

Fruit fly steps in to fight human disease

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Belgian scientists have successfully introduced genes coding for a variant of the Charcot-Marie-Tooth (CMT) disease, into fruit flies. CMT is one of the most common hereditary disorders of the peripheral nervous system. VIB research -- directed by Albena Jordanova, Patrick Callaerts and Vincent Timmerman -- shows that the flies recapitulate several symptoms of the human disease.

"By putting mutant genes from human patients into fruit flies, we've created the first ever fly model for this kind of [neuromuscular disease](#)," says Albena Jordanova. "Now we have the opportunity to unravel the molecular mechanism behind Charcot-Marie-Tooth, as well as to start looking for substances with therapeutic value."

The breakthrough is the result of collaboration between VIB researchers working at the University of Antwerp and the Katholieke Universiteit Leuven, and appears in the [Proceedings of the National Academy of Sciences](#) (PNAS).

Charcot-Marie-Tooth is a hereditary disorder of the [peripheral nervous system](#) that affects 1 in 2,500 people worldwide. Patients suffer from progressive motor impairment, muscle wasting and weakness, sensory loss, and foot deformities. Affecting children and adults, the disease often starts with minor symptoms, gradually worsening over time. Presently CMT cannot be cured or prevented.

New chapter for an old gene

In previous research Albena Jordanova and Vincent Timmerman (VIB, University of Antwerp) discovered that CMT patients in families in Belgium, Bulgaria and the US showed three specific changes in one of the most ubiquitous genes in life: the YARS gene. YARS is responsible for the production of one of the oldest enzymes in the history of life (tyrosyl-tRNA synthetase), which is vital for the production of proteins. This was an entirely unexpected breakthrough. YARS had been considered a closed chapter in the biology textbooks. No one had suspected the relationship with specific variants of CMT until the revelation by Jordanova and her colleagues. These VIB findings open up an entirely new field of research.

Fruit flies with CMT symptoms

The VIB researchers at the University of Antwerp, in collaboration with Patrick Callaerts (VIB, K.U.Leuven) introduced four variants of the YARS gene into fruit flies. The normal variant, showed no difference in ordinary fruit flies. However, [fruit flies](#) with the mutant YARS genes, showed clear symptoms of CMT such as a reduced capacity to move, decreased functioning of the nerve cells and degeneration of the nerve endings.

Source: VIB (the Flanders Institute for Biotechnology)

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