MASARYK UNIVERSITY

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Researchers from MU contributed to the discovery of a new blood disease

A new type of blood disease was discovered by scientists from the MU Faculty of Medicine in cooperation with their colleagues from abroad. The discovery of this hereditary genetic mutation was triggered by a non-standard case of a female patient who after treatment of the acute lymphoblastic leukemia, inexplicably, continued to have a low number of blood platelets. Although she recovered from the disease, her children inherited this mutation and hence have low number of blood platelets as well. The scientists have confirmed that the reason is a gene mutation which significantly increases the likelihood of its bearer to experience an outbreak of leukemia. Information about this new discovery was published in the prestigious scientific journal Nature Genetics.

„Bearers of this disease do not usually experience any symptoms. The higher risk of leukemia is thus more likely to be discovered by chance when blood tests reveal lasting thrombocytopenia, that is a low level of blood platelets. People with this anomaly in their genes should then be more attentive to manifestation of seemingly ordinary diseases, because there is a higher risk of leukemia," said the dean of the Faculty of Medicine and the head of the Department of Internal Medicine, Hematology and Oncology of the Faculty Hospital Brno, Jiří Mayer.

He emphasized that his colleagues were able to discover this disease because they were looking for the causes of an unusual blood picture of this specific patient which did not comply with what had previously been known. „The patient was treated for acute leukemia and after the treatment she had lower levels of blood platelets. When she had children, it turned out that they also have a lower number of blood platelets. That lead us to look for genetic reasons of this condition. Eventually we discovered that the whole family has a certain mutation of the gene ETV6 which is responsible for haematopoiesis," describes the case Michael Doubek, a hematologist. Consequently, the doctors discovered that this gene mutation was one of the reasons that caused her outbreak of leukemia.

To confirm the genetic basis of this haematopoiesis disease took the researchers twelve years. Besides the scientists in Brno, laboratories in Italy and the USA also contributed to this discovery. Moreover, this newly discovered disease might be more widely spread than the researchers first assumed. They are currently studying genetic information of another family that experienced a similar problem. The tests are carried out in cooperation with laboratories of the science institute Ceitec MU.

Although it is not possible to cure the disease now, it is important for patients and doctors to know about the possible hereditary disposition. Moreover, there is a chance that scientists will be able to prevent the mutated gene from being passed onto the next generation.

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