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'Unknown' neurological disorder often incorrectly diagnosed

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The very serious hereditary disease HDLS was discovered in 1984 in Sweden. Many HDLS patients are still incorrectly diagnosed with Alzheimer's disease, MS or Parkinson's disease, but researchers at the Sahlgrenska Academy, University of Gothenburg, Sweden, have now developed a more certain diagnosis method - and are seeking to find a treatment for the "unknown" neurological disorder.

In 1984, Sahlgrenska Professor Oluf Andersen for the first time described a new, hereditary and very serious neurological disease that was given the name hereditary diffuse leukoencephalopathy with spheroids, usually abbreviated HDLS.

The disease has been perceived as very uncommon. However, a HDLS international consortium with headquarter at the Mayo Clinic in collaboration with the researcher Christina Sundal at the Sahlgrenska Academy, University of Gothenburg, and her research colleagues have now succeeded in identifying the genetic mutation, called CSF1R, that is believed to cause the disease.

The discovery, which is presented in a dissertation at the Sahlgrenska Academy, University of Gothenburg, has resulted in a new gene test that has led to more than 100 new cases of HDLS being confirmed in the U.S. and Japan in recent months.

In Sweden, HDLS has to-date been limited to one single family, which currently consists of 166 individuals of which 15 have been diagnosed

with HDLS. There are many unreported cases, and since the Swedish family was found negative for the CSF1R gene mutation Dr. Christina Sundal and her research team are still doing genetic testing to find additional gene mutation that are causative for the Swedish family. Results of this analysis will soon be published.

Since knowledge of the disease is limited among doctors, patients with HDLS are often incorrectly diagnosed with Alzheimer's disease, MS or atypical Parkinson's disease, and a study is now under way where 100 Swedish MS patients are undergoing genetic analysis to see if their disease is actually HDLS.

The basic Swedish research has been followed up internationally - most successfully by researchers at the Mayo Clinic in Florida, who have now gathered information and samples from HDLS families around the world. In 2011, Christina Sundal, a doctoral student at the Institute of Neuroscience and Physiology at the Sahlgrenska Academy, University of Gothenburg, was invited to the Mayo Clinic to conduct research on HDLS.

Christina Sundal is now presenting her dissertation, which shows that the symptoms and characteristic changes of HDLS can be distinguished on magnetic resonance images of the brain. Together with the discovery of the CSF1R gene mutation, this has revolutionized the possibilities of making the right diagnosis and developing future treatments.

"Our research has shown that HDLS is often incorrectly diagnosed. We hope that the disease will now be easier to identify, and that it will eventually be possible to use the CSF1R gene mutation to develop new medicines that can treat both HDLS and other similar neurodegenerative diseases," says Christina Sundal.

The 36-year-old researcher, raised in Bergen, Norway, is now being

given the responsibility to lead the continued HDLS research at the Sahlgrenska Academy, University of Gothenburg and Sahlgrenska University Hospital.

"I hope that our research will lead us to find a treatment in the future that can stop this disease, which is very devastating and strikes many families hard," says Christina Sundal.

Provided by University of Gothenburg