BLOOD ADVANCES TALK

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Inherited predisposition to myeloid malignancies

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Abstract

Germline predisposition to myeloid malignancies is increasingly recognized, and numerous susceptibility syndromes are now well described. Both inherited and de novo germline mutations can be seen, and mutations range from point mutations to deletions and other structural chromosomal rearrangements. Because easily obtained tissues such as saliva, blood, and bone marrow are all potentially affected by the disease, care must be given to testing true germline tissue. Thus, comprehensive testing is complicated, both by the necessity of covering all mutations types as well as doing so using DNA derived from germline material. Identification of germline mutations has important implications for the treatment of the index patient as well as for family members who may share an inherited variant. Efforts by the American Society of Hematology and ClinGen to provide standardized functional annotation of variants will hopefully provide consistency in variant interpretation. As the recognition of these syndromes increases and the cost of DNA sequencing falls, additional predisposition syndromes are likely to be identified.

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