

Understanding Down Syndrome Features

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In the Down Syndrome Child it is imperative that the parents become educated to understand what is happening inside their child's body and cells. It does help to have a basic knowledge of cell biology, and also a basic knowledge of biochemistry, before attempting to master this important subject. It is the parent's responsibility to become educated in their child's condition to stay abreast of the dynamic needs of the Down Syndrome child.

Down Syndrome behaves much like a degenerative disease. In the Down Syndrome patient there is much biochemical damage done to the cells because of the replicated 21st chromosome that is a part of the patient's genetic make-up.

In order to gain an overview of this subject we will first discuss basic biology and cellular structure as it relates to Down Syndrome. This will be followed up by cellular function as it relates to Down Syndrome with a particular focus on nerve cells and neurotransmitters.

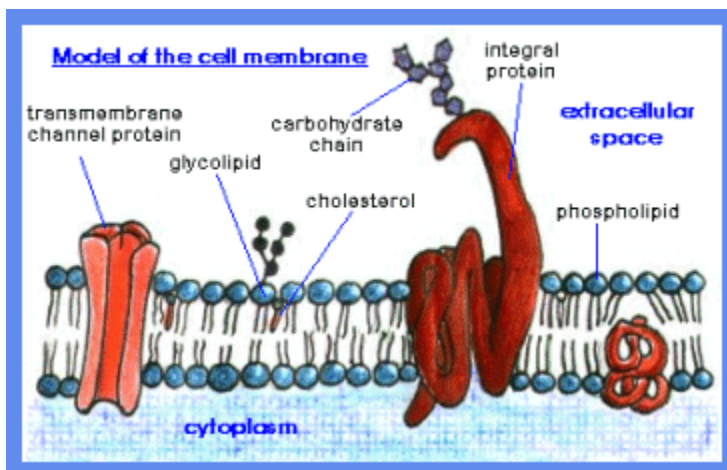
Cellular structure

The cell consists of an outer cell membrane, the liquid cytoplasm interior, the nucleus which contains the DNA, and various organelles to perform different tasks within the cell.

The Cell Membrane

The cell membrane is composed of a bi-layer of phospholipids. These phospholipids repel water on one end of the phospholipids and attract water on the other end. The outer ends of the phospholipid membrane is hydrophilic or "water loving". The other end is hydrophobic or "water hating" and is comprised of fatty acid chains. These fatty acid chains make up the inside of the phospholipids membrane.

These phospholipids spontaneously line up to form the cell membrane in bilipid layers by putting their heads outward and butts tail to tail with another phospholipid to form a unique selectively-permeable membrane.



This membrane has the ability to keep fat and water-based chemicals out of the cell while still allowing water, oxygen, and carbon dioxide and other nutrients to pass through. It also allows enough flexibility to incorporate different transport proteins among them.

Some transport proteins move nutrients and chemicals into the cell while other transport proteins move waste material out of the cell. This is critical for healthy function of the cell. For the transport to work correctly, the cell membrane must remain fluid for the proteins to change shape as it transports different shape and sized nutrients, chemicals, and wastes in or out of the cell. If the proteins are limited in their flexibility by limited flexibility of the cell wall they cannot properly perform their functions. Fluid movement of the cell walls is lost in the natural aging process of the body and also by transfatty acid incorporation into the cell membrane. Loss of fluid movement in the cell wall is accelerated in the Down Syndrome patient and is a particular problem for Down Syndrome patients.

In the Down Syndrome patient, the fatty acid portions of the cell membrane are more prone to chemical damage than the genetically "normal" subject. This damage is a process called lipid peroxidation, or "oxidative stress." The end result of this oxidative stress is that the cell membrane loses its flexibility and the transport proteins can no longer function properly. When transport proteins do not function properly, chemical waste begins to accumulate inside the cell and will ultimately kill the cell. The cell also starves for nutrients because it can no longer effectively transport its food from the outside world in.

Vitamin E, along with other antioxidants, is essential to protect against oxidative stress. While dietary intake of essential fatty acids provides material to repair damaged cell membranes.

The Cell Nucleus

The cell nucleus is where the DNA (Deoxyribonucleic Acid) resides in a cell. This is where the process of "protein synthesis" begins.

To make a protein, a portion of the DNA inside of the nucleus unzips to generate a section of RNA (a molecule similar to DNA but which is used in the making of proteins). A new strand of RNA is formed to encode the unzipped DNA portion. Amino acids are the building blocks of proteins that are then strung together according to the instructions in the RNA to make the protein.

Once the protein is made, it moves to its place in the cell where it is designed to perform a specific job.

DNA production in Down Syndrome is one of the places where the extra 21st chromosome comes into play. Chromosomes are made of DNA. But because there is an extra copy of the 21st chromosome in each cell of the Down Syndrome patient, therefore there are more copies of proteins produced than are needed for the normal chemical reactions that occur in a cell. In some cases, these extra proteins cause metabolic imbalances, which lead to an excess (over-expression) of some chemicals and a deficiency (under-expression) of others in the body. (Metabolism is the sum total of all the chemical reactions in the body.)

An example of an excess that causes metabolic imbalance is the superoxide dismutase or SOD gene which is on the 21st chromosome. SOD is half of a two-step process to take a poisonous form of oxygen, called superoxide, and turn it into oxygen and water. This SOD, because it is manufactured on the replicated 21st chromosome in Down Syndrome, is made in larger quantities than normal. The enzymes used in the second half of this process are either catalase or glutathione peroxidase. Glutathione peroxidase and catalase are not over-replicated as is SOD and is therefore not expressed in balance to SOD.

In the first step of this two-step process, SOD takes the toxic superoxide and turns it into hydrogen peroxide. However, in step two of the Down Syndrome patient, there is not enough glutathione peroxidase and catalase to break down the hydrogen peroxide into oxygen and water fast enough. This leaves excess hydrogen peroxide in the cells until it finally reaches toxic levels and eventually destroys the cell from within. This is called Oxidation (the same process that rusts metals and turns apples brown). Hydrogen Peroxide is especially toxic to nerve cells. Hydrogen peroxide also reacts when exposed to other chemicals and elements in the body such as iron that can cause even more cellular damage.

To address this problem what is required is a variety of anti-oxidants, (mostly vitamins and minerals) that keep the oxidative damage from occurring, and in some cases, even repair the cellular damage and loss of fluidity that has occurred.

Antioxidants include Vitamins A (beta-carotene), C, and E, the minerals zinc and selenium, alpha Lipoic Acid, Co-Q10, inositol and bioflavonoids.

Structure of a Nerve Cell

One significant difference between nerve cells (neurons) and other body cells is that mature neurons do not typically reproduce. If a neuron dies, if for example by oxidative damage, then that cell is normally lost forever.

The brain and spinal cord are comprised mainly of nerve cells (neurons). There are significant differences between a nerve cell and other cells in the body. Nerve cells have a number of "tentacles" which stick out from the cell body called dendrites. Dendrites receive messages from other nerve cells. Then there is the axon of the nerve cell that protrudes farther away from the cell body than do the dendrites. The axon sends the message to be picked up by other cell's dendrites. There are many dendrites on a nerve cell but there is only one axon.

The nerve impulse always flows to a dendrite, through the cell body, and down the axon to the next cell's dendrites. The next cell repeats the process until the message reaches its destination at the end of the neural pathway. At the end of a peripheral neural pathway, the nerve cell will connect to a muscle or an organ, etc in the body system.

The process of learning causes new connections to be made from one neuron to another. As the connections are repeated over and over, the cells form a deeper, wider neural pathway much like a cow-trail in a pasture. The more cows that repeatedly use the trail the deeper and wider the trail becomes. When connections are unused for a

period of time, they begin to die off and are pruned by the body much like the cow-trail will fade and overgrow when the cows quit using it.

These neural pathways are ultimately covered with an insulating coating called myelin. Myelin is a fatty sheath that is formed by another type of cell called a Schwann cell. The whole human brain takes years to myelinate starting in the lower parts of the brain and insulating upwards to finally myelinate the cerebral cortices. This process starts soon after conception and continues on until about 20-23 years of age. The myelin sheath forms by wrapping itself tightly around the axon like wrapping electrical tape around a live electrical wire. Myelin assists the neuron to send neural chemical messages more quickly and keeps the signals from bleeding over into other impulses from proximal neurons.

Myelination of the brain has a direct correlation to brain development and lack of myelination or slow myelination correlates directly to developmental delays. Beginning at about two months of age, myelin formation in Down Syndrome slows in the brain as compared to genetically normal children.

The Schwann cells which make up the myelin sheath consist mainly of the cell membrane and very little cytoplasm. The cell membranes of these cells are made up of a lipid-protein mixture that is slightly different than other types of cells. This makes the myelin to consist of a higher proportion of lipids or fats than any other type of cell. A majority of these fats are docosahexaenoic acid, or DHA. DHA is an essential fatty acid and part of the omega 3,6,9 supplements.

Function of Nerve Cells

When a neuron is stimulated, an electrical/chemical impulse is made which travels down the dendrites, through the cell body and then down the axon. At the end of the axon are bell-shaped synaptic knobs which connect to other neurons. The synaptic knobs contain the chemicals called neurotransmitters. The synapse is the gap between the two nerve cells where the exchange of neurotransmitter chemicals takes place. When the neuro impulse reaches the synaptic knob, the synaptic knob releases a neurotransmitter into the synapse. The next neuron in line uses receptors to detect the neurotransmitter. If enough neurotransmitter chemical has been released for the next nerve to fire then the neuro/chemical transmission continues through that neuron and then on to the next neuron and so on.

After the neurotransmitter has been released into the synaptic gap, the neurotransmitter is either reabsorbed into the synaptic knob or is destroyed by enzymes in the synapse. This process of removing the neurotransmitter from the synaptic gap prevents the recipient neuron from being continuously stimulated.

The end result of the synaptic firing of neurotransmitters is thought processes, muscle movement, organ and glandular function, and pain sensation to name a few.

Function of Neurotransmitters in Down Syndrome

Acetylcholine

Acetylcholine is the primary neurotransmitter in the central nervous system that activates muscle fibers, releases hormones, and is essential in the learning and memory processes of the brain. Choline and inositol are what the body uses to produce acetylcholine and can be supplemented to help overcome a shortfall in the body.

In Down Syndrome there are fewer acetylcholine receptors in the brain causing some of the memory and learning problems seen in Down Syndrome. This is because the nerves involved in these two processes are stimulated with less frequency and therefore have less opportunity to develop normally.

A lack of acetylcholine also accounts for faulty communications in the endocrine system resulting in glandular and hormonal shortfalls in the Down Syndrome patient. One such shortfall is in Human Growth Hormone (HGH) causing problems as short stature.

Serotonin

Serotonin is a neurotransmitter that controls blood pressure, sleep cycles, peristalsis (the movements of the intestines), and control of behavior (mood and aggression). Tryptophan is a primary amino acid which is converted into serotonin and requires vitamins B-6 and C for that conversion. Serotonin levels in the blood and cerebral spinal fluid are deficient in the Down Syndrome patient.

Serotonin is also used to produce Melatonin which works in the sleep center of the brain to promote proper sleep. The brain releases the highest levels of Human Growth Hormone during the stage of sleep called Rapid Eye Movement (REM) sleep which helps to provide normal growth.

Dopamine

Dopamine is the neurotransmitter that is involved what is called high order association which is the process in the brain that brings two separate pieces of information together for critical thinking. Dopamine is very important to the reward/pleasure sensation. Dopamine also controls physical movements and coordinates processes in the brain. Tyrosine is a precursor to the production of dopamine.

Dopamine levels are decreased in some Down Syndrome patients Though not all Down Syndrome are found to be deficient. Dopamine is also converted to another neurotransmitter called norepinephrine and then further converted to another neurotransmitter called norepinephrine.

Norepinephrine

Norepinephrine helps the brain to process sensory input from eyes and ears and tactile functions. It also regulates sleep cycles and influences anxiety and arousal.

Tyrosine supplements help to increase the levels of norepinephrine which is found to be decreased in Down Syndrome patients.

In conclusion: A comprehensive Neuro Development plan along with an appropriate diet and supplementation plan specific to the individual Down Syndrome patient are critical for the health and well-being of the Down Syndrome patient.