Simple blood test could one day diagnose motor neurone disease

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Scientists at the University of Sussex have identified a potential pattern within blood which signals the presence of motor neuron disease; a discovery which could significantly improve diagnosis.

Currently, it can take up to a year for a patient to be diagnosed with amyotrophic lateral sclerosis (ALS), more commonly known as motor neuron disease (MND).

But after comparing blood samples from patients with ALS, those with other motor-related neurological diseases, and healthy patients, researchers were able to identify specific biomarkers which act as a diagnostic signature for the disease.

Researchers hope that their findings, published in the journal *Brain Communications*, and funded by the Motor Neurone Disease Association (MNDA), could lead to the development of a blood test which will identify the unique biomarker, significantly simplifying and speeding up diagnosis.

With patients living, on average, just 2-5 years after diagnosis, this time could be crucial.

Professor Majid Hafezparast, a professor of Molecular Neuroscience at the University of Sussex, led the research in collaboration with Professors Nigel Leigh and Sarah Newbury from the Brighton and Sussex Medical School, Martin Turner from the University of Oxford, Andrea Malaspina from Queen Mary, University of London, and Albert Ludolph from the University of Ulm.

He said: "In order to effectively diagnose and treat ALS, we are in urgent need of biomarkers as a tool for early diagnosis and for monitoring the efficacy of therapeutic interventions in clinical trials.

"Biomarkers can indicate the disease is present and help us to predict its progression rate.

"In our study, we compared serum samples taken from the blood of 245

patients and controls, analyzing their patterns of non-coding ribonucleic acids (ncRNA).

"We found a biomarker signature for motor neurone disease that is made up of a combination of seven ncRNAs. When these ncRNA are expressed in a particular pattern, we are able to classify whether our samples come from ALS patients or controls."

Dr. Greig Joilin, the research fellow who undertook this work in Professor Hafezparast's team said: "We hope that, with further work to validate these biomarkers, a blood test could be developed to help improve diagnosis of motor neuron disease.

"We are now looking to see whether they can predict prognosis to give patients and their families some insight as they begin to understand the disease. Our work could also help other scientists to measure the effectiveness of potential drug treatments against the ncRNA levels. Further, it provides new insight into the cellular and molecular events that contribute to the disease."

ALS is a group of conditions which affects the nerves in the brain and spinal cord leading to weakness in the muscles and rapid deterioration.

Doctors still don't know why this happens and there is currently no cure, although existing drug treatments can help patients with daily life and extend life expectancy—but only by two to four months on average.

Stephen Hawking is perhaps one of the most famous cases of motor neuron disease, but more recently Geoff Whaley and his wife Ann brought to light the troubling situation of patients in the UK who wish to end their life before the final phase of the disease takes hold.

Professor Hafezparast hopes that his team's discovery will improve the

outlook for patients by improving diagnosis and giving other researchers a valuable tool to test potential treatments. The researchers are now looking to validate this biomarker signature in a larger cohort of patients and begin to understand why these ncRNAs change in ALS patients.

More information: 'Identification of a potential non-coding RNA biomarker signature for amyotrophic lateral sclerosis' *Brain Communications*, 2020.

Provided by University of Sussex