# 論 文 要 旨

## （Relationships between CAG repeat expansion length and disease progression history in patients with childhood-onset dentatorubral-pallidoluysian atrophy）

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Objective: To elucidate a relationship between CAG repeat expansion length and disease progression history in patients with childhood-onset dentatorubral-pallidoluysian atrophy (DRPLA). Methods: We retrospectively evaluated information from nine Japanese patients with disease onset reported as between 6 months and 12 years of age. In addition, we collected data of a total 13 patients from past literatures. Results: CAG repeat length in these patients ranged from 62 to 93. A strong correlation was confirmed for the age of disease onset, with the onset of epilepsy and involuntary movements, emergence of regression, and autonomic symptoms. The age at becoming wheelchair-bound and initiation of tube feeding also showed a significant correlation with CAG repeat length. In the previously reported cases, a significant correlation was confirmed for the age of disease onset, with the onset of epilepsy and emergence of regression. Conclusions: This is the article detailing this aspect of DRPLA focusing on the childhood-onset population. The first description of relationships between CAG repeat expansion length and natural history in patients with childhood-onset DRPLA was found in Maruyama’s report in 2012. Earlier disease milestones were revealed compared to the expected age based upon a previous report which contained data from the entire patient population, including adult-onset cases. These results provide a basis for predicting the outcome of patients in this particular age group, as well as for assessing the results of future clinical therapeutic trials.