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Validity of a Family History Form as a Genetic Screening Tool

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Background: In this era of advanced molecular genetics, genetic screening is becoming a vital component of medicine. Primary physicians need screening tools that will allow them to take advantage of this genetic revolution. Our primary objective was to validate a locally developed family history form (FHF) as a genetic screening tool for primary physicians. If successful, the FHF was to be implemented by our HMO for use by primary physicians.

Methods: The FHF was designed based on contributions from physicians, biostatisticians, and FHFs used by other institutions. The design was a bubble form with a list of approximately 50 genetic disorders. Twenty patients, seen in the genetic clinic, consented to participate in this study. Participants were asked to fill out the FHF at the beginning of their appointment. A genetic counselor then obtained a 3-generation pedigree from each patient. The FHF was considered valid if it agreed with at least 90% of the family history information gathered by the pedigree.

Results: Of the twenty FHFs completed, four matched at least 90% of the pedigree information ($p>0.9$). The overall percentage of agreement for all diseases and for all patients was 56.4%. The information that was captured by the pedigree and not by the FHF included ? siblings, 3rd degree relatives, carrier status and ages at diagnosis. In fourteen FHFs, the form picked up some family information that was not obtained by the pedigree. This unexpected finding is most likely due to the indication for their genetics appointment influencing the pedigree interview.

Conclusion: We do not recommend the use of a bubble FHF design as a genetic screening tool, and we plan to test different designs including fill-in-the-blank.