

A diagnostic test for ALS

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Researchers at VIB, KU Leuven, and UZ Leuven, in collaboration with researchers at the University of Jena, have demonstrated that measuring neurofilaments provides reliable confirmation of an ALS diagnosis. This diagnostic test represents a significant step forward because valuable time is still lost at present in diagnosing ALS. Diagnosis takes an average of one year from the first symptoms. The researchers hope that these tests will allow treatment to be started sooner.

ALS, a diagnosis feared by patients and doctors alike

Wednesday 21 June is World ALS Day. It is a day to stop and consider amyotrophic lateral sclerosis, or ALS for short. This neurodegenerative disease causes the death of motor neurons, the nerve cells controlling the muscles. The result is a loss of strength which spreads throughout the body. Besides the muscles in the limbs, it also affects the muscles used for swallowing, breathing, and speaking. The average survival rate after the onset of the first symptoms is just 2 to 5 years. ALS is relatively rare; about 400 people a year are diagnosed with the condition in Belgium.

As Prof. Philip Van Damme (VIB-KU Leuven, UZ Leuven) tells us: "Despite the severity of the disease, an ALS [diagnosis](#) relies heavily on the physician's clinical acuity. The typical disease progression of ALS, with the loss of strength extending from one body region to another, allows a definite diagnosis. In the early stages of the disease, diagnosis is difficult. Consequently the average time between the first symptoms and diagnosis is approximately one year. Better tests are needed for a faster

ALS diagnosis, which we hope to achieve with this [test](#)."

Neurofilaments can help to diagnose ALS

Neurofilaments are structural proteins in the cytoskeleton, which are present in high concentrations in motor neurons. It has been known for a long time that the lumbar fluid in ALS patients contains a higher concentration of neurofilaments, perhaps because they are released from sick [motor neurons](#). Researchers led by Prof. Koen Poesen (Laboratory of Medicine, UZ Leuven, and the Laboratory for Molecular Neurobiomarker Research, KU Leuven) and Prof. Philip Van Damme (Neurology UZ Leuven, and VIB-KU Leuven Center for Brain Research) have carried out detailed research into this phenomenon.

Prof. Koen Poesen (UZ Leuven, KU Leuven): "We have demonstrated that a certain type of neurofilament (pNfH, phosphorylated neurofilament heavy) in particular increases sharply in the lumbar fluid of ALS patients. This is even true when compared to patients presenting loss of strength symptoms due to other conditions (known as ALS mimics). The test meets all the requirements for use as a reliable diagnostic test. However, it requires an epidural because we can still only reliably measure the neurofilaments in the lumbar fluid."

The researchers have also demonstrated that there is a good correlation between the degree of neurofilament increase and the extent of the motor neuron loss. This indicates that the test reflects the underlying disease process. Whether the implementation of the test will also lead to a shorter time before diagnosis is currently still being researched.

More information: Koen Poesen et al. Neurofilament markers for ALS correlate with extent of upper and lower motor neuron disease, *Neurology* (2017). [DOI: 10.1212/WNL.0000000000004029](https://doi.org/10.1212/WNL.0000000000004029)

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