Study of Rett syndrome epidemiology and database in Japan

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Objective: In 2010, the recent revised criterion for diagnosis of RTT was published. We performed nationwide survey of RTT, according to the new criterion, to clarify the Japanese epidemiology of RTT.
Methods: We selected 1,020 hospitals or institutes by stratified sampling from 2,918 ones specialized for pediatrics and pediatric neurology in Japan to ask the number of RTT patients and clinical details from 2008 to 2009. We calculated prevalence of RTT patients, and reviewed individual symptoms with the new criterion. Moreover, we have established Japanese Rett Syndrome Database (J-RettDB) since 2013.
Results: We received 677 responses from 1,020 hospitals (total response rate: 66.4%), and they counted 480 definite RTT patients and 79 atypical patients. The estimated prevalence of RTT patients in Japan was 0.90 per 10,000 girls. Interestingly, over 80% of five main symptoms of the new criterion appeared in typical RTT patients until 3 year-old. We collected over 50 RTT patients on J-RettDB, and revealed several unique mutations of *MECP2*.
Conclusion: This is the first nationwide survey of RTT in Japan. It gave a prevalence of concomitant with previous studies in other countries. We have established the clinical and genetic database of Japanese RTT patients. Next, we have to use the database for clinical research to explore biological markers and treatment with international collaborations.