A Rare Case of Familial Multiple Sclerosis

Rima R*

Department of Neurology, Army hospital Research and Referal, New Delhi, Delhi P.O. Box: 110010, India

Abstract

Multiple sclerosis (MS) is a chronic disease confined to the central nervous system (CNS). While the cause is not clear, proposed causes for this include genetics and environmental factors such as infections. Although most MS cases occur sporadically, a considerable proportion, as many as 20 percent, of the patients are related by family. Whereas familial aggregation of MS is a well-known phenomenon in high prevalence areas, it is considered very rare in Asia, a low prevalence area. We are describing a case of two sisters presenting with Multiple sclerosis, one with RRMS and other with SPMS.

Keywords: Multiple sclerosis; Chronic disease; Central nervous system; Genetics

Introduction

Multiple sclerosis (MS) is a chronic disease confined to the central nervous system (CNS). Its pathological hallmarks are neuroinflammation, de- and remyelination, neurodegeneration and astrogliosis. This neuroinflammation disrupts the neurological circuits, resulting in a varied signs and symptoms including physical, cognitive and psychiatric problems. MS occurs either in relapsing form or in progressive form.

The underlying mechanism is either destruction of neurons by the immune system or failure of the myelin-producing cells. The proposed mechanism for MS includes genetics and certain environmental factors. MS is diagnosed according to McDonald criterion 2010, which is based on signs, symptoms and neuroimaging. Most of the MS cases occur sporadically, but about 20 percent, of them can be familial [1,2].

The risk of familial MS had been reported to be increased from 12- to 38-fold in siblings, six- to 25-fold in children of MS patients [3] and from seven- to 26-fold in parents [4-9]. Familial aggregation of MS is commonly seen in highly prevalent areas [10] but is very rare in Asia, a area having a low prevalence [11]. However, the few studies that have addressed the familial risk of MS in low prevalence areas have been seriously limited by modest numbers of MS patients P [12].

Case Report

A 34 year old lady became symptomatic in 2008 with acute onset of paresthesias in bilateral lower limbs, with spontaneous recovery in one month. Thereafter she had multiple similar episodes over next 2 years. In february 2010 she had a similar episode of paresthesias in bilateral upper and lower limb which remained static for further two years and thereafter she had partial recovery of symptoms. In November 2010 she had insidious onset progressive gait and appendicular ataxia leading to difficulties in activities of daily living. She had one missed abortion and had one normal delivery two years back. She is a non smoker and does not use illicit or recreational drugs.

Neurologically she had grade 2 spasticity in both lower limbs, all DTI, were pathologically brisk, both planters were extensors, vibration and proprioception loss in both LL. She had clumsy finger nose, past pointing and knee shin test. She had intentional tremors in all limbs and atactic gait. Neuroimaging was suggestive of multiple discrete T2W and flair hyperintensities in pericallosal, justacortical, periventricular regions with no Gad enhanced lesions. Black holes were also seen in T1W images. CSF for oligoclonal bands was positive. Her autoimmure profile was negative.

Her elder sister presented at a age of 33 years with paresthesia in both hands and gait ataxia. Neuroimaging was suggesting of multiple discrete areas of signal attenuation in periventricular region, subcortical white matter, brainstem and spinal cord suggesting of demyelinating plaques. Diagnosed a case RRMS, started on prophylaxis initially on interferon. She had multiple relapses while on interferon thus subsequently started on fingolimod with no recurrences for last 2 years.

Discussion

Multiple sclerosis (MS) is an inflammatory autoimmune disorder of the central nervous system characterized by recurring neurologic symptoms MS typically presents in early adulthood and is considered the most common cause of neurologic disability in young adults [1]. In the contemporary history of multiple sclerosis (MS), the familial recurrence rate is estimated at about 15% and is variable in geographic locations or absent in some ethnic groups [13,14]. The earliest collection and report of familial cases were by Curtius in 1933 [15]. Pratt et al. in 1951 described the first incidence rate of 6.5% in a collection of 310 cases and proposed the concept of polygenetic inheritance [16]. Jersild et al. [17] was the first to report an association between the disease and certain alleles of the HLA complex thus providing evidence in support of the genetic nature of MS. Further the genome-wide association studies confirmed about 59 genetic variants associated with MS [18].

Our patient had disease onset at 28 yrs and her elder sister had MS at 33 yrs of age. It has been seen in some studies, that the onset of familial MS is in younger age groups than non-familial MS but this was not in our case. Familial MS has been found in younger ages than non-familial MS in other studies. A study on familial MS in Spain concluded that it affects individuals about 8 years earlier than non-familial MS (22 vs. 30 years old). Another large study based on PPMS patients found that familial MS starts sooner, than non familial MS with a median age of 37.6 vs. 42.7 years [19].

Some studies delineated that familial MS is more common among

*Corresponding author: Rumpaul Rima, Army hospital Research and Referal, New Delhi, Delhi P.O. Box: 110010, India, Tel: 01145151695; E-mail: rumpaulrima@gmail.com

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first and third degree relatives with MS. In other studies frequency of MS in first and second degree relatives of MS patients has been estimated 22.8%. A meta-analysis found that relative risk for MS is 9.2, 3.4 and 2.9 in first, second and third degree relatives of MS patients, respectively [20]. Our patients are product of nonconsanguous marriage and are first degree relatives.

Familial MS was more common among siblings, with sister-sister relationship having the highest rate. The lowest relation was among father-son and mother son. Mantomoli showed that the risk of MS among siblings is 4.7% which is 31 times higher than general population [21] Ebers [22] showed that the probability of having MS of MS among siblings is 4.7% which is 31 times higher than general among father-son and mother son. Mantomoli showed that the risk first degree relatives.

Our patients is a product of nonconsanguinous marriage, are sisters and the younger sibling had severer disease than the elder sister. Younger had SPMS and the elder one had RRMS. Symptomology is different in these two individuals. In ASIA the incidence of familial MS is lower.

Concordant siblings tend to share age of symptoms onset rather than year of onset, suggesting that inherited factors distinct from a common environmental exposure influence susceptibility. Studies of half-siblings and adoptees support the concept that genetic, and not environmental factors, are primarily responsible for familial aggregation [20,25] In this case both were brought up in India but elder sibling had migrated to Canada 8 years before the onset of the disease.

A strong genetic component in MS pathogenesis is indicated foremost by the relative high-recurrence risk in family members of affected individuals and frequent occurrence in some ethnic populations (particularly those of northern European origin) compared with others (African and Asian groups), irrespective of geographic location [26-28].

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