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A Literature Review of Hereditary Sensory and Autonomic Neuropathy (HSAN/HSN)

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Abstract: **Hereditary Sensory and Autonomic Neuropathy (HSAN/HSN) are genetically heterogeneous disorders of the peripheral nervous system that affect the sensory and autonomic neurons. Hereditary Sensory and Autonomic Neuropathy (HSAN/HSN) there are total of 13 different genes are mutated and the disease is transmitted through Autosomal recessive and Autosomal dominant inheritance. Which all 13 genes will cause different types of Hereditary Sensory and Autonomic Neuropathy (HSAN/HSN) each type of Hereditary Sensory and Autonomic Neuropathy (HSAN/HSN) has different symptoms according to each type.**

Objective: In order to collect information about the Hereditary Sensory and Autonomic Neuropathy (HSAN/HSN)

Methods: A literature review on Hereditary Sensory and Autonomic Neuropathy (HSAN/HSN) related articles and researches

Keywords: **Hereditary sensory and Autonomic neuropathy, Hereditary Sensory and Neuropathy, Hereditary sensory and Autonomic neuropathy Type I, Hereditary sensory and Autonomic neuropathy Type IA, Hereditary sensory and Autonomic neuropathy Type IB, Hereditary sensory and Autonomic neuropathy Type IC, Hereditary sensory and Autonomic neuropathy Type ID, Hereditary sensory and Autonomic neuropathy Type IE, Hereditary sensory and Autonomic neuropathy Type II, Hereditary sensory and Autonomic neuropathy Type IIA, Hereditary sensory and Autonomic neuropathy Type IIB, Hereditary sensory and Autonomic neuropathy type III, Hereditary sensory and Autonomic neuropathy Type IV, Hereditary sensory and Autonomic neuropathy Type V, Hereditary sensory and Autonomic neuropathy Type VI, Hereditary sensory and Autonomic neuropathy Type VII, Hereditary sensory and Autonomic neuropathy Type VIII, HSAN, HSAN I, HSAN IA, HSAN IB, HSAN IC, HSAN ID, HSAN IE, HSAN IIA, HSAN IIB, HSAN III, HSAN IV, HSAN V, HSAN VI, HSAN VII, HSAN VIII, HSN, Congenital sensory neuropathy, Familial dysatonomia, Congenital insensitivity to pain with anhidrosis, CIPA, Congenital insensitivity to pain, CIP.**

I. INTRODUCTION

Hereditary Sensory and Autonomic Neuropathy (HSAN/HSN) is a genetic disorders a type caused by a single gene disorder which describes the neurodegenerative disease that progressive loss of function that predominantly affects the peripheral sensory nerves. There are a total of 8 types different clinical entities have been described under hereditary sensory and autonomic neuropathies. Each Hereditary Sensory and Autonomic Neuropathy (HSAN/HSN) are all characterized such as Cause, Gene, Symptom.

Hereditary Sensory and Autonomic Neuropathy Type I (HSAN I / HSN I)

Hereditary Sensory and Autonomic Neuropathy Type I (HSAN I / HSN I) is a condition characterized by nerve abnormalities in the legs and feet. People with Hereditary Sensory and Autonomic Neuropathy Type I (HSAN I / HSN I) have tingling, weakness, and a reduced ability to feel pain and sense hot and cold in their leg and feet. somebody affected individuals do not lose sensation, but instead feel shooting pains. When the disorder progresses without treatment the sensory abnormalities can affect the hands, arms, shoulders, and abdomen. People with Hereditary Sensory and Autonomic Neuropathy Type I (HSAN I / HSN I) often have open sores on their feet, hands or infections of the soft tissue of the fingertips. The patient has a slow self-healing because affected individuals cannot feel the pain of these sores before the infection sores is knows, the sores may have dead tissue all ready. It rarely, people with Hereditary Sensory and

Autonomic Neuropathy Type I (HSAN I / HSN I) may develop hearing loss caused by abnormalities of the inner ear and may have Anhidrosis, making the body's temperature venting unable to do therefore or fainting without consciousness.

Hereditary Sensory and Autonomic Neuropathy Type I (HSAN I / HSN I) is the most common from among the 8 type but Hereditary Sensory and Autonomic neuropathy Type I (HSANI/HSN I) can be divided into 5 types is Hereditary Sensory and autonomic Neuropathy Type IA (HSAN IA/ HSN IA), Hereditary Sensory and autonomic Neuropathy Type IB (HSAN IB/ HSN IB), Hereditary Sensory and autonomic Neuropathy Type IC (HSAN IC/ HSN IC), Hereditary Sensory and autonomic Neuropathy Type ID (HSAN ID/ HSN ID), Hereditary Sensory and autonomic Neuropathy Type IE (HSAN IE/ HSN IE). The only difference between type is the genes are mutated but the symptoms are all the same.

Cause of Hereditary Sensory and Autonomic Neuropathy Type IA (HSAN IA/ HSN IA)

Hereditary Sensory and Autonomic Neuropathy Type IA (HSAN IA/ HSN IA) which inherited as an autosomal dominant trait the mutated gene is the Serine Palmitoyltransferase Long Chain Base Subunit 1 (SPTLC 1) provides instructions for making one part of an enzyme called serine palmitoyltransferase (SPT). This Enzyme is involved in making lipids called sphingolipids, which are essential components of cell membranes, have many functions within cells, such as protecting cell surfaces from various factors that can cause cell damage. If Serine Palmitoyltransferase Long Chain Base Subunit 1 (SPTLC 1) mutations reduce the amount of Serine Palmitoyltransferase Long Chain Base Subunit 1 (SPTLC 1) that is produced and result in an SPT enzyme with decreased function or there was a change in the work which made Deoxysphingoid bases are produced. This step made sphingolipids decrease. And as more and more Deoxyshingoid bases accumulate, they become toxic to nerve cells, causing nerve cell destruction resulting in loss of sensation and muscle weakness. This gene was identified in patients with hereditary sensory neuropathy type I.

Cause of Hereditary Sensory and Autonomic Neuropathy Type IB (HSAN IB/ HSN IB)

It is not yet possible to determine the exact mutation genes present in chromosome 3 and gene inherited as an autosomal dominant.

Cause of Hereditary Sensory and Autonomic Neuropathy Type IC (HSAN IC/ HSN IC)

Hereditary Sensory and Autonomic Neuropathy Type IC (HSAN IC/ HSN IC) which inherited as an autosomal dominant trait the mutated gene is the Serine Palmitoyltransferase Long Chain Base Subunit 2 (SPTLC 2). This gene encodes a long chain base subunit of serine palmitoyltransferase. Serine palmitoyltransferase, which consists of two different subunits, is the initial enzyme in sphingolipid biosynthesis. It catalyzes the pyridoxal 5'-phosphate dependent condensation of L-serine and palmitoyl CoA to 3-oxosphinganine. Mutations in this gene were identified in patients with hereditary sensory neuropathy type I.

Cause of Hereditary Sensory and Autonomic Neuropathy Type ID (HSAN ID/ HSN ID)

Hereditary Sensory and Autonomic Neuropathy Type ID (HSAN ID/ HSN ID) which inherited as an autosomal dominant trait the mutated gene is the Atlastin GTPase 1 Gene (ATL1). The protein encoded by this gene is a GTPase and a Golgi body transmembrane protein. The encoded protein can form a homotetramer and has been shown to interact with spastin and with mitogen-activated protein kinase. This protein may be involved in axonal maintenance as evidenced by the fact that defects in this gene are a cause of spastic paraplegia type 3. Three transcript variants encoding two different isoforms have been found for this gene.

Cause of Hereditary Sensory and Autonomic Neuropathy Type IE (HSAN IE/ HSN IE)

Hereditary Sensory and Autonomic Neuropathy Type IE (HSAN IE/ HSN IE) which inherited as an autosomal dominant trait the mutated gene is the DNA (cytosine-5)- methyltransferase 1 Gene (DNMT1). This gene encodes an enzyme that transfers methyl groups to cytosine nucleotides of genomic DNA. This protein is the major enzyme responsible for maintaining methylation patterns following DNA replication and shows a preference for hemi-methylated DNA. Methylation of DNA is an important component epigenetic gene regulation. Aberrant methylation patterns are found in human tumors and associated with developmental abnormalities. Variation in this gene has been associated with Hereditary Sensory and Autonomic Neuropathy type IE (HSAN IE/HSN IE). Alternative splicing results in multiple transcript variants.

Hereditary Sensory and Autonomic Neuropathy Type II (HSAN II/ HSN II)

Hereditary sensory and Autonomic Neuropathy Type II (HSAN II/HSN II) or Congenital sensory neuropathyit is a condition that affects sensory nerve cells that transmit information about pain, temperature, touch. In some patient with Hereditary sensory and Autonomic Neuropathy Type II (HSAN II/HSN II) may have autonomic disorders. This puts you

at risk of worsening your heart rate, digestion and breathing. which these disorders start from childhood. The first sign of Hereditary sensory and Autonomic Neuropathy Type II (HSAN II/HSN II) is numbness in the hands and feet. Soon after, he lost his senses. Babies with Hereditary sensory and Autonomic Neuropathy Type II (HSAN II/HSN II) will have trouble sucking. Causing inability to eat or may have a short pause in breathing Digestive problems, such as backflow of stomach acid The taste of food has deteriorated. and the response may be slower because all myelin fibers are gone. Hereditary Sensory and Autonomic neuropathy Type I (HSANI/HSN I) can be divided into 2 types is Hereditary Sensory and autonomic Neuropathy Type IIA (HSAN IIA/ HSN IIA) and Hereditary Sensory and autonomic Neuropathy Type IIB (HSAN IIB/ HSN IIB) The only difference between type is the genes are mutated but the symptoms are all the same.

Cause of Hereditary Sensory and Autonomic Neuropathy Type IIA (HSAN IIA/ HSN IIA)

Hereditary Sensory and Autonomic Neuropathy Type IIA (HSAN IIA/ HSN IIA) which inherited as an autosomal recessive trait the mutated gene is the WNK Lysine Deficient Protein Kinase 1 (WNK1). The WNK1 gene is the gene that provides instructions for making the protein WNK1, which is important in many functions in the body. For example, it regulates blood pressure as well as pain. WNK1 protein contains several isoforms such as protein L-WNK1 which is found throughout the body, protein KS-WNK1 which can be found in kidney cells. Both act as kinases. To alter the activity of an enzyme that alters the activity of proteins by adding oxygen and phosphorus atoms. at a specific location where both are transmitting controls. Sodium and potassium get into cells. In the kidneys, sodium channels help deliver sodium to specialized cells and then transfer it into the bloodstream. makes it possible to maintain sodium in the body called the reabsorption process. The potassium channels deal with excess potassium by eliminating it in the form of urine. Protein L-WNK1 increases potassium reabsorption and decreases potassium excretion. Protein KS-WNK1 has opposite effect on protein L-WNK1. Sodium and potassium affect blood pressure regulation and protein WNK1 also plays a role in Regulates membrane channels that transport chlorite ions, which are important in the activation of neurons. Mutations in the WNK1 gene cause HSAN1A and Pseudohypoaldosteronism type2, in which the WNK1 gene must mutate more than 12 genes. Therefore, it will result in the gene being shortened and unable to work. All of these mutations lead to an abnormally shortened WNK1/HSN2 protein that is probably nonfunctional. People with Hereditary Sensory and Autonomic Neuropathy Type IIA (HSAN IIA/ HSN IIA) have a reduction in the number of sensory neurons; however, the role that the abnormal WNK1/HSN2 protein plays in that loss is unclear. The loss of sensory neurons results in the signs and symptoms of Hereditary Sensory and Autonomic Neuropathy Type IIA (HSAN IIA/ HSN IIA).

Cause of Hereditary Sensory and Autonomic Neuropathy Type IIB (HSAN IIB/ HSN IIB)

Hereditary Sensory and Autonomic Neuropathy Type IIB (HSAN IIB/ HSN IIB) which inherited as an autosomal recessive trait the mutated gene is the Reticulophagy Regulator 1 Gene (RETREG1). The RETREG1 gene provides instructions for making a protein that is involved in a cellular process called autophagy. Cells use this process to recycle cell parts and break down certain proteins when they are no longer needed. In particular, the RETREG1 protein helps direct autophagy of a cell structure called the endoplasmic reticulum, which is important in protein processing and transport. Autophagy may be a way for the cell to remove parts of the endoplasmic reticulum when they are no longer needed or to break down excess or abnormal proteins that are being processed within the structure. The RETREG1 gene mutations may lead to an abnormally short and nonfunctional protein. The resulting lack of functioning RETREG1 protein impairs autophagy of the endoplasmic reticulum and alters the structure of the Golgi apparatus in sensory and autonomic neurons.

Hereditary Sensory and Autonomic Neuropathy Type III (HSAN III/ HSN III)

Hereditary Sensory and Autonomic Neuropathy type III (HSAN III/HSN III), also known as familial dysatonomia, is a hereditary disorder that affects the development and survival of autonomic nervous system. The first symptoms you see are hypotonia, scoliosis, cyanosis, poor bone quality, have heart and liver problems have speech and walking developmental problems delayed vision problems, difficulty eating food, lack of tears, easy lung infections difficult to maintain body temperature poor balance. Many patients died in childhood.

Cause of Hereditary Sensory and Autonomic Neuropathy Type III (HSAN III/ HSN III)

Hereditary Sensory and Autonomic Neuropathy Type III (HSAN III/HSN III) which inherited as an autosomal recessive trait the mutated gene is the Elongator complex protein 1 Gene (ELP1). The ELP1 gene provides instructions for making a protein called elongator complex protein 1 (ELP1). This protein is found in a variety of cells throughout the body. It is part of a six-protein complex called the elongator complex. The elongator complex plays a key role in

transcription, the process that transfers information in genes to the cell machinery that makes proteins. elongator complex is important for the transcription of proteins that affect the cytoskeleton and cell motility. The cytoskeleton and cell motility are essential for the growth and development of cells. If a mutation occurs in the ELP1 gene, more than two copies of the mutation must occur in a cell. This mutation can disrupt how information in the ELP1 gene is spliced together during transcription. As a result of this splicing error, a reduced amount of ELP1 protein is produced. Reduced amounts of ELP1 protein may impair the growth and development of nerve cells by disrupting the cytoskeleton and cell motility.

Hereditary Sensory and Autonomic Neuropathy Type IV (HSAN IV/ HSN IV)

Hereditary Sensory and Autonomic Neuropathy Type IV (HSAN IV/HSN IV), also known as Congenital insensitivity to pain with anhidrosis (CIPA) it is an autosomal recessive inherited disease that manifests only when both parents are carriers. Hereditary sensory neuropathy type IV (HSAN IV/HSN IV) is a rare genetic disorder characterized by the loss of sensation especially in the feet and legs The sensory loss is due to abnormal functioning of small unmyelinated nerve fibers and portions of the spinal cord that control responses to pain and temperature Hereditary sensory neuropathy type IV (HSN4) is a rare genetic disorder characterized by the loss of sensation especially in the feet and legs The sensory loss is due to abnormal functioning of small unmyelinated nerve fibers and portions of the spinal cord that control responses to pain and temperature. Symptoms of CIPA are insensitivity to pain, anhidrosis, intellectual disability. The ability to sense all pain is absent, resulting in repeated injuries, scarring, infection of the skin; multiple bone fractures and recurrent joint dislocations resulting in joint deformity. Sense of touch, vibration, and position are normal. Anhidrosis predisposes to recurrent febrile episodes that are often the initial manifestation of CIPA. Intellectual disability of varying degree is observed in most affected individuals; hyperactivity and emotional lability are common.

Cause of Hereditary Sensory and Autonomic Neuropathy Type IV (HSAN IV/ HSN IV)

Hereditary Sensory and Autonomic Neuropathy Type IV (HSAN IV/HSN IV) which inherited as an autosomal recessive trait the mutated gene is the Neurotrophic Receptor Tyrosine Kinase 1 gene (NTRK1). The NTRK1 gene provides instructions for making a protein that is essential for the development and survival of nerve cells, especially those that transmit information about sensations such as pain, temperature, and touch. The NTRK1 protein is found on the surface of cells, particularly sensory neurons. It acts as a kinase, which is an enzyme that changes the activity of other proteins by adding a cluster of oxygen and phosphorus atoms at specific positions. This process is called phosphorylation. The NTRK1 protein is turned on when another protein called nerve growth factor beta (NGFβ) attaches to it and signals the NTRK1 protein to phosphorylate itself. Then, the activated NTRK1 protein phosphorylates other proteins; this process is needed to transmit signals for cell growth and survival. Mutations in the NTRK1 gene cause Congenital insensitivity to pain with anhidrosis (CIPA), a condition characterized by anhidrosis. Many of the NTRK1 gene mutations lead to a protein that cannot be activated by phosphorylation, which means the mutated NTRK1 protein cannot transmit cell growth and survival signals to neurons. Without the proper signaling, neurons die by a process of self-destruction called apoptosis. Loss of sensory neurons leads to the inability to feel pain in people with CIPA. In addition, people with Congenital insensitivity to pain with anhidrosis (CIPA)lose the nerves leading to their sweat glands, which causes the anhidrosis seen in affected individuals.

Hereditary Sensory and Autonomic Neuropathy Type V (HSAN V/ HSN V)

Hereditary Sensory and Autonomic Neuropathy Type V (HSAN V/HSN V), also known as Congenital insensitivity to pain (CIP) It is a condition that affects the sensory nerve cells. It is a condition that affects the sensory nerve cells that are born in the baby. in which the baby loses sensation temperature perception and deep pain perceptions, leading to neuropathic arthropathy, a disease in which the tissue and bone around the joint are destroyed. The patient may have a deficiency of sweat (Anhidrosis).

Cause of Hereditary Sensory and Autonomic Neuropathy Type V (HSANV/HSN V)

Mutations in the Nerve Growth Factor Gene (NGF) cause Hereditary Sensory and Autonomic Neuropathy Type V (HSAN V/HSN V). The NGF gene provides instructions for making a protein called nerve growth factor beta (NGFβ). This protein is important in the development and survival of nerve cells, especially those that transmit pain, temperature, and touch sensations. The NGFβ protein functions by attaching to its receptors, which initiates signaling pathways inside the cell. The NGFβ protein can bind to two different receptors, the NTRK1 receptor or the p75NTR receptor. Both receptors are found on the surface of sensory neurons and other types of neurons. The binding of the NGFβ protein to the

NTRK1 receptor signals these neurons to grow. Mutation of the NGF gene leads to the production of a protein that cannot bind to the receptor and does not transmit signals properly. Without the proper signaling, sensory neurons die and pain sensation is altered, resulting in the inability of people with HSAN5 to feel pain.

Hereditary Sensory and Autonomic Neuropathy Type VI (HSAN VI/ HSN VI)

Hereditary sensory and autonomic neuropathy type VI (HSAN IV/HSN IV) is a severe autosomal recessive disorder characterized by neonatal hypotonia, respiratory and feeding difficulties, lack of psychomotor development, and autonomic abnormalities including labile cardiovascular function, lack of corneal reflexes leading to corneal scarring, areflexia, and absent axonal flare response after intradermal histamine injection

Cause of Hereditary Sensory and Autonomic Neuropathy Type VI (HSAN VI/HSN VI)

Hereditary Sensory and Autonomic Neuropathy Type VI (HSAN VI/HSN VI) is a recessive genetic disorder that arises because of mutations in the dystonin gene (DST). Number of heterozygous mutations in DST that result in milder forms of the disease.

Hereditary Sensory and Autonomic Neuropathy Type VII (HSAN VII/ HSN VII)

Hereditary Sensory and Autonomic Neuropathy Type VII (HSAN VII/HSN VII), also known as Congenital insensitivity to pain with hyperhidrosis and gastrointestinal dysfunction is a genetic condition that causes the inability to feel pain, excessive sweating, and gastrointestinal issues. signs and symptoms of Hereditary Sensory and Autonomic Neuropathy Type VII (HSAN VII/HSN VII) usually appear at birth or during infancy.

Cause of Hereditary Sensory and Autonomic Neuropathy Type VII (HSAN VII/ HSN VII)

Hereditary Sensory and Autonomic Neuropathy Type VII (HSAN VII/ HSN VII) which inherited as an autosomal dominant trait the mutated gene is the Sodium Voltage-Gated Channel 11 subunit alpha gene (SCN11A) so it have a 1 in 2 or 50% chance of passing the condition on to each of their children. This protein mediates the voltage-dependent sodium ion permeability of excitable membranes. Assuming opened or closed conformations in response to the voltage difference across the membrane, the protein forms a sodium-selective channel through which sodium ions may pass in accordance with their electrochemical gradient. It is a tetrodotoxin-resistant sodium channel isoform. Also involved, with the contribution of the receptor tyrosine kinase NTRK2, in rapid BDNF-evoked neuronal depolarization.

Hereditary Sensory and Autonomic Neuropathy Type VIII (HSAN VIII/ HSN VIII)

This is a rare autonomic neuropathy. It is characterized by a congenital impaired sensation and acute pain or inflammation. with no indication of heat or cold; other symptoms include no corneal reflexes resulting in corneal scarring; Less sweating and tearing wounds and easy skin infections Sensory with large fibers, such as a light touch vibration and perception of abnormal sensations

Cause of Hereditary Sensory and Autonomic Neuropathy Type VIII (HSAN VIII/ HSN VIII)

Hereditary sensory and autonomic neuropathy type 8 is a disease characterized by sensory disorders that are inherited through autosomal recessive. It is caused by a mutation in the PR/SET Domain 12 gene (PRDM12) that causes impaired sensory neuron development. This gene encodes a transcriptional regulator of sensory neuronal specification that plays a critical role in pain perception.

Diagnosis

A diagnosis of Hereditary Sensory and Autonomic Neuropathy (HSAN/HSN) is based on upon identification of symptoms, detail patient history and family history consistent with Hereditary Sensory and Autonomic Neuropathy (HSAN/HSN). Diagnosis can be used Electromyography (EMG) the electromyography, show h ow well a muscle responds to the nerve and can determine whether muscle weakness is caused by the muscle themselves or by the nerves that control the muscle. Surgical removal and microscopic examination (biopsy) in order to check the condition of the nerve. Molecular genetic testing can confirm a diagnosis in some case because molecular genetic testing can detect mutations in the specific gene.

Treatment

This disease like a nuclear bomb because we can't feel pain or temperature so Current treatment of Hereditary Sensory and Autonomic Neuropathy every Type is symptomatic treatment only don’t have specific treatment. Therefore, the patient must have a physical examination almost all the time.

II. CONCLUSION

Hereditary Sensory and Autonomic Neuropathy (HSAN/HSN) is a Genetic Disorders that is transmitted through genes. There are eight types of HSAN, each of which has a cause. Mutated genes and different symptoms. The disease is like a nuclear explosion that is always ready to explode because the patient is unable to feel pain or feel cold at all. There is currently no specific treatment, only symptomatic treatment.

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