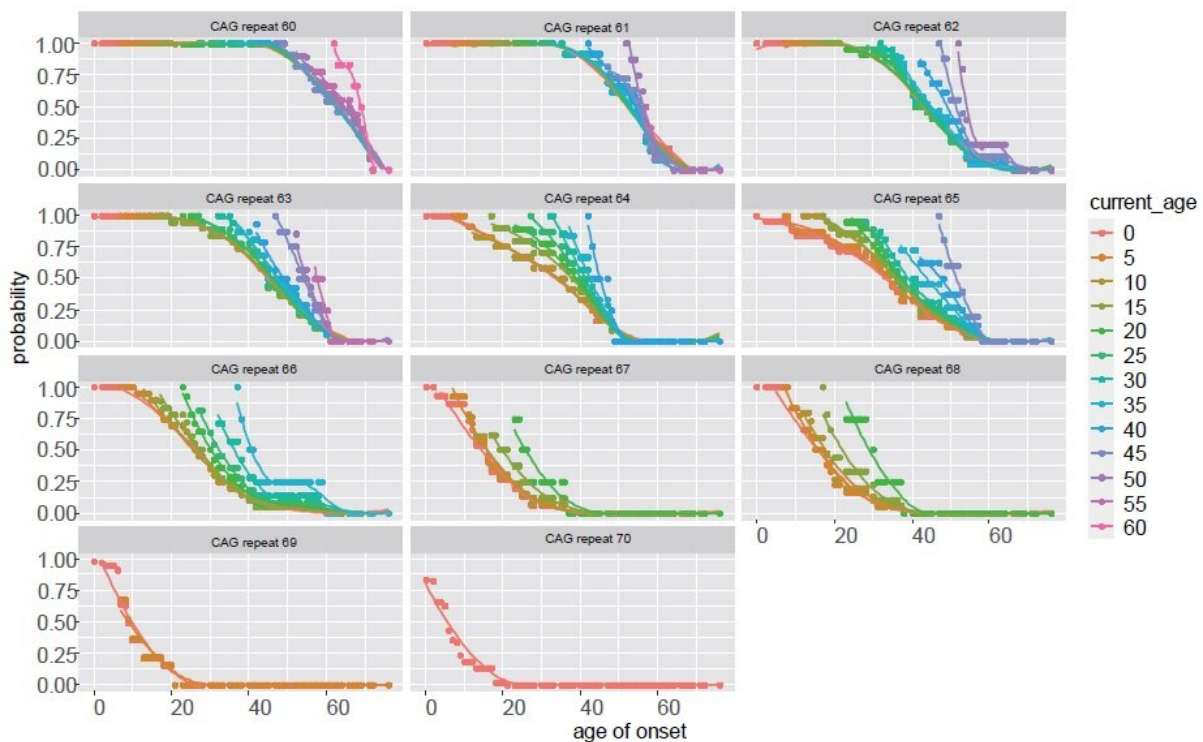


Researchers predict the age of onset of polyglutamine diseases with machine learning

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The image is under development.

Predicting the asymptomatic probability at a given age in DRPLA. Credit: Niigata University

Using machine learning, researchers at the Department of Neurology at

Niigata University have developed a model to predict the asymptomatic probability at each age from the current age and number of CAG repeats in carriers of spinocerebellar degeneration.

Polyglutamine diseases such as DRPLA and SCA3 are caused by an expansion of CAG repeats in the causative gene. In polyglutamine diseases, the number of CAG repeats is known to be inversely related to age of onset. Parametric survival analysis has traditionally been used to predict age of onset, but a more accurate prediction method has been desired.

The researchers used two [machine-learning](#) survival analyses to predict age of onset and compared their [accuracy](#) with six parametric survival analyses; the two machine-learning methods (Random Survival Forest and DeepSurv) showed higher prediction accuracy than parametric survival analyses. In particular, Random Survival Forests had the highest prediction accuracy and was used for the final prediction.

"This study is important for genetic counseling for career life planning. In the future, we will continue the analysis with more cases at several centers, aiming for more accurate prediction of the probability of onset of the [disease](#)," said Dr. Hatano and Dr. Ishihara.

The results of the study, "Machine Learning Approach for the Prediction of Age-Specific Probability of SCA3 and DRPLA by Survival Curve Analysis," were published in the online edition of the journal *Neurology Genetics*.

More information: Yuya Hatano et al, Machine Learning Approach for the Prediction of Age-Specific Probability of SCA3 and DRPLA by Survival Curve Analysis, *Neurology Genetics* (2023). [DOI: 10.1212/NXG.0000000000200075](#)

Provided by Niigata University

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