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My46 Trait Profile

Multiple Sclerosis

Multiple sclerosis is a chronic neurodegenerative disease of the central nervous system. It is caused by a combination of genetic and environmental risk factors.

Characteristics of Multiple Sclerosis

Multiple sclerosis (MS) is a neurodegenerative disease of the central nervous system (CNS) characterized by inflammation and demyelination. The myelin sheath protects the nerves, however in individuals with MS, demyelination (loss of myelin) occurs leading to problems in communication between the CNS and the rest of the body. The common symptoms of MS include sensory problems such as tingling or numbness, visual loss or double vision, lack of coordination, motor problems or muscle weakness, difficulty with speech and fatigue.

The average age of diagnosis is between 20 and 40 years however, MS can be diagnosed in children and older adults. MS is much more common in certain parts of the world than others. For example, MS is more common in the northern most latitudes such as Canada, northern United States and northern Europe than it is in Africa and Asia. MS is also more common in women than men.

There are four subtypes of MS. Relapsing-remitting (RRMS) is the most common, where individuals have “attacks” or periods of symptoms followed by a period of no symptoms (i.e., remission). The second most common type, primary-progressive (PPMS), involves steadily worsening symptoms overtime without relapses. In PPMS, there may be plateaus where the symptoms are stable but no true period of remission. Secondary progressive MS (SPMS) involves a period of relapses and remissions followed by a period of progressive increase in disability. The least common type, progressing-relapsing MS (PRMS), involves distinct relapses but in between there is ongoing progression of disease rather than periods of remission.

Diagnosis/Testing

The diagnosis of MS is typically made using the criteria of dissemination in space and time. Dissemination in space means that there must be evidence of symptoms or a lesion occurring in more than one of the four characteristic locations in the CNS. Dissemination in time means that there must be evidence of new lesions occurring over time. Brain imaging (e.g., MRI) is often used to locate these lesions and a contrast agent called gadolinium may also be used to view areas of active inflammation in the CNS. Occasionally, a lumbar puncture (a procedure where a needle is put into the lower spine to collect a sample of cerebrospinal fluid (CSF) that surrounds the CNS) may be performed to look for markers in the CSF, which show signs of inflammation.

There are many factors that influence an individual’s risk of developing MS. While there is no single gene known to cause MS, certain genetic factors include the human lymphocyte antigens (HLA) gene, which are involved in the immune system. There are several different subtypes (or alleles) of the HLA gene and in certain populations, specific alleles have been associated with an increased susceptibility to develop MS. Environmental factors that are thought to be associated with MS include certain infections such as Ebstein-Barr virus (EBV) as well as vitamin D. When environmental factors occur in combination with specific genetic factors, this can lead to an individual being susceptible to develop MS.

Management/Surveillance

Management of MS often involves medication either to treat a relapse or to slow the progression of the disease. Steroids may be used during an attack as they can reduce the amount of acute inflammation. Other medications work by suppressing the immune system, which can decrease the inflammation that causes the attacks. There are also many other treatments that may be prescribed to address the specific symptoms associated with MS such as fatigue or pain.

Individuals with MS are generally followed by a neurologist but often have interdisciplinary teams involved in their care. A scoring system called the Expanded Disability Status Scale (EDSS) is often used to quantify and track the level of disability in individuals with MS.

Mode of inheritance

MS is a complex condition, which means that it is caused by a combination of many different factors. These factors can be genetic or non-genetic (such as environmental factors and lifestyle choices). Complex conditions are inherited in a multifactorial pattern. This means that the chance for an individual to develop MS is influenced by the number and type of genetic and non-genetic factors that occur together to which an individual is exposed. In other words, no single gene, and no single environmental factor cause MS. However, not all of these genetic factors and environmental factors are known.

Risk to family members

Although there is no single gene that causes MS, the risk to first-degree relatives of individuals with MS is higher than that of the general population. In general, for a sibling or child of an individual with MS, the risk is about 3%. This is above the general population risk of approximately 0.2%. However, the risks may be different depending on the specific family history and a genetic counselor may be able to provide more information.

Special considerations

None

Resources

National Multiple Sclerosis Society
http://www.nationalmssociety.org
Multiple Sclerosis Foundation
http://www.msfocus.org/
MS Society Canada
http://mssociety.ca
Genetics Home Reference: Multiple sclerosis
http://ghr.nlm.nih.gov/condition/multiple-sclerosis

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