

Development of a Pediatric Oncology Screening Tool to Identify Individuals Appropriate for Genetics Referral

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Background. Current medical literature highlights the importance of detecting childhood cancers due to a hereditary syndrome, suggesting that the identification of a predisposing genetic condition will improve survival and treatment outcomes for the patient and potentially offer family members with an accurate risk assessment. Presently, there are a number of published guidelines that outline which adult cancer patients and survivors should be referred for hereditary cancer risk assessment; however, there is limited guidance in the pediatric oncology setting.

Procedure. The goal of this study was to develop a simple, practical, and efficient clinical tool to aid pediatric oncology providers in the determination of whether a child diagnosed with cancer should be referred for genetic evaluation. Tool development involved systematic literature search of hereditary cancer syndromes as well as expert review. The tool was retrospectively applied to a sub-set of patient records from the oncology and genetics clinics in the Department of Pediatrics at the University of Massachusetts Memorial Center. To assess whether a genetics referral as identified by the tool would agree with recommendations of a practicing genetic counselor, review of the records by an expert in the field was separately performed.

Results. The sample consisted of 131 eligible patient records, 7 of which had an identified genetics referral. Overall, application of the screening tool identified 23 (17.5%) total records that were eligible for genetics referral, 4 (57%) of which were among the 7 records with a known genetics referral. Expert review of the records identified 38 total records (29%) for genetics referral. Of these, the tool agreed with 22 (57.8%) for genetics referral. Expert review identified the same 4 of the 7 (57%) known genetics referrals as the tool. Kappa analysis revealed good agreement between the expert and the tool ($k=0.66$).

Conclusion. This project lends evidence that a pediatric oncology screening tool at the time of diagnosis is relevant. A missed opportunity for a genetics referral may lead to incomplete risk assessment and recommendations for short and long-term management.