Cell/tissue culture *in vitro* that aims to mimic organ structure and function.

Outer Radial Glia (oRG): Found in the outer subventricular zone of the neocortex, outer radial glia preferentially express genes related to extracellular matrix formation, migration, and stemness.

Outer Subventricular Zone (oSVZ): A uniquely structured germinal zone that generates the expanded primate supragranular layers.

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

Phylogeny: Historical relationships of species or genetic loci.

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

***POM121*:** A gene that encodes for transmembrane nucleoporin, a protein that localizes to the inner nuclear membrane and forms a core component of the nuclear pore complex, which mediates transport to and from the nucleus.

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

Pyramidal Neurons: A type of multipolar neuron found in areas of the brain including the cerebral cortex, the hippocampus, and the amygdala. Pyramidal neurons are the primary excitation units of the mammalian prefrontal cortex and the corticospinal tract.

Reading-Frame: A way of dividing the sequence of nucleotides in a nucleic acid (DNA or RNA) molecule into a set of consecutive, non-overlapping triplets called *codons*.

Reading-Frame Shift: A genetic mutation caused by indels (insertions or deletions) of a number of nucleotides in a DNA sequence that is not divisible by three.

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

Sequencing: Reading the order of nucleotides in DNA.

Single Cell Clonal Lineage Analysis: A system for labeling and following a single progenitor cell and its daughter cells as they proliferate and mature.

Species: A population that can produce viable and fertile offspring. (debated definition)

SRGAP2: A gene on chromosome 1 that encodes for a protein that plays a role in cortical neuron development. Duplications of this gene are unique only to humans.

Subventricular Zone (SVZ): Describes both embryonic and adult neural tissues in the vertebrate nervous system.

Transcription Factor Protein: A protein that alters gene expression by binding directly or indirectly to DNA.

Variant: DNA that differs among groups studied.

Ventricular Zone (VZ): A transient embryonic layer of tissue containing neural stem cells, principally radial glial cells, of the central nervous system of vertebrates.

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