MedicalXpress

Research team identifies new genetic syndrome

March 4 2014

Researchers at the National Institutes of Health (NIH) have identified a new genetic syndrome characterized by a constellation of health problems, including severe allergy, immune deficiency, autoimmunity and motor and neurocognitive impairment. The researchers, led by scientists at the NIH's National Institute of Allergy and Infectious Diseases (NIAID), observed that the syndrome's diverse symptoms are the result of mutations in a single gene associated with sugar metabolism. They plan to evaluate certain types of sugars as a potential treatment for people with this rare genetic condition in an upcoming clinical trial.

The study, published in the *Journal of Allergy and Clinical Immunology*, involved eight patients from two families. The families were originally referred to NIH because of severe eczema and recurrent skin and lung infections. By studying this group, the investigators found that the syndrome is caused by mutations in the *PGM3* gene that result in the production of underactive PGM3 protein. The NIH team showed that underactive PGM3 leads to lower levels of sugars that are essential for glycosylation, or the attachment of sugars to proteins. Glycosylation is necessary for the normal growth and function of all tissues and organs in the human body. The variety of symptoms in people with *PGM3* mutations likely reflects the production of abnormally glycosylated proteins throughout the body. In the laboratory, adding a certain type of sugar to cells from patients with *PGM3* mutations boosted cellular levels of the sugars necessary for glycosylation, thus suggesting a potential treatment.

The NIH scientists have yet to unravel how glycosylation defects influence the immune systems of people with *PGM3* mutations. However, their findings suggest that other, less severe defects in glycosylation may play a role in more common allergic and immunologic diseases, opening potential new avenues for developing treatments. The study was conducted by scientists from NIAID, the National Human Genome Research Institute and the National Institute of Neurological Disorders and Stroke, all components of NIH, and collaborators. Additional funding was provided by the NIH's National Institute of Diabetes and Digestive and Kidney Diseases, under grant number R01DK55615, and by The Rocket Fund.

More information: Y Zhang et al. Autosomal recessive PGM3 mutations link glycosylation defects to atopy, immune deficiency, autoimmunity, and neurocognitive impairment. *Journal of Allergy and Clinical Immunology* DOI: 10.1016/j.jaci.2014.02.013 (2014).

Provided by NIH/National Institute of Allergy and Infectious Diseases

Citation: Research team identifies new genetic syndrome (2014, March 4) retrieved 30 April 2023 from https://medicalxpress.com/news/2014-03-team-genetic-syndrome.html

This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.