Novel inherited variants may raise risk for Hodgkin lymphoma

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Novel coding and noncoding gene variants have been identified that may

increase the risk for developing Hodgkin lymphoma (HL), according to a study published online Aug. 20 in *Blood*.

Jamie E. Flerlage, M.D., from St. Jude Research Hospital in Memphis, Tennessee, and colleagues performed whole-genome sequencing on 234 individuals with and without HL from 36 pedigrees with two or more first-degree relatives with HL. To identify coding and noncoding variants, a family-based segregation analysis was performed.

The researchers identified 44 HL risk variants in 28 pedigrees, of which 33 and 11 were coding and noncoding, respectively. The top four recurrent risk variants were a coding variant in *KDR*, a 5'UTR in *KLHDC8B*, and noncoding variants in an intron of *PAX5* and in an intron of *GATA3*. For one pedigree, a newly identified splice variant in *KDR* and high confidence stopgain variants affecting *IRF7* and *EEF2KMT* were seen. In three independent pedigrees, multiple truncating variants in *POLR1E* were observed.

"We married several existing tools as well as customized approaches to make a pipeline that could process data from these families in a meaningful way," a coauthor said in a statement. "The work that went into making this pipeline is not specific to Hodgkin lymphoma; the pipeline can be used for any other disease where families are involved."

More information: Abstract/Full Text (subscription or payment may be required)