Artificial intelligence to save the day? How clever computers are helping us understand Huntington's disease.

Scientists at IBM and the CHDI Foundation have used artificial intelligence to analyse datasets from Huntington's disease observational trials to model progression of the disease. They hope their findings will help improve clinical trial design.

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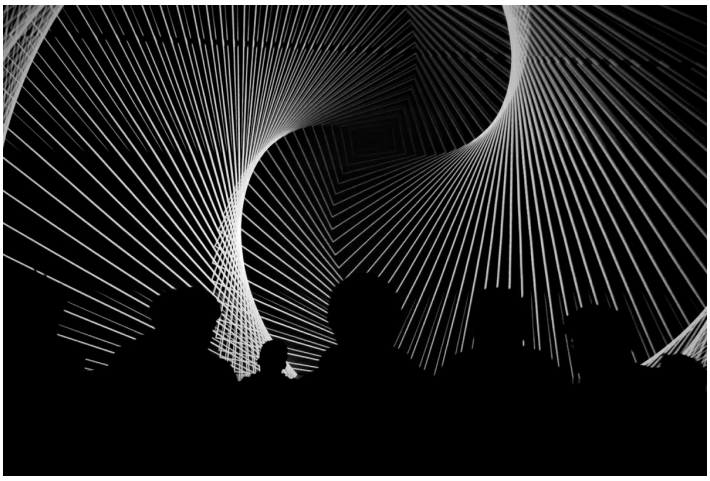
Scientists have developed a new model that maps out the different stages of Huntington's disease (HD) in detail. Using artificial intelligence approaches, the researchers were able to sift out information from large datasets gathered during observational trials contributed by Huntington's disease patients. A team of researchers from IBM and the CHDI Foundation have published a new model of HD progression in the journal Movement Disorders that they hope will improve how HD clinical trials are designed in the future.

Predicting the progression of HD symptoms is complicated

HD is caused by an expansion in the huntingtin gene which leads to the production of an expanded form of the huntingtin protein. Studies of lab models of HD as well as people carrying the HD gene, show that having the expanded gene and making the expanded form of the protein causes a cascade of problems. Starting with small molecular changes, people with HD will eventually end up experiencing a range of different symptoms related to thinking, movement and mood that get worse over time.



Scientists have been in the dark about how best to categorise the different phases with HD, but this new study using artificial intelligence hopes to shed some light on this problem.

Image credit: Ars Electronica / Robert Bauernhansl

Symptoms of HD typically start to show between the ages of 30 and 50, but a number of factors influence when this happens. We have known for a long time that people with bigger expansions in their huntingtin gene tend to get symptoms earlier, healthy lifestyle choices like a balanced diet and regular exercise can delay symptom onset, and other so-called genetic "modifiers" can also influence how early the disease might affect a gene carrier.

However, there's still a lot we don't understand about how Huntington's disease progresses over time and how the symptoms get worse. To try and tackle this problem, scientists from around the world have run numerous observational trials and natural history studies where patients' symptoms, biomarkers, and other measurements are monitored over time. These include PREDICT-HD, REGISTRY, TRACK-HD, and Enroll-HD. Together these studies have generated very large datasets which comprise more than 2000 different measurements recorded from 25,000 participants. This is tons of really helpful data, all made possible by the dedication of HD families to participating in these trials.

Machine learning helps us learn more about HD progression

Scrutinising all these datasets at once can help scientists spot new patterns and make novel conclusions but doing this type of analysis manually is extremely laborious and challenging. This is where the clever computer scientists come in! Scientists are able to use cool new methods to get the computers to look at all the data at the same time using special types of programs often referred to as artificial intelligence or AI.

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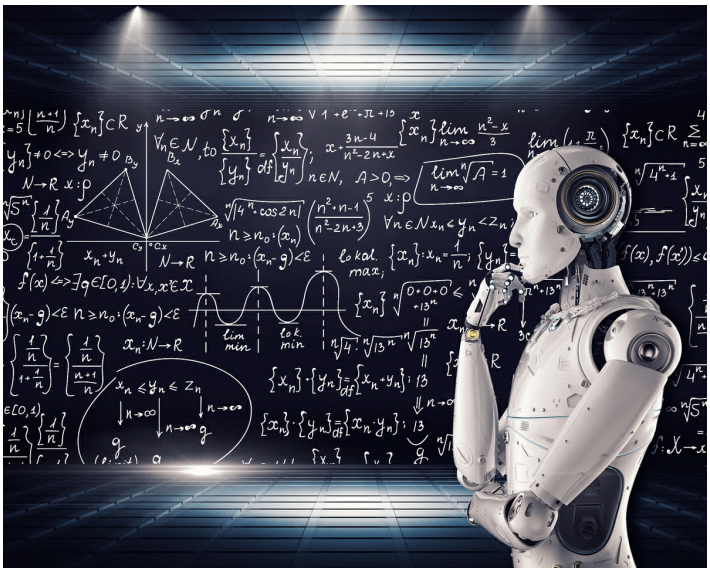
One commonly used AI approach is called machine learning. This type of AI software becomes better at making predictions of certain outcomes by building models from training data sets which it uses to “learn” without being explicitly programmed to do so. Machine learning is a field in its own right in biomedical research but also has lots of different applications for things like email filtering and speech recognition.

IBM and CHDI researchers used machine learning approaches to build and test a new model to understand how HD progresses and to categorise different disease stages. The model was then tested against a number of different measurements commonly collected and compiled in HD research that track disease progression, including the Unified Huntington’s Disease Rating Scale (UHDRS), total functional capacity (TFC), and the CAG-age product, also called the CAP score.

The new model defines 9 states of HD, all specified by different measurements that assess movement, thinking, and day-to-day function. These states span from the early stages of the disease before motor symptoms begin, all the way through to the late-disease stages that have the most severe symptoms. The model was able to predict how likely participants in the studies were to transition between states as well as how long participants spend in the different phases of HD. While other studies have determined that the entire disease course occurs over a period of about 40 years, this is the first time researchers have predicted the expected amount of time HD patients will spend in each of the 9 states that were described in the new model.

New models of HD progression will hopefully inform clinical trial design



Artificial intelligence is being used in lots of different ways to solve problems in areas such as medicine, business, communications, and transport.

Image credit: Image via www.vpnsrus.com

Having this handy new 9-state model of HD progression can help scientists and clinicians learn more about the different stages of HD and the timeframes it takes people with HD to move from one state to the next. With this information in hand, the researchers at IBM and CHDI believe this could help select the best-suited participants for particular HD clinical trials, identify robust biomarkers for monitoring how the disease progresses, and also help design better clinical trials.

This is an exciting step forward for HD research and we look forward to learning more about other AI applications in HD research as novel approaches are designed and this exciting field of science matures further.

The authors have no conflicts to declare. For more information about our disclosure policy see our FAQ...

GLOSSARY

Total Functional Capacity A standardized rating scale for function in HD, used to assess capacity to work, handle finances, perform domestic chores and self-care tasks

huntingtin protein The protein produced by the HD gene.

clinical trial Very carefully planned experiments designed to answer specific questions about how a drug affects human beings

observational A study in which measurements are made in human volunteers but no experimental drug or treatment is given

UHDRS A standardized neurological examination that aims to provide a uniform assessment of the clinical features of HD