Clinical Applicability of Proposed Algorithm for Identifying Individuals at Risk for Hereditary Hematologic Malignancies

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Over the past decade, more than 12 genes have been identified to cause hereditary predispositions to hematologic malignancies. These syndromes are characterized by an increased risk to develop myelodysplastic syndrome (MDS), acute myeloid leukemia (AML), or aplastic anemia (AA) at young ages, with various phenotypic features including peripheral cytopenias, immune dysfunction and skeletal defects. In 2013, Churpek et al. proposed a referral algorithm which consists of certain criteria for identifying leukemia patients who may benefit from genetic assessment for these hereditary syndromes. These criteria assess personal history of cytopenias, skin or nail abnormalities, immune deficiencies/atypical infections, and other associated clinical characteristics. The algorithm also assesses family history of leukemia and personal/family history of other malignancies.

Our study aimed to assess the applicability of these criteria on an unselected population of adults with leukemia by retrospective chart review at The University of Texas M.D. Anderson Cancer Center. These patients presented for initial consultation from March 1, 2014 to December 31, 2014. Six-hundred and eight individuals diagnosed with MDS/AML/AA were included in this study. Key demographic information was obtained from a clinical database maintained by the Department of Leukemia. The median age at diagnosis was 67 years, 387 (64%) were male, and at the time of data collection, 315 (51.8%) individuals were alive. Of the 608 individuals in this study, 334 (54.9%) were diagnosed with AML, 199 (32.7%) with MDS, 59 (9.7%) with MDS/MPD, and 16 (2.6%) with AA.

Regarding clinical/medical record documentation of referral criteria, three hundred and sixty-four (59.9%) individuals reported at least one first or second-degree relative with cancer. Thirty-one (5.1%) individuals reported a family history of leukemia, which was also the most consistently reported criteria in the medical record (n=580, 95.4%). Overall, 406 individuals (66.8%) had insufficient documentation to determine whether any criteria were met. Two hundred and two (33.2%) individuals met at least one of the proposed criteria for genetic counseling referral; however, only nine received a referral (4.5%) to genetic counseling.
Increased documentation of the presence or absence of phenotypic features associated with these hereditary syndromes is necessary to better assess the applicability of these criteria, and to ensure that individuals receive appropriate referral for cancer genetics risk assessment.

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