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Genetic abnormality may increase risk of blood disorders

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Researchers at Memorial Sloan-Kettering Cancer Center (MSKCC) have shown for the first time that a tendency to develop some blood disorders may be inherited. Their research, published online today in *Nature Genetics*, identifies a common genetic sequence abnormality that enhances the likelihood of acquiring a mutation in a gene linked to certain blood diseases.

The investigators carried out a genome-wide study to identify inherited DNA sequence changes that frequently occur in patients with myeloproliferative neoplasms, in which several types of blood cells are excessively produced in the bone marrow. They found that an inherited alteration in the gene for JAK2 - a protein with enzymatic activity that is linked to the abnormal production of blood cells - is more common in patients with these disorders. Importantly, patients who inherited this JAK2 alteration were predisposed to acquiring another JAK2 mutation on the same DNA strand. According to the research, these mutations do not arise randomly, but are specifically determined by the DNA sequence.

More than half of patients afflicted with myeloproliferative neoplasms - which affect an estimated 140,000 people in the US - carry the JAK2 mutation and suffer from the overproduction of red blood cells, platelets, or fibrous connective tissue. According to the authors, understanding the underlying inherited sequence partly explains the predisposition for acquiring mutations in certain disease-specific genes and may help explain why some individuals are at higher risk in

developing the disease.

Source: Memorial Sloan-Kettering Cancer Center

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