

# Breakthrough in the treatment of inherited genetic disorder which damages muscle and nerve cells in the body

April 28 2016

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Scientists at the Universities of York and Leiden have made a significant breakthrough in the treatment of an inherited genetic disorder which damages muscle and nerve cells in the body.

Pompe disease is caused by a defective gene that results in a deficiency of an [enzyme](#) called acid alpha-glucosidase (GAA) which causes [progressive muscle weakness](#) in people of all ages.

The crippling disease came to wider public attention through the 2010 film Extraordinary Measures starring Harrison Ford.

But the new research, which included a team from Leiden University in the Netherlands, has led to the synthesis of fluorescent chemical 'probes' which can be applied for medical diagnosis.

These tools allow scientists to measure the level of the GAA enzyme in human cell extracts, allowing rapid detection of enzyme levels.

The researchers say the compounds will help inform more effective treatments, eventually allowing for better therapies and personalised medicine.

Similar diagnostic approaches have already improved medical treatment for other inherited conditions.

The research team included Dr Liang Wu and Professor Gideon Davies of the Structural Biology Laboratory, Department of Chemistry, at York.

Professor Davies said, "These tools allow scientists to measure levels of enzyme in healthy and sick people. Imaging active enzyme levels will help assess disease severity and also help inform more effective treatments."

Dr Wu added "It is an exciting breakthrough, made possible by the power of European collaboration. We believe that this technology will have widespread application beyond genetic disease and into cancer therapies."

The work, funded by the European Research Council, is reported in the journal *ACS Central Science*.

**More information:** Jianbing Jiang et al. Detection of Active Mammalian GH31  $\alpha$ -Glucosidases in Health and Disease Using In-Class, Broad-Spectrum Activity-Based Probes, *ACS Central Science* (2016).

[DOI: 10.1021/acscentsci.6b00057](https://doi.org/10.1021/acscentsci.6b00057)

Provided by University of York

Citation: Breakthrough in the treatment of inherited genetic disorder which damages muscle and nerve cells in the body (2016, April 28) retrieved 6 May 2024 from

<https://medicalxpress.com/news/2016-04-breakthrough-treatment-inherited-genetic-disorder.html>

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