Genetics and Genomic: Spinal Muscular Atrophy

Name

University

Course

Date

**History of Spinal Muscular Atrophy (SMA)**

Spinal Muscular Atrophy (SMA) was first discovered in children by scientists Johann Hoffman and Guido Werdnig from Germany and Austria in 1891. They noticed some babies developed muscle weakness within the first months of life, and the condition was genetic. Further investigation indicated the feebleness was caused by the loss of particular spinal cord nerve cells called anterior horn cells or motor neurons. The progressive muscle wasting or atrophy observed overtime proved the disease was indeed SMA. The condition is first detected when babies have difficulty crawling, controlling head, sitting, and walking. Some severe types of SMA have more severe effects, such as swallowing, feeding, and breathing problems. Now that it is mentioned, there are several categories of SMA. This rare genetic disorder develops into four forms. Type 1 is the most common Werdnig-Hoffman, while Type 2 is an intermediate SMA. Type 3 is milder, while Type 4 is very seldom.

**Statistical Information**

Studies conducted on all classifications of SMA indicate the predominance of around one to two per 100,000 persons sampled. However, some studies show different prevalence. For instance, a 1992 study conducted in Bologna, Italy, resulted in 6.56 per 100,000 individuals. Three similar inquiries in Scandinavia indicated a dominance of 4.18 per 100,000 people aged 18 years and below. The prevalence of teenagers aged below 16 years ranged between 2.78 to 3.23 per 100,000 individuals (Verhaart et al., 2017). The incongruent findings indicate there could be regional differences in the incidences of SMA. The differences in prevalence could also be caused by genetics. The disease is inherited through an autosomal recessive configuration, where every type of SMA in a cell mutates, producing two copies. It is no wonder that although parents of individuals with SMA carry a copy of autosomal recessive gene in most cases, there are no observable signs of the condition (MedlinePlus, 2021). Its onset determines the severity of the disease. According to De Sanctis et al. (2018),although the disease attacks children at an age, its manifestation in infancy and early childhood is related to worse results. A more positive prognosis is recorded for patients who develop the signs later in childhood or adolescence.

**Signs and Symptoms**

The signs and symptoms advance according to the SMA type. For individuals with SMA1, indications of the disease appear at birth or within six months. Children have difficulty sucking and swallowing while not meeting the usual milestone of sitting or holding their heads up. SMA2 mainly occurs between six and eighteen months and tends to affect hind limbs. Babies are unable to walk, a condition that stays until adulthood. SMA3 manifests after 18 months, but symptoms may remain hidden until early adulthood. Minors experience recurrent respiratory infections and general muscle weakness. Over time, standing or walking becomes complicated. Finally, SMA4 manifests slowly, and signs may be unnoticeable until the mid-30s. It rarely affects the population, and most people retain their mobility and live fully.

**Screening, Diagnosis, and Treatment**

Since SMA symptoms are similar to other muscle diseases, most screening tools are structured the same across the board. Doctors mostly use genetic testing to look for mutations that might cause the condition. Electromyography (EMG) shows how muscles receive nerve signals. Thirdly, muscle biopsy involves the removal of a muscle sample and examining it under a microscope. Finally, the blood test is sought to evaluate the levels of creatine kinase often released by weakening muscles. A conclusive SMA diagnosis is done after the physician collects the medical history and performs a physical exam (Pera et al., 2020). Despite the existence of a wide range of screening tools, there is no explicit cure for SMA. However, assistive medications are recommended to assist in the management of the conditions. Some of the drugs used include disease-modifying medications such as Spinraza for children and Evrysdi for adults and older children that control the production of motor neuron protein. Gene replacement therapy is administered to babies under two years using the Zolgensma drug to replace faulty or missing mechanical neurons. Alternatively, support devices such as wheelchairs and braces are used to assist in independence maintenance.

**Use of Pharmacogenomics**

Pharmacogenomics is the study of how people’s genes affect their response to drugs. Currently, the field is used to address SMA as the one-size-fits-all medical approach is often ineffective. The FDA approved Zolgensma as the main drug for this population of interest as its mechanism of action addresses the genetic cause that lead to SMA. Notably, the gene therapy mentioned was approved to treat children aged less than two years. The research of pharmacogenomics is expected to continue as inheritance remains the primary cause of the condition.

References

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