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Progressive Myoclonus Ataxia (PMA), A Review on Some Important Notes

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Abstract

Progressive Myoclonus Ataxia or PMA which also was known as the Ramsay Hunt syndrome, is a disorder which shows itself with ataxia and myoclonus which are progressive in their nature. Seizures can be seen in the PMA but their occurrence is not very common. Cognitive dysfunction can also be seen in the PMA but such dysfunction is not very much significant. This is a short review of some important notes about the Progressive Myoclonus Ataxia disorder, its clinical picture, and relevant causes for the occurrence of this disorder.

Keywords: Progressive Myoclonus Ataxia; Ramsay Hunt Syndrome; Myoclonus; Ataxia; Seizure; Dystonia

Body

Progressive Myoclonus Ataxia or PMA which also was known as the Ramsay Hunt syndrome, is a disorder which shows itself with various signs and symptoms like ataxia, myoclonus, seizures, dystonia, cognitive impairment etc.

The common clinical picture of the PMA is a patient who is presenting with myoclonus and ataxia. The myoclonus may be multifocal, generalized, or segmental. Myoclonus can be triggered by sensory stimuli. It can be present at rest and be increased intentionally and with an action which is called Mimicking dysmetria. The myoclonus is originally cortical in most of the cases with PMA although focal reflex myoclonus may be originated from subcortical regions in some of the cases. In comparison with subcortical myoclonus, cortical myoclonus tends to have a shorter duration in its time [1,2].

Regarding the etiology, there are various causes for the occurrence of the PMA. Also the syndrome may be sporadic or inherited. Creutzfeldt-Jakob disease, Neuronal ceroid lipofuscinosis, Sialidosis, Mitochondrial disorders, Unverricht-lundborg disease, Progressive multifocal leukoencephalopathy, Dentatorubral-pallidoluysian atrophy or DRPLA, Spino-cerebellar ataxias or SCAs specifically Spino-cerebellar ataxia type 14 or SCA14 and Friedreich's ataxia are among the etiologies for the occurrence of the PMA.

There are some gene mutations which can lead to the occurrence of the PMA. Mutation in the MRE11 and GOSR2 are the examples of the gene mutations which can cause the PMA. MRE11 gene is also related to Ataxia-telangiectasia-like disorders. GOSR2 or Golgi Qb-SNARE protein can be mutated and cause the PMA which is presenting with myoclonus, ataxia, areflexia, seizures and chronic degeneration of the anterior horn cell. Progressive Myoclonus Ataxia which is related to the GOSR2 or GOSR2-related PMA is also known as the Progressive Myoclonic Epilepsy type 6 [3,4].

Celiac disease can also cause the PMA although the relationship between the PMA and Celiac disease is controversial. Gluten-free regimens can improve the neurological symptoms of the patients with PMA and concomitant Celiac disease. As a result in case of encountering with a patient with suspicious neurological symptoms for PMA, doing some workups for diagnosing Celiac disease like checking the levels of Antitissue transglutaminase and Antigliadin antibodies can be useful to find the cause of the PMA in the relevant patient.

Patients who are presenting with ataxia and myoclonus which are progressive in their nature should be more evaluated for possible diagnosis of the PMA. Paying attention to possible concomitant pathologies is of importance to diagnose the patients with the PMA [1,5].

Conclusion

a) It is important for the clinicians and researchers in the field, to have knowledge about the Progressive Myoclonus Ataxia, its clinical picture and etiologies to approach the affected patients with more precision at the bedside for the clinicians and to try to understand the pathophysiology of the Progressive Myoclonus Ataxia to study more about this disorder and relevant pathologies for the researchers.

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