

Medical Update Memo

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Disease onset in familial and sporadic primary progressive multiple sclerosis

Summary

The pathophysiology of primary progressive multiple sclerosis (PPMS) involves diffuse axonal degeneration which is believed to start early in the disease process, even before the onset of clinical symptoms. Canadian research suggests a hereditary component to the disease process of PPMS. Koch M., Zhao M, Yee, I., Guimond C., Kingwell E., Rieckmann P., Sadovnick D., Tremlett H. and the UBC MS Clinic Neurologists. *Multiple Sclerosis* 2010 Apr 8. [Epub ahead of print]

Details

The pathophysiology of primary progressive multiple sclerosis (PPMS) involves diffuse axonal degeneration which is believed to start early in the disease process, even before the onset of clinical symptoms. Symptomatic onset then occurs when this process reaches a threshold after which the axonal loss can no longer be compensated. A preliminary study showed that patients with familial PPMS had an earlier clinical onset than patients with sporadic disease, suggesting a hereditary component to the disease process of PPMS.

In this study, data was combined from two large, population-based, longitudinal MS databases to investigate disease onset in familial and sporadic PPMS.

411 patients with PPMS were reviewed. There were no differences in gender distribution or onset symptoms between familial and sporadic PPMS. Patients with familial PPMS were significantly younger at disease onset (n=84, median age: 37.6 years) than patients with sporadic disease (n=327, median age: 42.7, p=0.007). This difference was due to a greater proportion of familial cases with a disease onset before the age of 30 and a smaller proportion with disease onset between 40 and 50 years of age (p=0.002). Gender

had no significant effect on the age at disease onset. Further analyses showed that these findings were unlikely due to ascertainment bias towards an earlier diagnosis in familial cases.

Conclusion: Findings suggest a hereditary component to the disease process of PPMS. It would be worthwhile to identify patients with familial PPMS for future research on disease modifying genes in MS.

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