BIOMARKERS FOR SEGAWA'S DISEASE

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Background/Objective: Segawa's disease is an autosomal dominant dopa-responsive dystonia (DRD) caused by partial defects in guanosine 5'-triphosphate cyclohydrolase I (GTPCH). We investigated the number and distribution of patients with Segawa's disease in Japan in 2009. In this study, in patients with Segawa's disease, we measured neopterin and biopterin levels and evaluated their use as diagnostic markers for Segawa's disease.
Methods: Between 2010 and 2011, new DRD patients suspected of having Segawa's disease were diagnosed by serum and cerebrospinal fluid (CSF) pteridine analyses, and the diagnosis was confirmed by GTPCH gene (GCH1) analysis. The data were analyzed statistically.
Results: Twenty-four and 25 patients were newly diagnosed to have Segawa's disease in 2010 and 2011, respectively. The plasma neopterin level was significantly lower in patients with Segawa's disease than in controls. However, no significant difference was observed in plasma biopterin level. Significant differences were also seen in CSF neopterin level and biopterin levels.
Conclusions: The plasma neopterin concentration plays a potential role in the diagnostic workup for Segawa's disease, as its decrease showed a statistically significant association with the disease. Although the prevalence of Segawa's disease in Japan, combining the results of our study in 2009 and in this study, was similar to that in the previous report by Nyggard et al. in 1993 (0.5-1.0 patients/million), plasma pteridine screening may reveal many more patients who potentially have Segawa's disease.