Identification of genetic mutations involved in human blood diseases

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A study published today in *Nature Genetics* has revealed mutations that could have a major impact on the future diagnosis and treatment of many human diseases. Through an international collaboration, researchers at the Montreal Heart Institute (MHI) were able to identify a dozen mutations in the human genome that are involved in significant changes in complete blood counts and that explain the onset of sometimes severe biological disorders.

The number of red and white blood cells and platelets in the blood is an important clinical marker, as it helps doctors detect many hematological diseases and other diseases. Doctors can also monitor this marker to determine the effectiveness of therapy for certain pathologies.

"Complete blood counts are a complex human trait, as the number of cells in the blood is controlled by our environment and the combined expression of many genes in our DNA," explained Dr. Guillaume Lettre, a study co-author, an MHI researcher, and an Associate Professor at the Faculty of Medicine at Université de Montréal.

In collaboration with their colleagues at the University of Washington in Seattle and the University of Greifswald in Germany, these MHI researchers analyzed the DNA of 6,796 people who donated specimens to the MHI Biobank by looking specifically at segments of DNA directly involved in protein function in the body. They specifically identified a significant mutation in the gene that encodes erythropoietin, a hormone that controls the production of red blood cells. "Subjects who carry this

mutation in their DNA have reduced hemoglobin levels and a 70% greater chance of developing anemia," explained Dr. Lettre. The scientists also identified a mutation in the JAK2 gene, which is responsible for a 50% increase in platelet counts and, in certain cases, for the onset of bone marrow diseases that can lead to leukemia. Dr. Jean-Claude Tardif, Director of the MHI Research Centre, Full Professor at the Faculty of Medicine at Université de Montréal, and a study co-author, added that "after reviewing pre-existing clinical data from the MHI Biobank, we observed that these donors also had a higher risk of having a stroke during their lifetime."

Dr. Lettre believes that these findings are very encouraging, as they suggest that the experimental approach used in the study can be applied to other human diseases. "Thanks to the existing genetic data and wealth of other clinical information available from the MHI Biobank, we will be able to identify other rare genetic variations that may impact the risk of cardiovascular disease and open the door to the development of new therapies."

Provided by Montreal Heart Institute

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