New test for rare immunodeficiency

August 22 2017

Researchers at the University of Basel have developed a test to quickly and reliably diagnose a rare and severe immune defect, hepatic veno-occlusive disease with immunodeficiency. They reported on their findings in the *Journal of Clinical Immunology*.

Rare diseases are often only diagnosed very late, especially since in many cases, diagnostic tests are not available or are only available in a few laboratories. Hepatic Veno-occlusive disease with immunodeficiency is a clear example of this. It is an immunodeficiency caused by a mutation in the gene Sp110, and children suffering from it often die of severe infections or liver failure during the first year of life.

First described in 1976, this disease is still difficult to treat and is often fatal. It is also unclear what function the Sp110 gene actually has and why Sp110 protein deficiency leads to a severe immune defect and liver disease.

Rapid detection of missing Sp110 protein

One reason for the lack of progress in the treatment of this disease is that it had previously not been possible to detect the expression of the Sp110 protein in immune cells without significant effort. Mike Recher's research group from the University of Basel's Department of Biomedicine has now published a new, quick and easy-to-use test that shows the presence of the Sp110 protein in immune cells from the patient's blood.

The detection is based on flow cytometry, a method in which the properties of individual cells are measured using laser light. The researchers were able to use the test to reliably determine patients with different mutations in the Sp110 gene in a matter of hours.

"We expect that this test will soon be used in laboratories to diagnose a Sp110 deficiency," says Professor Recher. "The test will also help us quickly discover more about the biological function of Sp110."

More information: Florian A. Marquardsen et al. Detection of Sp110 by Flow Cytometry and Application to Screening Patients for Veno-occlusive Disease with Immunodeficiency, *Journal of Clinical Immunology* (2017). DOI: 10.1007/s10875-017-0431-5

Provided by University of Basel