

References

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Glossary

amino acid: One of several molecules that join together to form proteins. There are 20 common amino acids found in proteins.

attention deficit hyperactivity disorder (ADHD): A disorder that may be characterized by a pattern of inattention (inability to concentrate) sometimes combined with hyperactivity and impulsivity that is persistent and developmentally inappropriate, and occurs in at least two different settings.

blood-brain barrier: A network of blood vessels and tissue that is made up of closely spaced cells and helps keep harmful substances from reaching the brain. The blood-brain barrier lets some substances, such as water, oxygen, carbon dioxide, and general anesthetics, pass into the brain. It also keeps out bacteria and other substances, such as many anticancer drugs.

brain protein synthesis: Brain protein synthesis is the group of processes that are involved in the generation of mature brain protein molecules required for normal brain functioning.

cofactor: Something that must join with another substance to produce a given result. There are many different cofactors in the body. One example is BH₄, which is a cofactor required for PAH to convert Phe to Tyr in the liver.

conceptual reasoning: The ability to problem solve by a creative search for new ideas or solutions.

dopamine: One of the neurotransmitters in the brain. It is derived from tyrosine and is converted to norepinephrine and epinephrine. Dopamine is important in regulating movement. It communicates with the nerves via a family of receptors.

enzyme: A protein that speeds up chemical reactions in the body.

enzyme substitution therapy: A method of injecting enzymes to substitute for those that are missing or altered because of genetic mutations.

essential amino acids: Amino acids that cannot be synthesized in the body and can only be obtained through food supply.

executive function (EF): Deliberate, conscious control over your own thoughts, actions and emotions.

gene therapy: Treatment that alters a gene. For example, in studies of gene therapy for cancer, researchers are trying to improve the body's natural ability to fight the disease or to make the cancer cells more sensitive to other kinds of therapy.

hepatocyte: A liver cell.

impulse/inhibitory control: The ability to suppress actions and distractions that would otherwise interfere with the attainment of a goal.

information processing speed: How quickly you can react to incoming information, process it, understand it and use it.

magnetic resonance imaging (MRI): A procedure in which radio waves and a powerful magnet linked to a computer are used to create detailed pictures of areas inside the body. These pictures can show the difference between normal and diseased tissue. MRI makes better images of organs and soft tissue than other scanning techniques, such as computed tomography (CT) or X-ray. MRI is especially useful for imaging the brain, the spine, the soft tissue of joints and the inside of bones.

maternal PKU syndrome: A condition caused by elevated Phe harming the developing fetus. Children born to mothers who have untreated PKU can have mental retardation, heart defects, small heads, low birth weight and cognitive and behavioral problems.

mental/cognitive flexibility: The ability to switch between tasks in response to changing task demands.

myelin sheath: An insulating layer found on neuronal axons (long extensions of nerve cells). This structure allows nerve impulses to travel faster by keeping the electrical current inside the nerve.

neurotransmitter: A chemical that is made by nerve cells and used to communicate with other cells, including other nerve cells and muscle cells.

phenylalanine (Phe): An essential amino acid in humans (provided by food). Phenylalanine plays a key role in the production of other amino acids and is important in the structure and function of many proteins and enzymes. Phenylalanine is converted to tyrosine, which is used in the production of dopamine and norepinephrine neurotransmitters.

phenylalanine hydroxylase (PAH): An enzyme that helps change phenylalanine to tyrosine. In PKU, PAH is mutated and possesses less activity than a normal PAH. There are over 500 different known mutations of PAH that can cause PKU, some more severe than others.

phenylketonuria (PKU): An inherited disorder that causes a build-up of phenylalanine (an amino acid) in the blood. This can cause mental retardation, behavioral and movement problems, seizures and delayed development. Using a blood test, PKU can easily be found in newborns. The treatment is a diet low in phenylalanine.

protein: A molecule made up of amino acids that are needed for the body to function properly. Proteins are the basis of body structures such as skin and hair and of substances such as enzymes, cytokines and antibodies.

psychiatric disorder: A disorder characterized by behavioral and/or psychological abnormalities, often accompanied by physical symptoms. The psychiatric symptoms may significantly affect many aspects of a person's day-to-day life, causing significant distress or impairment in social and work settings.

selective and sustained attention: The ability to keep focus over time on attaining a goal.

tryptophan: The least plentiful of all amino acids and an essential amino acid in humans (provided by food). Tryptophan is found in most proteins. Tryptophan is converted to 5-hydroxy-tryptophan (5-HTP), which is converted in turn to serotonin, a neurotransmitter essential in regulating appetite, sleep, mood, and pain. Tryptophan is a natural sedative. It is present in dairy products, meats, brown rice, fish, and soybeans.

tyrosine (Tyr): Considered a non-essential amino acid; however, in patients with phenylketonuria who lack phenylalanine hydroxylase and cannot convert phenylalanine into tyrosine, it is considered an essential nutrient. Tyrosine plays a role in protein synthesis and is necessary for the production of some neurotransmitters and hormones.

white matter: The nerve tissue forming the bulk of the deep parts of the brain and the outer parts of the spinal cord. It is white because of a white-colored insulating layer on the nerve fibers, the myelin sheath. It is composed of nerve cell extensions (axons), which connect various grey matter areas of the brain to each other and carry nerve impulses to and from the nerve cell bodies within the central nervous system (neurons).

working memory: The ability to hold information in your mind and work with it over a short period of time.