

Exploring Family Relationships in Cancer Risk Counseling Using the Genogram¹

Mary Daly,² Jennifer Farmer, Christine Harrop-Stein, Susan Montgomery, Mark Itzen, Josephine Wagner Costalas, Andre Rogatko, Suzanne Miller, Andrew Balshem, and Doris Gillespie

Division of Population Science, Family Risk Assessment Program, Fox Chase Cancer Center, Cheltenham, Pennsylvania 19012 [M. D., C. H. S., S. M., M. I., J. W. C., A. R., A. B., D. G.], and the Division of Medical Genetics, University of Pennsylvania Health System, Philadelphia, Pennsylvania 19104 [J. F.]

Abstract

Objectives. The genogram is a tool that has facilitated counseling in family therapy and social work for many years. It is hypothesized that genograms may also be useful in genetic counseling, because they help the counselor to acquire more objective and consistent information from the client, as well as to incorporate family dynamics and psychosocial issues into the counseling approach.

Materials and Methods. A pilot study of genograms used as an adjunct to genetic counseling was performed at Fox Chase Cancer Center's Family Risk Assessment Program. A questionnaire was developed to elicit genograms from 38 women at risk for familial breast and/or ovarian cancer. After standard pedigree expansion, a series of questions was asked about the consultand's relationship with other family members, communication patterns within the family, attitudes toward genetic testing, family reactions to cancer, roles individuals play in the family, and significant historical or anniversary events. Relationships were defined by the consultand as close, very close, conflictual, fused and conflictual, distant, or estranged.

Results. The majority of relationship types reported by 38 individuals was "very close" or "close." Eighty-one % reported having close/very close relationships with their spouses, 83% reported close/very close relationships with their mothers, and 70% reported close/very close relationships with their fathers. The degree of familial cohesion as depicted by the genogram correlates positively with scores obtained on the standardized Social Adjustment Scale Self-Report ($P = 0.01$).

Conclusions. Given the family-wide implications of genetic testing, the genogram may offer important guidance in family-targeted interventions.

Introduction

As the HGP³ nears completion of its goal of mapping the entire human genome, more and more individuals and their families

will qualify for presymptomatic DNA testing of adult-onset genetic diseases. The cancer genetic counseling literature has reported that receiving predictive genetic testing results affects family dynamics, as well as individual psychosocial functioning (1, 2). Because the information provided in a genetic counseling session is likely to have important implications for family members, genetic counseling needs to focus on not just the individual client but also the family system within which the individual operates. Eunpu (3) proposes the use of the genogram, a multigenerational family tree that plots biological relationships and graphically records information about social relationships and psychosocial issues in the family, to provide genetic counselors with a tool to help families adjust to a genetic diagnosis and enhance decision making.

This study reports the use of the genogram as an adjunct to genetic counseling sessions for women who are at increased risk for breast and/or ovarian cancer and are interested in pursuing genetic testing. Additionally, this pilot attempts to validate information depicted on the genogram by having individuals take an independent survey, the SASSR.

Materials and Methods

FRAP. The FRAP was designed for women who have a family history of breast and/or ovarian cancer. The FRAP is a multidisciplinary team, consisting of oncologists, genetic counselors, a social worker, nurses, health educators, and a physician assistant. Eligible participants complete a series of questionnaires, detailing their medical, family, reproductive, and exposure history before attending two 1-h education sessions. In the first session, breast and ovarian anatomy, risk factors associated with breast and ovarian cancer, and the benefits and limitations of screening for these cancers are reviewed and discussed. This session is facilitated by a health educator or nurse. In the second education session, the genetic counselor reviews basic information on the genetics of familial cancers, the different patterns of inheritance of genes within a family, and the benefits, risks, and limitations of *BRCA1* and *BRCA2* genetic testing. At an individual follow-up visit, the consultand meets with the cancer risk counselor to expand and review the family history. For consultands identified as having a possible hereditary family history of breast and/or ovarian cancers, participation in the program's genetic testing research protocol is offered. However, before any decisions about genetic testing are made, all women in the FRAP program are provided with estimates of their risks for breast and ovarian cancer, based on their personal and family history. The Gail model (4) is used to estimate breast cancer risk, whereas data from published epidemiological studies are used to estimate ovarian cancer risk. These estimates are then revised for any woman undergoing genetic testing, based on her test results.

The informed consent process for genetic testing includes an in-depth discussion of inheritance of mutated *BRCA1* and *BRCA2* genes, the spectrum of cancers identified in families carrying alterations in these genes, and the potential risks, benefits, and implications for receiving results from genetic

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² To whom requests for reprints should be addressed, at Fox Chase Cancer Center, Division of Population Science, 510 Township Line Road, Cheltenham, PA 19012.

³ The abbreviations used are: HGP, Human Genome Project; FRAP, Family Risk Assessment Program; SASSR, Social Adjustment Scale Self-Report.

testing. When test results are available, the consultand is invited to meet with a team consisting of a medical oncologist, genetic counselor, and social worker to discuss the test results. The medical oncologist reviews the types of cancers for which the consultand may be at risk, based on the genetic test results and the consultand's medical and family history. Appropriate screening and prevention guidelines are also reviewed. The social worker helps explore, with the consultand, unexpected reactions to the results and reminds her of support resources identified at the prediscovery session. The genetic counselor reviews the benefits and limitations of the results, the inheritance of the gene mutation in the family (if a mutation had been identified in the family), and identifies at-risk relatives. All participants, regardless of genetic testing status, are encouraged to share their cancer risk information with their family. In a follow-up survey conducted after the receipt of genetic risk information, we have found that >75% of women share the information with their family members, but many report considerable emotional difficulty in doing so (5). To date, however, there has been no standardized tool to help counselors address family issues critical to the communication of genetic information and to assist the sharing of information within the family.

Study Participation. Thirty-eight women who are participants in the FRAP enrolled in the study, "Exploring the Use of the Genogram as an Aid in the Genetic Counseling Process." These women had completed participation in the HGP research at Fox Chase Cancer Center, "Coping with Genetic Risk for Breast and Ovarian Cancer." The goal of the HGP Coping Study was to assess how women cope with their risk of breast and ovarian cancer. This supplemental research expanded upon the Coping research by looking in more depth at the impact of the genetic counseling process on the consultand and her family. The nature and purpose of the genogram study were explained to every participant, and voluntary consent was obtained. The genogram session was conducted as an interview in a small meeting room just outside of the clinic area after the individual cancer risk counseling session. All sessions lasted between 30 and 60 min. A total of 38 genograms were constructed using a standardized questionnaire in interview style.

Questionnaire Development. McGoldrick and Gerson (6) standardized genogram construction by drawing lines between individuals to represent the nature of their relationships (Fig. 1). The conceptual framework of genogram construction and interpretation is rooted in Murray Bowen's Family Systems Theory, which describes the association between family structure and family functioning and identifies the characteristics of family relationships that foster or erode support systems (6). According to this perspective, behavior patterns within families are often stable and transmissible over time. Therefore, the genogram is constructed to examine patterns of function and relationships from one generation to the next. Reactions to a family crisis, such as a diagnosis of cancer, are of particular importance in defining the strength and nature of relationships (6).

The questionnaire developed for this pilot was designed to elicit information for genogram construction. It was intended to be administered in an interview format that contained forced choice as well as open-ended items. The questionnaire was initially developed with guidance from Eunpu, published suggestions from McGoldrick and Gerson (6) and Rolland (7), as well as members of the FRAP team who identified the common content areas that typically emerge in the course of cancer risk counseling. In our previous research (8, 9), for example, we have found that the motivation to seek cancer risk counseling is a function of beliefs about personal risk, perceived coping skills, and concerns about

children. Thus, the questionnaire included items, based on Family Systems Theory that probed health beliefs, coping strategies, family communication patterns, and affective climate. Pretest of the questionnaire was administered to 14 women to assess clarity, language, content, and feasibility. On the basis of their feedback, additional questions dealing with roles within the family and the family's experience with cancer were added.

Genogram Construction. A genogram uses symbols, like those in a pedigree, to represent individuals and biological relationships in a family. The template for the genogram is the computer-generated pedigree illustrating at least three generations of the consultand's family. This pedigree forms a framework for adding family information including demographics, critical family events, and relationships between family members.

The genogram interview was conducted with 38 participants by either a genetic counseling student ($n = 18$), nurse ($n = 2$), a master's prepared social worker ($n = 11$), or health educator ($n = 7$).

The consultand was first asked to define her relationships with specified individuals as close, very close, conflictual, distant, estranged or cut off, and/or fused and conflictual. Definitions of these relationship types were explained to the consultand and based upon the response, a particular pattern of lines was drawn between her and other family members (Fig. 1). In an attempt to quantify and further define the reported relationship, the consultand was asked about the frequency and nature of communication between individuals and whether information was censored during communication. For example, two family members who constantly argue and cannot seem to agree on most issues would be defined as having a conflictual relationship. The consultand watched the interviewer draw lines between individuals on her family pedigree based on the relationship type reported by the consultand.

At the end of each genogram session, the consultand was asked to review the genogram to be certain that the relationships depicted reflected the information given. The consultand was permitted to amend the genogram, if desired.

SASSR. After completion of the genogram session, participants completed the SASSR, which is an instrument for measuring social adjustment by patient self-report. The SASSR is a modification of the Structured and Scaled Interview to Assess Maladjustment developed by Gurland *et al.* (10, 11). The self-report scale is simple to execute and requires no trained interviewer for administration.

Weissman and Bothwell (12) validated the use of the SASSR as an effective means of assessing social adjustment. With the use of the Pearson r correlation, a comparison of means between the interview scale and the self-report scale was completed. Correlations between the two were significant, ranging from 0.40 for the family unit subscale to 0.76 for the marital role subscale. The correlation for overall adjustment was 0.72, indicating excellent agreement (12).

The SASSR contains 42 forced-choice questions that measure either instrumental or expressive role performance in six major areas of functioning: (a) role performance; (b) satisfaction with work; (c) social activities; (d) leisure activities; (e) relationships with extended family; and (f) family role as spouse, parent, or member of a family unit (12). The SASSR takes 15–20 min to complete, depending on reading ability and comprehension. For our purposes, scores on the social and leisure activities, extended family, and family roles subscales were totaled for a composite SASSR score.

Database. A database of genogram relationships and SASSR scores was created on a Microsoft Excel spreadsheet. Each type

of relationship reported by the consultand in reference to the consultand and another family member was entered. Frequencies of relationship types were calculated from the database. Linear regression and correlation analyses were used to model the dependency of the response variable SASSR with the predictor %C/VC (percent close/very close). Other categories, such as fused and conflictual, conflictual, distant, and estranged were eliminated from this analysis because the numbers of these types of relationships were very small.

Results

Study Population. Thirty-eight genograms were obtained from 34 unique families. There were four sister pairs that were interviewed independently. Two women were interviewed on separate occasions by different counselors, and independent genograms were constructed. For these two participants, the questionnaire responses as well as the genogram relationships were almost identical at each interview.

Table 1 compares the genogram subset with the larger study population enrolled in the HGP Coping Research. Participants shared similar demographics including age, educational level, and marital status. Breast/ovarian cancer risk was equivalent between the two groups based on the mean Gail Model Scores and family history information. These data illustrate that the genogram study population mirrors the larger at-risk population that presently participates in the Fox Chase Cancer Center FRAP. Of the 38 participants in this pilot study, two have received genetic testing results. One participant received results in the year after the study, whereas the other received results about 4 months before the genogram session.

Genogram Construction. Sample genograms appear in Figs. 2–4. Fig. 2 illustrates a family whose pattern of functioning and communication is conflictual and in some cases estranged and has been repeated in successive generations. The consultand said her mother should not receive genetic testing results and if she did, she should not receive the results with her daughters present. The consultand in Fig. 3 reported receiving messages of denial from individuals diagnosed with cancer. “Everyone said that everything will be fine.” She felt that family members took the diagnoses lighter than they should. The consultand had been the caregiver to her mother, father, and spouse. Note the repetition of troubled relationships in three generations. Sadness was difficult for the consultand to express in her family, but she had less trouble with humor and sarcasm. The consultand in Fig. 4 felt that she would be the next in her family to get cancer. She reported that everyone tended to be supportive. However, she received the most support from her husband and sister. Although she described her family as close, they seemed to have great difficulty expressing anger.

The majority of relationships between family members were reported as “very close” or “close.” The frequency of relationship types was calculated based on the total of all relationships reported by the consultand. Relationships between the consultand and her children were revealed unless the children were <12 years old. The consultand’s relationships with aunts, uncles, or grandparents were not always elicited because of time constraints or lack of contact with extended family. Of the 38 consultands, 81% reported close/very close relationships with their spouse, whereas 13% reported distant or fused and conflictual relationships with their spouse. When a consultand did not report a relationship with a spouse, it was because the spouse had died or the couple was divorced; this occurred in 6% of genograms. If the consultand was remarried, the relationship with the current spouse was recorded. Eighty-three % of con-

Table 1 Comparison of demographic and risk characteristics for genogram pilot and human genome study participants

	Genogram pilot	Human genome study
Demographics		
Mean age	46.6	41.5
Education		
≤ High school	21%	24%
> High school	79%	76%
Ethnicity		
Caucasian	100%	97%
Married	71%	73%
Risk characteristics		
Mean Gail score	14.09	15.8
Cancer family history ^a		
>1 FDR with Br/Ov Ca	16%	15%
1 FDR only	32%	33%
1 FDR and SDR	45%	51%
SDR only	5%	1%
Other	2%	

^a FDR, first-degree relative; Br/Ov Ca, breast/ovarian cancer; SDR, second-degree relative.

sultands reported having a close/very close relationship with their mothers, but only 70% reported these relationships with their fathers. Eight % reported having a fused and conflictual relationship with their mothers, and 11% reported fused and conflictual relationships with their fathers. Distant and estranged relationships were reported with lower frequency. Only 1% of consultands said that they had this type of relationship with their mothers, but 17% said that they had a distant or estranged relationship with their fathers. If the consultand did not report a relationship with a parent, that was because the parent had died when the consultand was a child. Of all consultands, 8% did not report relationships with their mothers.

Genogram Questionnaire. A great deal of additional information was learned during the interview about the consultand’s relationships and communication patterns within the family. Seventy-four % of those interviewed reported that they have a relative who has expressed interest in genetic testing. Thirty-nine % reported that they would feel most comfortable discussing genetic testing with their sister, whereas 34% of those questioned felt they would be most comfortable talking to their spouses about genetic testing. Seventy-six % of our sample has actually donated blood for genetic testing research.

We explored at length whether cancer was a subject that could be discussed openly within the family. Eighty-three % of those questioned felt that their family was able to talk about cancer openly with the majority of family members, although an occasional family member might be excluded from these discussions. When asked whether family members agreed about the cancer diagnosis in the family, only 57% felt that all members of the family agreed about the implications of the cancer diagnosis within their family.

The consultand was then asked about her involvement in the physical care of the family member with cancer and how the caretaking decisions were made. Fifty-seven % of those interviewed had responsibility for the physical care of a family member with cancer. The majority of these felt that their proximity to the sick relative determined their involvement, although physical health, a nursing background, and position in sibship (oldest *versus* youngest) were key factors.

The immediacy of the cancer threat was explored by asking whether anyone in the family thought they would be next to develop cancer. Forty-five % responded affirmatively,

Fig. 1. Lines used to standardize genogram construction. The different types of lines are drawn between individuals depicted in a genogram to represent the nature of their relationships. Developed by McGoldrick and Gerson (6).

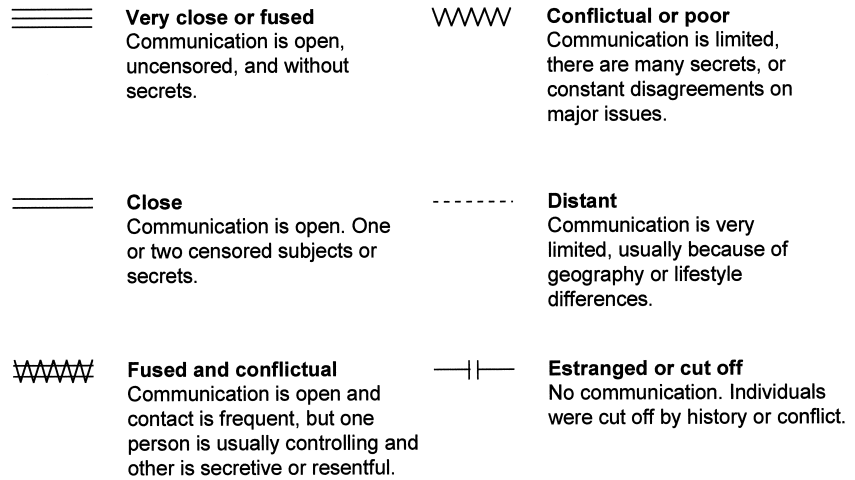
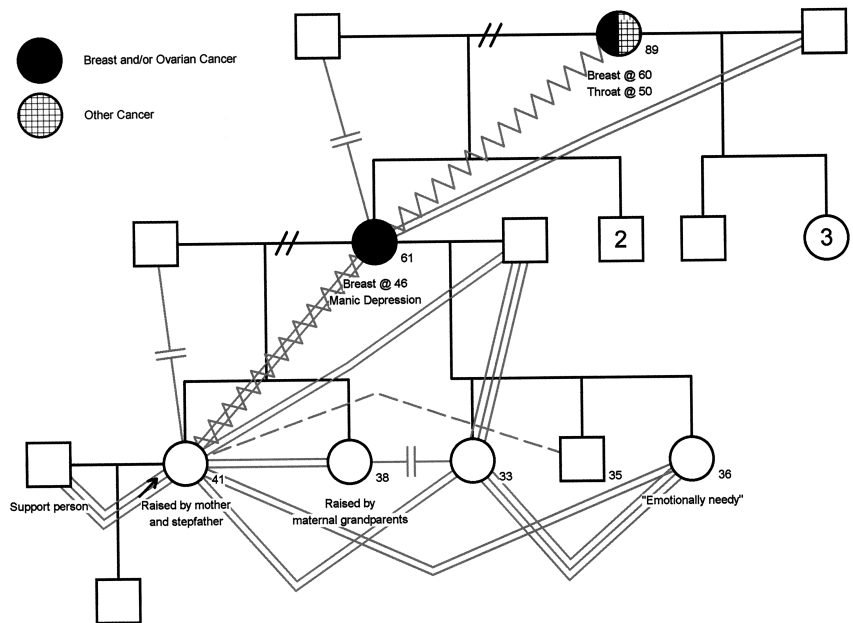


Fig. 2. Genogram of a family exhibiting conflictual and sometimes estranged patterns of communication. Lines depict the quality of communication between two individuals; see Fig. 1 for definitions.



with 21% of the consultands feeling that they would be the next person in their family to develop cancer.

Genogram Validity. Finally, in an attempt to determine the validity of the genogram, the percentage of close and very close relationships reported was correlated with SASSR scores from the spare time, family of origin, and family unit subscales. The SASSR is a validated tool and an excellent measure of social adjustment (12). SASSR scores ranged from a minimum of 25 to a maximum of 45, with a mean score of 32.9. SASSR scores and % close/very close (%C/VC) relationships were found to be significantly correlated ($r^2=0.15$; $P = 0.014$). That is, the higher the percentage of close and very close relationships represented on the genogram, the greater the social adjustment recorded on the SASSR subscales.

Discussion

Genetic counseling traditionally acknowledges the importance of family in a client’s life. Concern for the emotional well being

of the client, maintenance of the quality of family relationships, and respect for the beliefs and attitudes of the extended family have long characterized the role of the genetic counselor (3). However, very few studies have systematically evaluated the communication of genetic knowledge within a family, and fewer still have systematically investigated the effect of cancer risk information on family relationships. The genogram, first used by Murray Bowen in 1978 (6) as part of Family Systems Theory, has been used extensively in family therapy practice, providing insight into how the individual functions in the context of the family system, and likewise, how individuals interact as a functional whole (13). McGoldrick and Gerson (6) later standardized genogram construction, thereby providing practitioners with a consistent and reliable structure for recording family dynamics. Eunpu (3) supports the use of the genogram to incorporate the exploration of family relationships within the genetic counseling setting.

In this study, genograms were constructed for 38 high-risk

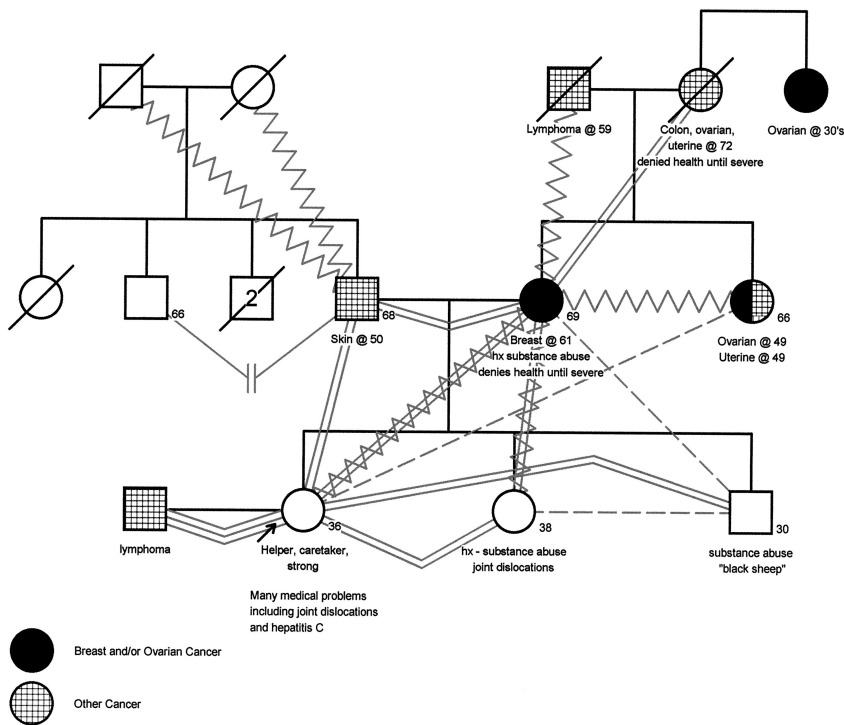


Fig. 3. Genogram of a family exhibiting troubled relationships that are repeated across generations. Lines depict the quality of communication between two individuals; see Fig. 1 for definitions.

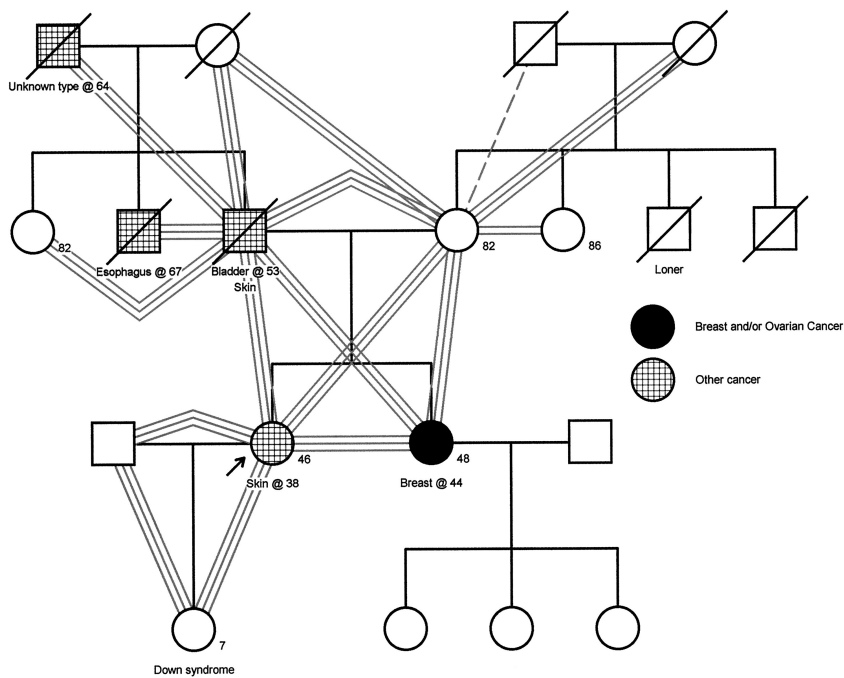


Fig. 4. Genogram of a family exhibiting supportive relationships. Lines depict the quality of communication between two individuals; see Fig. 1 for definitions.

women to pilot the efficacy of this technique for assessing family relationships. Participants reported the majority of their family relationships to be “close or very close.” The degree of family cohesion reflected by the genogram correlated significantly with social adjustment as measured by the SASSR. This finding helps establish the validity of the questions used in genogram construction and indicates that the genogram is ac-

curately capturing the consultant’s assessment of familial relationships, without biased interpretation by the interviewer.

There are several potential limitations to using the genogram in the context of research and counseling. The first of these is the subjective nature of the information recorded by the counselor. By standardizing both the content and the structure of the questions asked and adhering to defined symbols and nomenclature, we have

attempted to convert qualitative data into a graphical representation of family dynamics that may be used in comparative and longitudinal research and counseling settings.

A second potential limitation is the issue of sample bias; this was a self-referred, highly motivated and educated population. The majority of the genograms collected in this pilot study illustrate relationships that are close and/or very close. Perhaps it is a reflection of the individual's own attitudes, behaviors, and status within the family network (14) that enable the individual to seek cancer risk counseling. It is also possible that our study population comes from families who have a history of chronic illness and have therefore developed, as a family, coping strategies for successfully dealing with chronic illness. Gerson (15) outlines the phases and stages through which families evolve, the challenges they deal with during each stage, and the impact a crisis, such as illness, can have on the family at each stage. Some stages of the family life cycle may involve closer interaction with other family members and represent a "contraction phase." It is possible that the majority of families in this study are in a similar family life cycle stage that involves a more interactive mode of behavior and/or coping among family members.

A third limitation of genogram construction is interviewer bias. Although information was collected using the standardized questionnaire, the interviewer was ultimately responsible for interpreting the consultant's responses and diagrammatically completing the genogram. Although it is difficult to address interrater reliability in this small sample, 95% of the genogram sessions were conducted by only three interviewers. They followed the same format, using the genogram questionnaire as a foundation for exploring family relationships. All questions were asked in the same order, and the genogram was constructed based on the relationship questions in the survey. All participants were given the opportunity to amend the genogram upon its completion by the interviewer. Because of the structure and the unique personal nature of the information used to construct the genogram, it may lack the rigor of a more formal psychometric tool. However, the richness of the data provided and its potential usefulness in the individual counseling setting offset this limitation.

A final limitation is the possibility that the consultants answered questions about their familial relationships in a socially acceptable way. This is a problem inherent in most social research studies. However, our study population was already focused on family issues by virtue of their selection into a FRAP. We were careful in the informed consent process to emphasize that the information provided would be kept confidential and that there was no obligation to participate.

Genetic counseling is a communication process, helping individuals and their families understand all of the options available for coping with a genetic condition or undergoing genetic testing, to make decisions consonant with a family's goals, and to make the best possible adjustment to a genetic diagnosis or risk (3). The drawing or construction of one's family history, in the form of a pedigree, is the most commonly used and one of the most powerful tools in medical genetics and genetic counseling (15–17). It is the first and often best source of information for genetic risk analysis, making it a basis of treatment (3). Its various symbols and markings provide a universal language that attempts to display a family's complex biological relationship as a graphic model (17). It is drawn to establish the pattern of inheritance of traits, clarify a clinical genetic diagnosis, and assist in identifying at-risk individuals in a family (15, 16). Relied upon by molecular and medical geneticists, it is used to calculate recurrence risks and to do gene mapping via linkage analysis and other genetic family studies (18).

In a similar fashion, the genogram can serve as a tool to members of a multidisciplinary clinical genetics team to provide a

recorded memoir of a family's past and present attitudes and beliefs about genetic risk, as well as a record of the quality of relationships and dynamics within the family. For the consultant anticipating the receipt of genetic testing results, the genogram may be used in a therapeutic way to point out support networks or possible difficulties within the family. Although both the pedigree and the genogram are derived from interview data, the visual representation of that data can enhance the appreciation of familial patterns of both medical and social significance.

For genetic counseling to be truly helpful, it will be important to identify efficient and innovative methods to make genetic information available and relevant for the counselors and consultants (3). We propose that the genogram may serve as an innovative adjunct to genetic counseling to meet the future demands and challenges facing the genetic counseling profession.

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