Propionic Acidemia Foundation
Glossary of Commonly Used Terms

Amino Acids: When proteins are digested in the diet, amino acids remain - amino acids are either essential (obtained through diet) or non-essential (made by the body from the essential amino acids). *

Ammonia: A by-product of protein metabolism. *

Anion Gap: The difference between the sum of cations and anions found in plasma or serum. The anion gap is used to aid in the differential diagnosis of metabolic acidosis. It is calculated by subtracting the chloride and bicarbonate levels from the sodium plus potassium levels.

Asymptomatic: Showing no symptoms. *

Autosomal Recessive Inherited Disorder: A characteristic or disorder occurring when an individual receives two copies of a mutated gene for that condition, one from the mother and one from the father

Biochemical Pathway: Systems in the body for processing molecules for useful purposes. *

Biotin: A vitamin Cofactor for carboxylase enzymes. Essential for metabolism of proteins, carbohydrates and fats.

Branched Chain Amino Acid (BCAA): L-Leucine, L-Isoleucine, L-Valine are essential amino acids because humans cannot survive unless they are present in the diet. They are easily converted to ATP, critical to energy and muscle metabolism. They aid in hemoglobin formation, which helps to stabilize blood sugar and lower elevated blood sugar levels. L-Leucine decreases blood sugar and boosts tissue healing, including bone. L-isoleucine is essential for hemoglobin formation and regulates blood sugar and energy levels. L-Valine acts as a natural stimulant and is involved in tissue regeneration and nitrogen balance. *

Cardiomyopathy: The Cardiomyopathy is a group of chronic disorders affecting the muscle of the heart resulting in impairment of the pumping function of the heart.

Carnitine: This essential fatty acid metabolism cofactor helps to move the fatty acid to the mitochondria from the cytoplasm of the cell.

Carrier: Individuals carrying an abnormal gene that can be transmitted to their offspring. These individuals do not show evidence of the disorder. *

Catabolism: The breakdown of lean muscle mass to obtain amino acids (for growth and development) and energy, resulting from inadequate supply in the diet. Results in excess production of ammonia. *
Any metabolic process by which organisms convert substances into excreted compounds

Chronic: A situation or disease with a long duration. *

Cofactor: A Cofactor is any substance that needs to be present in addition to an enzyme to catalyze a certain reaction.

Constipation: Difficult, incomplete, or infrequent evacuation of dry hardened feces from the bowels. Can cause PA's serious illness. **

Cyclic: Recurring or moving in cycles. **

Deficiency: A lower amount than necessary for functioning. *

Dehydration: Excessive loss of water from the body or from an organ or body part, as from illness or fluid deprivation. **

Developmental Disabilities: A chronic mental or physical impairment that results in decreased ability of an individual to reach appropriate age-level developmental goals. *

DNA: Deoxyribonucleic acid (DNA) is the chemical inside the nucleus of all cells that carries the genetic instructions for making living organisms. *

Electrolytes: Any of various ions, such as sodium or chloride, required by cells to regulate the electric charge and flow of water molecules across the cell membrane. The primary ions of electrolytes are sodium, potassium, calcium, magnesium, chloride, phosphate and bicarbonate. **

Enzyme: A protein molecule that helps other organic molecules enter into chemical reactions with one another but is itself unaffected by these reactions. **

Enzymatic Assay: laboratory methods for measuring enzymatic activity. **

Etiology: The origins of a disease. *

Fibroblasts: A cell that is present in connective tissue and active in making and secreting collagen. * Skin cells.

Gene: A gene is, in essence, a segment of DNA that has a particular purpose, i.e., that codes for (contains the chemical information necessary for the creation of) a specific enzyme or other protein. **

Hyperammonemia: Abnormally high levels of ammonia in the blood; if untreated, causing severe agitation, vomiting, lethargy, coma and death. *

Hypothermia: Abnormally low body temperature below 95 degrees F, causing heart and respiration slowing and paleness. *

Hypotonia (low tone): A condition in which there is diminution or loss of muscular tonicity, resulting in stretching of the muscles beyond their normal limits. **

Isoleucine: An essential amino acid found in proteins. One of the restricted amino acids for PA patients.

Ketone or Ketone Bodies: A ketone is an intermediate product of the breakdown of fats in the body; any of three compounds (acetoacetic acid, acetone, and/or beta-hydroxybutyric acid) found in excess in blood and urine of persons with metabolic disorders ** Ketones are used as a measure of metabolic instability in PA patients.

Ketosis: A pathological increase in the production of ketone bodies. Ketosis is a stage in metabolism occurring when the liver has been depleted of stored glycogen and switches to a fasting mode such as occurs during sleep, during dieting, and during the body's response to starvation.** In PA, a measure of metabolic instability.

Late-onset disorder: Characterized by mild, moderate or severe symptoms (occurring anytime after the neonatal period) in early or late childhood resulting from mutations allowing varying degrees of partial enzyme activity. Also sometimes referred to as "partial" defects. * A late-onset metabolic crisis can be as severe and life-threatening as the neonatal form.

Lethargy: Sleepiness. *

Liver: A large vascular organ in the body that causes important changes in substances in the body in order for the body to use these substances. *

Metabolic Acidosis: Decreased pH and bicarbonate concentration of the body fluids caused either by the accumulation of excess acids stronger than carbonic acid or by abnormal losses of bicarbonate from the body. ** A metabolic derangement of acid-base balance where the blood pH is abnormally low.

Metabolic Pathway: A cascade of chemical reactions by which the chemical changes in living cells provide energy for vital processes in the body. Energy production in the cell occurs in the mitochondria.

Metabolite: A substance produced by metabolic action or necessary for metabolic process. In PA, certain metabolites can reach toxic levels. * Any substance produced by metabolism or by a metabolic process

Methionine: Amino acid found in most proteins and essential for nutrition. Restricted amino acid for patients with Propionic Acidemia. **

Mutation: A change in genetic material occurring spontaneously or by induction, which changes the original expression (function or purpose) of the gene. *

Neonatal Onset Disorder: Severe, catastrophic disorder with life-threatening symptoms occurring in the neonatal period resulting from null/zero enzyme mutations or severely impaired enzyme activity.*

Neutropenia: An abnormal decrease in the number of neutrophils in the blood. **

Odd Chain Fatty Acid: fatty acids with an odd number of carbon atoms.

Organic Acidemia: Inherited disorders of amino acid catabolism in which toxic substances are produced as a result of an enzymatic blockage

Partial Activity: Not completely active, may be missing vital components. *

Plasma: Liquid part of the blood in which blood cells are suspended. *

Proband: An individual with a particular disorder who causes a study of his hereditary and genetic factors to determine if other members of the family have the same disease or carry it.* The proband might for example be a baby with propionic acidemia.

Prenatal: Before birth. *

Protein: Essential to all living cells, simplified by body processes to simple alpha-amino acids. * Twenty different amino acids are commonly found in proteins and each protein has a unique, genetically defined amino acid sequence which determines its specific shape and function.

Quantitative Amino Acids: Blood test done to measure levels of all amino acids individually.

Rapid Onset: Beginning quickly without warning.*

Seizures: A temporary change in brain performance due to abnormal electrical activity of a specific group of cells in the brain that either present with sudden muscle contractions, decreased level of consciousness, and several other symptoms. *

Serum or Plasma Ammonia level: Amount of ammonia concentration present in blood or plasma, used to monitor ammonia levels in PA's. *

Supplementation: A substance added to the diet to counteract a deficiency or potential deficiency. *

Threonine: Amino acid found in most proteins and essential for nutrition. Restricted amino acid for patients with Propionic Acidemia. **

Transport: To carry from one area to another in the body or within a cell. *

Tremor Ataxia: Trembling or shaking and lack of control of voluntary muscles. *

Triggering Event: An episode that causes a reaction or illness. *

Urea: A product of protein breakdown of amino acids, excreted in the urine. *

Valine: Amino acid found in most proteins and essential for nutrition. Restricted amino acid for patients with Propionic Acidemia. **

Waste: Unusable or excess material, lost by breaking down of the body's tissues. * Toxic by-products of cellular processes that are excreted from the body.

References:

* National Urea Cycle Foundation

** The American Heritage® Dictionary of the English Language, Fourth Edition. Houghton Mifflin Company, 2004.

** The American Heritage Stedman's Medical Dictionary. Houghton Mifflin Company, 2002.

** WordNet 1.7.1. Princeton University, 2001