

## Improving the accuracy of diagnosis for neurodegenerative disorders with artificial intelligence

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**Objective:** Diagnostic criteria for rare diseases are often revised with increased numbers of cases, because the pathogenesis of such diseases is complicated and heterogeneous. Therefore, nationwide surveillance is needed to clarify the pathogenesis of rare diseases. In this study, we applied AI (Artificial Intelligence) to diagnose neurodegenerative disorders, such as multiple system atrophy (MSA) and spinocerebellar degeneration (SCD).

**Methods:** We constructed an AI diagnostic system based on Chainer. After machine learning, diagnostic probability (0–1.0) was estimated for each case. Medical records of cases involving patients with MSA and SCD were provided from the Ministry of Health, Labour and Welfare. 4,949 cases involving patients with MSA and 7,073 cases involving patients with SCD between 2004 and 2008 were used for this study.

**Results:** Diagnostic probabilities of SND (Striata-Negra Degeneration) and OPCA (Olivo-Ponto-Cerebellar Atrophy) were estimated at 0.97 and 0.88. In contrast, the probability of SDS (Shay-Drager Syndrome) was lower than that of SND and OPCA. Diagnostic probabilities of sSCD, AD\_SCD, or SP (Spastic paraplegia) were estimated at 0.95, 0.86, and 0.83. On the other hand, the probabilities of AR\_SCD and Other\_SCD were estimated at 0.04 and 0.03.

**Conclusion:** An AI diagnostic system could correctly categorize cases with SND, OPCA, sSCD, AD\_SCD, and SP. Although cases involving patients with familial SCD required genetic testing, those with AD\_SCD were correctly estimated by the AI diagnostic system.