

The Three-Generation Pedigree: A Critical Tool in Cancer Genetics Care

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Pedigree construction is an important component of cancer risk assessment and comprehensive genetic care. Pedigrees must be updated and re-evaluated on a regular basis. Complete pedigrees are needed to select genetic tests and interpret genetic testing results accurately, as well as to enroll patients and families in research and variant reclassification studies to advance the science of genetics. Identified barriers to pedigree construction and assessment are described with implications for nursing practice.

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The family history, a rather low-tech tool, is the backbone of genetic assessment and guides risk assessment and genetic testing decisions. The importance of the pedigree and its application to genetic practice is often overlooked and underestimated. Unfortunately, particularly with electronic health records, standard pedigrees are not routinely constructed. A clear understanding of how pedigrees are employed in clinical oncology practice may lead to improved collection and use of family history data.

Standardized Pedigree Nomenclature

Family histories of disease have been recorded in standardized formats for centuries (Resta, 1993). In ancient Greece, the physician Hippocrates assembled family histories to understand disease prognoses (Hinton, 2008). In 1995, standardized criteria for the graphic presentation of data were published by the National Society of Genetic Counselors (NSGC) (Bennett, French, Resta, & Doyle, 2008). Pedigrees can be constructed with pencil and paper and scanned into the record. More commonly, a pedigree software program is used to construct the pedigree, making the process of updating it as more pertinent information becomes available an easier task.

American Society of Clinical Oncology and NSGC guidelines emphasize that the family history should be obtained and evaluated at the initial visit and should be reevaluated at least annually (Berliner, Fay, Cummings, Burnett, & Tillmanns, 2013; Lu et al., 2014). A pedigree should include three generations, ancestry from the maternal and paternal sides, current age and age at death, and, in oncology, information about malignancies and age of diagnosis.

Clinical Applications

Like many aspects of health care, collecting an accurate family history is a science and an art (Venne & Scheuner, 2015). Obtaining a family history provides a unique opportunity to connect with the patient on a different level and learn more about family dynamics and social norms, providing insight as to how the family may manage information gleaned from risk assessment and genetic testing. If patients know prior to their appointment what information is required from them, they can then provide a more accurate family history and will have greater confidence in their risk assessment and screening recommendations (Armel et al., 2009). This also communicates the importance of the family history in clinical decision making. Despite these benefits, in addition