

NATIONWIDE EPIDEMIOLOGICAL STUDY OF PEDIATRIC NEUROTRANSMITTER DISEASE IN JAPAN

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Objectives: Pediatric neurotransmitter disease (PND) is a relatively new concept in medical science. PNDs, which are induced by genetic disorders that affect the regulation of neurotransmitters in children, include dopamine-/serotonin-related diseases and GABA-related diseases. Left untreated, PNDs can lead to severely compromised neurological function in patients. However, diagnosing these diseases has been difficult. Thus, it is assumed that many of those patients were not diagnosed accurately and they did not receive appropriate treatments. We investigated the numbers and the distributions of patients with these diseases in Japan.

Methods: We sent a questionnaire to pediatricians or neurologists of 1622 Japanese hospitals in 2009.

Results: We received replies from 60.3% of those hospitals. In dopamine- /serotonin- related diseases, 116 patients of Segawa disease (autosomal dominant guanosine triphosphate cyclohydrolase I (GTPCH) deficiency) were reported from 44 hospitals. Those patients were in every 10 region of Japan. The prevalence rate of Segawa disease was calculated as 0.96 patients/ million. Three patients of aromatic-L-amino acid decarboxylase (AADC) deficiency, another dopamine- /serotonin- related PND, were also reported from 2 hospitals. No patient with tyrosine hydroxylase (TH) deficiency or sepiapterin reductase (SR) deficiency was reported in this study. In GABA-related disease, 3 patients of succinic semialdehyde dehydrogenase (SSADH) deficiency were reported from 3 hospitals.

Conclusion: The prevalence rate of Segawa disease in this study was similar to that of a previous report by Nyggard et al. in 1993 (0.5-1.0 patients/ million). This is the first report of a nationwide epidemiological investigation of PND in Japan.