HSP/PLS – Words to Know

A

Acupuncture is an alternative therapy treatment. It is an ancient system of Chinese medicine where thin needles are inserted into specific points on the body. These points are thought be connected to specific organ systems. The purpose is to alleviate various health conditions, including headaches, nausea and addiction.

(AFO) Ankle Foot Orthotics are special shoe inserts, splints, or braces that are used to relieve various gait problems, foot problems, help increase balance, or take pressure off sore spots on the feet. Orthotics may be custom molded for your feet, or may be pre-formed supports, such as simple arch supports.

Allele. Any one of a series of two or more different genes that occupy the same position (locus) on a chromosome. In the case of dominant and recessive alleles, the dominant allele will prevail over a normal allele.

Amyotrophic Lateral Sclerosis (ALS) is a rapidly progressive, fatal, neuromuscular disease. Fifty percent (50%) of ALS patients die from respiratory failure or pneumonia within three to five years of diagnosis. The condition is marked by degeneration of the upper motor and lower motor neurons. It is characterized by progressive muscle weakness, atrophy, stiffness, spasticity and fasciculations. Symptoms commonly appear in middle to late adulthood. There are some slow progressive forms of ALS. Also called Lou Gehrig's Disease or Motor Neuron Disease.

Amino Acid. One of the twenty building blocks of protein. They are the key components in all living things.

Anticholinergics are agents that inhibit the involuntary contractions of the bladder. They can also help increase the capacity of the bladder. Anticholinergics are often prescribed for treating urge incontinence.

Antioxidant. A chemical compound or substance that inhibits oxidation.

Antispasmodics drugs help reduce spasms, stiffness and cramps. For PLS and HSP, various types of antispasmodics may be prescribed to help reduce muscle spasticity and spasms.

(Apparently Sporadic) Spastic Paraplegia (SP) is often a term used when all the signs and symptoms indicate Hereditary Spastic Paraparesis but there is no documented family history to prove that the disorder is genetic. Clinicians may diagnose this situation as HSP, SP or Primary Lateral Sclerosis.

ATAXIA. A neurological condition characterized by a lack of coordination and control over muscle movements. Poor muscle control that causes clumsy movements.

Atrophy (Muscle Atrophy). Loss of muscle fiber volume characterized by a visible decrease in muscle size. This occurs because muscles no longer receive impulses or signals from nerve cells. It is not a symptom in HSP and PLS, although muscles may exhibit atrophy due to misuse.

Autosome/Autosomal is a chromosome that is not the sex determining chromosome. Humans have 23 pairs of autosomes and one pair of sex chromosomes. The term autosomal means that the gene responsible for the disorder is located on one of the autosomes.

Axon. The long, hair like extension of a nerve cell that carries a message to the next nerve cell. Each nerve cell (neuron) has one axon, which can be over a foot long, and sometimes many feet long. Nerve cells communicate with each other by transmitting electrical signals from the branches at the ends of their axons. They receive signals at extensions called dendrites.

B

Babinkski's signs. A pathological reflex where the great toe extends and flexes toward the top of the foot and the other toes fan out when the sole of the foot is firmly stroked. Babinkski's signs are commonly seen in PLS and HSP.

Baclofen Pump Is surgically implanted under the skin of the abdomen and delivers a small, continuous dose of medication directly to the spinal canal through a catheter. The "pump" can be programmed to release a specific amount of medicine at select times, which can be adjusted without surgery. The pump needs to be refilled every one (1) to three (3) months.

Bulbar Muscles. The muscles that control the speech, chewing and swallowing. Bulbar muscles are affected in PLS but rarely in HSP.

C

Carrier. A person who has an affected recessive gene but usually does not show the effects of that gene because he or she also has an unaffected gene. Normally, being a carrier of a recessive disorder is not harmful to the carrier. Because carriers have one affected gene, they can pass that gene to their children – however, unless the other parent is also a carrier, it is unlikely that any children will be affected by the disorder.

Celebrex is an FDA-approved drug for the treatment of rheumatoid arthritis, osteoarthritis and pain.

Central Nervous System (CNS). The brain and spinal cord combined.

Cerebellum. The portion of the brain in the back of the head between the cerebrum and the brain stem. It is responsible for motor control.

Cerebra Spinal Fluid Analysis (Spinal Tap). A procedure used to isolate cerebrospinal fluid for evaluation or diagnosis of disease. The analysis is normal in HSP and PLS patients.

Chiropractic. A treatment method that depends primarily on manipulating or adjusting the spine to prevent disease and treat pain and other ailments.

Chromosomes are self-replicating, genetic structures of cells. They are located within every cell of the human body and contain pieces of information (genes) called DNA. The DNA in each chromosome contains many genes.

Chronic. Marked by long duration or frequent recurrence.

Clonus is a repetitive jerking of muscles (a series of muscle contractions) that occurs when there is a disruption of the signals from the brain that normally inhibit muscle contractions. This disruption may be due to the effects of HSP/PLS on stretch reflexes. A video example can be viewed at: https://www.youtube.com/watch?v=4SrhgjGIZ30

Co-Enzyme Q10 is a non-prescription dietary supplement that is involved in a variety of cellular processes.

Complicated HSP. Some forms of HSP are considered "complicated" because they involve additional neurologic symptoms in addition to progressive spasticity and weakness in the legs. These conditions include peripheral neuropathy, ichthyosis (a skin disorder), epilepsy, ataxia, optic neuropathy, retinopathy, dementia, mental retardation, deafness, or problems with speech, swallowing or breathing.

Cortico-Spinal Tracts also called the upper motor neurons, are the long nerves that run from the brain to the spinal column. They synapse onto spinal neurons (lower motor neurons) that begin in the spinal cord and travel to the arms and legs.

Cortex. The outer layer of the cerebrum densely packed with nerve cells.

Creatine is a dietary supplement that is promoted for its ability to enhance muscle strength and physical endurance.

D

Dementia is characterized by a decline in intellectual functioning that is severe enough to interfere with the ability to perform routine activities.

Demyelinating. Loss of the myelin sheath that surrounds nerves. Loss of myelin is not a factor in most forms of HSP & PLS.

Dendrite. Extensions from the neuron cell body that take information to the cell body. A single nerve may possess many dendrites.

Dominant. If a dominant gene produces a particular trait or disorder, a person only needs to have one gene with that trait for it to appear, even if the other corresponding gene is normal.

Dysarthria. A speech disorder that is due to a weakness or incoordination of the speech muscles. Speech is slow, weak, imprecise or uncoordinated.

Dysphagia. The medical term for any difficulty or discomfort when swallowing. A normal swallow takes place in four stages and involves 25 different muscles and five different nerves.

E

Emotional Lability is uncontrolled laughter and crying that does not reflect how a person actually feels. This condition can affect people who have diseases or injuries of the brain and nervous system.

F

Fasciculation. Small, involuntary, irregular, visible contractions of individual muscle fibers caused by damage to the lower motor neurons. This symptom is usually not associated with PLS or HSP. A video example can be viewed at: https://www.youtube.com/watch?v=2pohtj8TeDY

FDA (U.S. Food and Drug Administration). The branch of federal government responsible for protecting the public health by assuring the safety, efficacy, and security of human and veterinary drugs, biological products, medical devices, our nation's food supply, cosmetics, and products that emit radiation.

Familial. Occurring within a family. Although this term may be used when referring to hereditary disorders, the cause may not necessarily be hereditary.

Familial Spastic Paraplegia. See Hereditary Spastic Paraplegia.

Free Radicals. Chemicals that are highly reactive and can oxidize other molecules (i.e. Superoxide).

G

Genes. Pieces of genetic information stored in codes in DNA that carry the instructions for making all of the proteins a cell needs and which determine traits such as hair and eye color. Genes contain the hereditary information that is passed on from parents to children.

Genome. All the genetic material in a particular organism that is contained in the chromosomes. The "human genome" is the collection of genes that is necessary to make a human being.

Glutamate is a neurotransmitter that sends signals in the brain and throughout the nerves in the body. Glutamate plays an important role during brain development. Normal levels of glutamate also help with learning and memory.

H

Hereditary. A trait or disorder that is inherited from the parents through genetic information.

Hyperactive Reflexes, Hyperreflexia. Under normal circumstances, the stretch receptors in muscles and tendons send reflexes to the spinal cord and brain telling them when a muscle is stretched and to contract. In some cases, there is a disruption of those signals, resulting in hyperactive reflexes, in which the reflex is greatly exaggerated. This is a common symptom of HSP and PLS.

Hyporeflexia. Weak or absent muscle response when a normal stimulus is applied.

Hereditary Spastic Paraplegia (HSP) is a term for a group of inherited primary upper motor neuron disorders that cause progressive spasticity (stiffness) and weakness of the leg and hip muscles. There is 90+ types of HSP.

Homeopathy. One of the original alternatives to conventional medicine homeopathy uses small, diluted doses of substances that cause illnesses as a means of curing them.

Hydrotherapy. The use of water, from sources such as natural springs, seaweed baths, or mud baths, as medical treatment. Hydrotherapy can be used to treat stress, headaches, muscle aches, arthritis and bursitis.

I

Ichthyosis. A skin disorder causing dry, rough, scaly skin that may be a symptom in some forms of complicated HSP.

Incidence is commonly measured in new cases per 1,000 (or 100,000) of population at risk. Due to rarity and diagnosis inconsistencies, however, it is difficult to estimate an accurate incidence rate for HSP and PLS. The incidence of HSP is estimated at 25,000 in the United States, and 3,000 for PLS.

Immune System. A complex system that is responsible for distinguishing us from everything foreign to us and for protecting us against infections and foreign substances. The immune system works to seek and kill invaders.

Intrathecal. Injection into the innermost membrane surrounding the central nervous system. Usually done by lumbar puncture.

L

Locus. The site on a chromosome where the gene for a particular trait is found. Any one of the alleles for a gene may be found at this site.

Lower Motor Neuron. Nerve cells starting at the spinal cord or brain stem and ending at the muscle fibers. The loss of lower motor neurons leads to weakness, twitching of muscles (fasciculations), and loss of muscle bulk (atrophy).

M

Magnetic Resonance Imaging (MRI). A non-invasive procedure that produces a two-dimensional view of an internal organ or structure, especially the brain and spinal cord.

Massage. The use of touch and various manipulation techniques to move muscles and soft body tissues to relieve stress, tension, and pain.

Minocycline is an antibiotic that has been shown to delay onset and slow progression of symptoms in a mouse model for ALS. Previous studies have shown the antibiotic protects neurons from dying in animal models for a variety of neurologic disorders.

Mitochondria are the principal energy source of the cell. Mitochondria convert nutrients into energy as well as doing many other specialized tasks.

Molecule. The smallest unit of a substance that can exist alone and retain the character of that substance.

Motor Neuron Disease (MND). A group of disorders in which motor nerve cells (neurons) in the spinal cord and brain stem deteriorate.

Muscle Cramp. Unexpected, involuntary, painful shortening of muscles. Usually, a knotting of the muscles is visible. Muscle cramps (spasms) are common in PLS and HSP.

Muscle Weakness. Loss of strength, increased fatigue, loss of coordination and difficulty with motor skills, and lack of ability to carry out certain skills.

Myelin Sheath. The coating on the neurons (nerve cells) that acts as an insulator to speed the conduction of nerve impulses. It is similar to the insulation coating on an electrical wire.

N

Nerves. A bundle of fibers that uses electrical and chemical signals to transmit sensory and motor information from one body part to another. See Neuron.

Neuron is a special cell that transmits electrical signals. It consists of a nucleus, a single axon which conveys electrical signals to other neurons and several dendrites which deliver incoming signals. A single neuron can be several feet in length. Neurons receive input from sensory cells or other neurons and send a signal to muscles or other neurons. Neurons that have sensory input are called “sensory neurons”. Neurons with muscle outputs are called “motor-neurons”, and those that only connect to other neurons are called interneurons.

Nervous System. The system of cells, tissues and organs that regulates the body’s responses to internal and external stimuli.

Neuronal Receptors. Neurons use chemical signaling mechanisms to communicate with one another. These impulses are transmitted at junctions between nerves called synapses. The sending neuron triggers the release of neurotransmitters (chemicals) into the synaptic cleft.

Neuroscience. The scientific disciplines concerned with the development, structure, function, chemistry, pharmacology, clinical assessments and pathology of the nervous system.

Neurotransmitters. Chemical substances that carry impulses from one nerve cell to another; found in the space (synapse) that separates the transmitting neuron’s terminal (axon) from the receiving neuron’s terminal (dendrite).

O

Onset. The time of appearance of the first symptoms of a condition, prior to seeking diagnosis.

Oxidative Stress. Accumulation of destructive molecules called free radicals can lead to motor neuron death. Free radicals damage components of the cells’ membranes, proteins or

genetic material by "oxidizing" them which is the same chemical reaction that causes iron to rust.

P

Paraparesis. Muscle weakness.

Paraplegia. Loss or impairment of motor function of the legs and lower part of the body.

Paresthesia. An unusual sensation which may include numbness, tingling, burning, or prickling. It is sometimes described as "the sensation of pins and needles" or "as if my leg were asleep".

Phenotype. The expression of the genes present in an individual. This may be directly observable (eye color) or apparent only with specific tests (blood type).

Primary Lateral Sclerosis (PLS) is the term for a group of Upper Motor Neuron (UMN) disorders that cause progressive spasticity (stiffness) and weakness in the legs, arms, and speech and swallowing muscles. Also considered a benign variant of ALS.

Proteins are large molecules required for the structure, function, and regulation of the body's cells, tissues, and organs. Each protein has unique functions. Proteins are essential components of muscles, skin, bones and the body as a whole.

R

Recessive gene requires that both corresponding genes are affected in order for the disorder to appear. If the person has one affected gene and one unaffected gene, the disorder will not appear, but the person is considered a "carrier" of the gene. Therefore, usually only children who inherit an affected gene from both parents will show the effects of the disorder.

RNA. A nucleic acid found in all living cells. The primary function of RNA is protein synthesis within a cell.

S

Sclerosis. A hardening within the nervous system, especially of the brain and spinal cord, resulting from degeneration of nervous elements such as the myelin sheath.

Sex Chromosomes. One of the pair of chromosomes linked to gender, known as the X chromosome and the Y chromosome. Females have two X chromosomes; males have an X and a Y chromosome.

Sialorrhea. Drooling resulting from the lack of spontaneous, automatic swallowing to clear excessive saliva in the mouth.

Spasticity (Spastic). Increased muscle tone, resulting in "stiffness". Often involves an exaggeration of the tendon reflexes.

Spasm is a sudden uncontrollable contraction of a muscle. It can be a single muscle cramp along with a burst of pain or a series of spasms lasting several minutes.

Spinal Cord. Part of the central nervous system extending from the brain through the vertebrae of the spinal column. The spinal cord carries information from the body to the brain and signals messages from the brain to the body.

Spinal-bulbar Muscular Atrophy. An X-linked motor neuron recessive disorder characterized by slowly progressive skeletal muscle weakness, especially in the muscles of mouth and throat.

Spinal Muscular Atrophy. A group of predominantly autosomal recessive motor neuron disorders characterized by severe hypotonia and muscle weakness due to lower motor neuron dysfunction.

Stem Cells. Undifferentiated cells that can differentiate into many different cell types when subjected to the right biochemical signals. Stem cells come from embryos, bone marrow and umbilical cords.

Stem Cell Transplant. Transplantation of stem cells from various sources has provided improvement in animal spinal neurodegenerative disease models such as stroke, epilepsy, Parkinson's and spinal cord injury. Human trials are promising, but not complete.

Stretch Receptors. Nerve endings in muscles and tendons that send messages to the spinal cord when the muscles are stretched.

Superoxide Dismutase (SOD). An enzyme that destroys superoxide, a highly reactive form of oxygen. It protects against superoxide damage.

Synapses. Junctions at which neurons communicate with target cells, such as a muscle cell or axon of another neuron. A tiny gap between the ends of nerve fibers across which nerve impulses pass from one neuron to another; at the synapse, an impulse causes the release of a neurotransmitter, which diffuses across the gap and triggers an electrical impulse in the next neuron.

T

Therapeutic Index. A general way of measuring an effective dose of a drug and its toxicity.

Trait. A genetic trait is any sort of distinguishing quality that can be inherited, such as hair color or eye color. Traits are determined by information contained in genes.

U

Uncomplicated HSP. Most forms of HSP are referred to as "uncomplicated" HSP because symptoms are confined to the lower body.

Upper Motor Neurons. Nerve cells (neurons) originating in the brain's motor cortex and running through the spinal cord.

X

X Chromosome. One of the two sex chromosomes. All humans have one X chromosome.

X-Linked. A gene that is located on the X chromosome. Since females have two X chromosomes, females with an affected gene on one X chromosome will not show symptoms unless the corresponding gene on the other X chromosome is also affected.

Since men only have one X chromosome, if they have an affected gene on the X chromosome, they will have the disorder, since there is no “normal” gene to compensate for it.

Y

Y Chromosome. One of the two sex chromosomes. Only males have a Y chromosome.