

The Need for Developing Standardized Family Pedigree Nomenclature

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To assess the variation in usage of symbols used in recording a genetic family history, full members of the National Society of Genetic Counselors were surveyed by questionnaire. The questionnaire return rate was 55.3% and genetic counselors from a broad range of clinical experience, genetic counseling training programs and geographic regions responded. There was striking variation in symbols used for recording routine medical information in a genetic family history (i.e., pregnancy, spontaneous abortion, termination of pregnancy). There was even less consensus in recording situations representing new reproductive technologies (i.e., artificial insemination by donor semen, donor ovum, surrogate motherhood). The results of this survey document the need for developing standardized nomenclature in recording genetic family histories as a quality assurance measure in the delivery of genetic services. Such standardization will reduce the chance of incorrect interpretation of patient and family medical and genetic information.

KEY WORDS: family history; genetic counseling; pedigree; quality assurance; reproductive technology.

INTRODUCTION

The family pedigree is one of the most powerful tools of a genetic counselor. It serves not only as a history-taking tool to record biological

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relationships and facts, but also as a sociological aid in counseling by serving as a record of family social relationships. The human pedigree as constructed by human genetics professionals lies somewhere between a purely biological pedigree, such as would be recorded by animal breeders, to the genogram used by family therapists in which social relationships and interactions are emphasized. An example of a biological pedigree is shown in Fig. 1 in which the lineage of a quarter horse with hyperkalemic periodic paralysis is recorded and features such as color and height may be noted (courtesy of the American Quarter Horse Association, A. Q. H. A.). This contrasts to the genogram of Sigmund Freud's family in Fig. 2 in which personal relationships between individuals are recorded, such as the conflicted relationship between Sigmund and his sister Anna (recorded as a jagged line), the overly close relationship between Sigmund and his daughter Anna and mother Amalia (recorded as three horizontal lines), and the distant relationship between Sigmund and his father Jakob (recorded as a dotted line) (McGoldrick and Gerson, 1985).

The purpose of the family pedigree in genetic counseling is to record medical information and family relationships in a short-hand fashion so that such information can be easily and quickly interpreted. This facilitates establishing a clinical diagnosis, assists in identifying the pattern of inheritance of a condition and identifies at-risk individuals in a family.

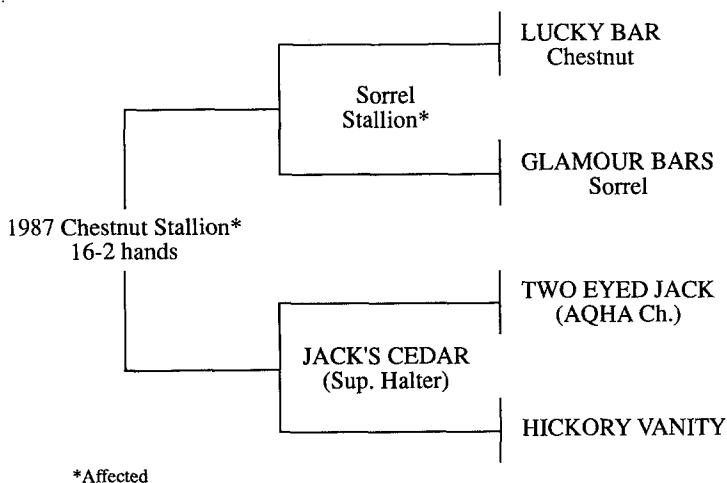


Fig. 1. Lineage of quarterhorse with hyperkalemic periodic paralysis.

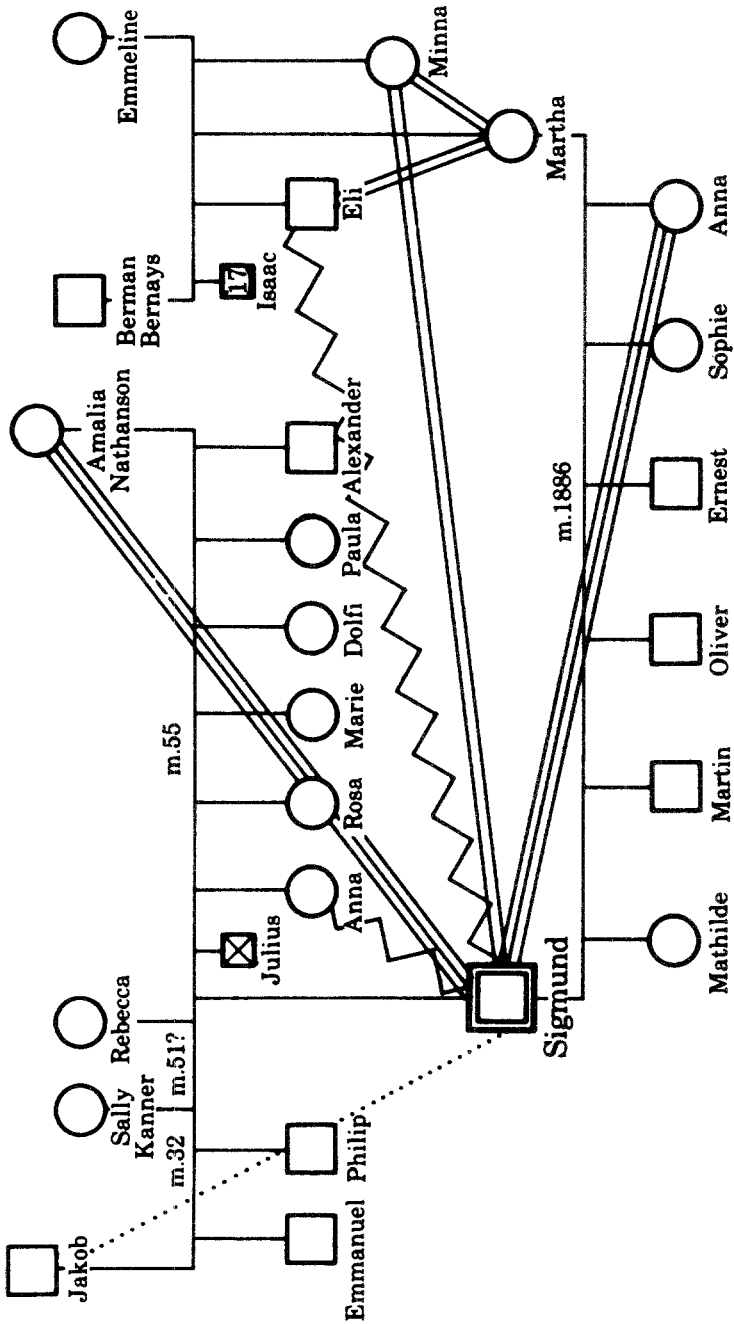


Fig. 2. Genogram of Freud family (printed with permission from McColdrick and Gerson, 1985).

If the short-hand information is not easily interpretable, the utility of a family pedigree is decreased and incorrect information may be conveyed to the family or to medical professionals. For example, if a client has had three spontaneous abortions but the pedigree is interpreted as showing three voluntary pregnancy terminations, incorrect information may be conveyed to other medical professionals and/or the client. Another example is if a symbol representing a woman who is pregnant through artificial insemination by an unidentified donor is interpreted by a genetic counselor as meaning the woman is pregnant with an unmarried partner (Fig. 6m), previously established rapport with the client may be disrupted. A pedigree should be a tool to facilitate genetic counseling; if symbols are not interpreted in a standardized fashion the pedigree may be an impediment.

The challenge of drawing easily interpreted pedigrees increases with the ever expanding myriad of new reproductive technologies. How should surrogate motherhood be symbolized? How should artificial insemination be symbolized? If information about a gamete donor is known should this information be recorded in the pedigree and in whose medical record should this information be placed? The complexity of drawing pedigrees grows as genetic counseling is being provided to more and more blended families and nontraditional family units. Should a purely social relationship such as a same sex relationship be recorded on a pedigree? How should the pregnancy of a lesbian couple conceiving a pregnancy with a known or unknown sperm donor be recorded on the pedigree?

To document the need for establishing standard pedigree nomenclature, in the summer of 1992 we polled full members of the National Society of Genetic Counselors (NSGC). We elicited their choice of symbols for many routine pedigree situations and symbols for individuals using alternate means of reproduction.

METHOD

A questionnaire was sent to full members of the NSGC requesting individuals to choose from limited options regarding pedigree symbolization for over 40 situations commonly encountered in genetic counseling. Copies of the questionnaire are available upon request. We chose to poll only full members as these are individuals who have been specifically trained to take genetic family histories and have significant experience with this activity.

RESULTS

Sample Demographics

Of the 790 questionnaires distributed, 437 responses were received for a return rate of 55.3%. The respondents represented a wide geographic distribution with representation from all six of the NSGC regions (Table I). A number of genetic counseling training programs were represented (Table II). The respondents had varied backgrounds in terms of their clinical experience (Table III). Thus the opinions of a wide spectrum of experienced genetic counselors were assessed.

Survey Responses

Although the respondents were given limited options to choose for each pedigree scenario, many respondents illustrated other choices, making the data difficult to interpret and confirming our hypothesis that there is wide variation in symbols used. There was no overall agreement on which symbol should be used for *any* of the pedigree scenarios, even for the common pedigree symbols such as pregnancy or miscarriage. For this reason, we will not discuss the responses for each survey question individually.

Table I. Survey Respondents by NSGC Region

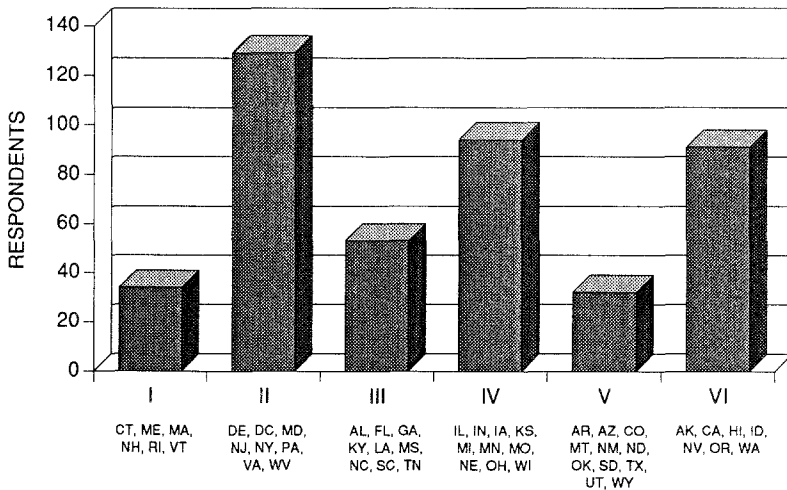


Table II. Genetic Counseling Training Background of Respondents

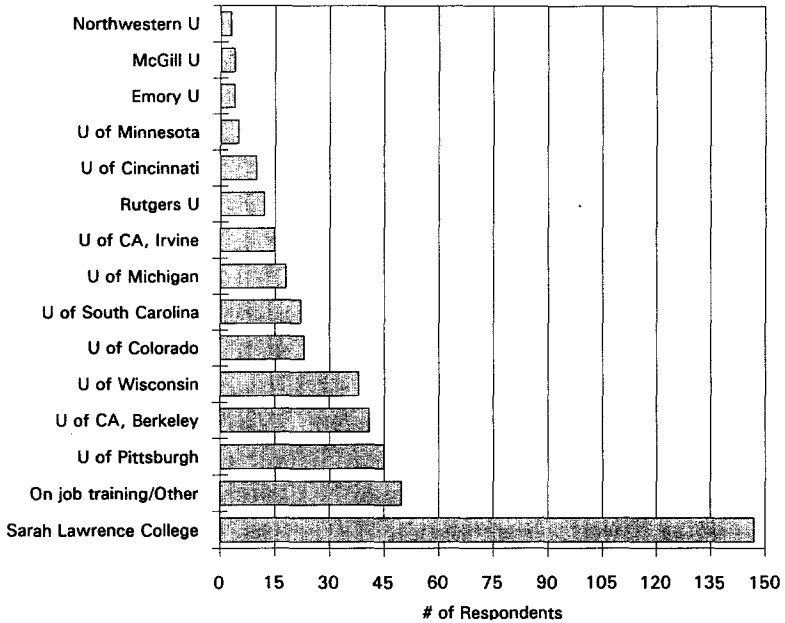
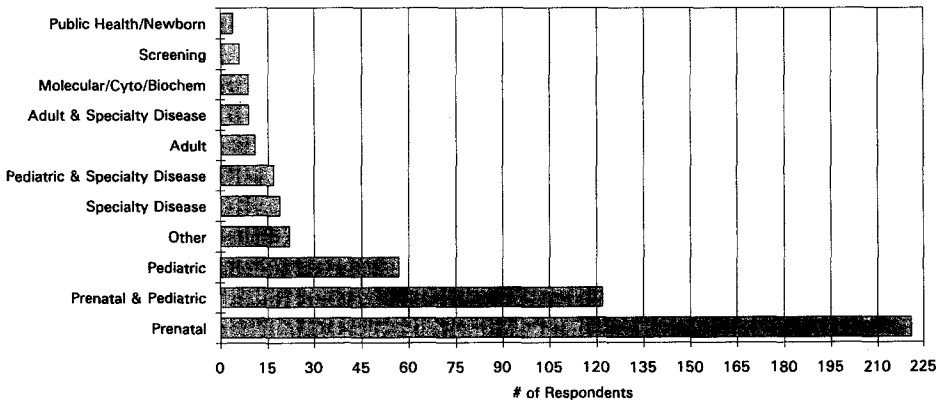


Table III. Respondents' Primary Focus of Genetic Counseling



Common Pedigree Symbols and Abbreviations

Pregnancy is one of the symbols most frequently used by genetic counselors, yet no standard symbol emerged from the survey. In fact, pregnancy was symbolized at least 17 different ways (Fig. 3). Some of the symbols used for pregnancy were given another meaning by other genetic counselors. For example, Figs. 3e and 3g were used by some genetic counselors to symbolize spontaneous abortion. Figure 3i was used to represent a pregnancy in which the sex of the fetus was known to be female. Figure 3n was used when the number of offspring of an individual was unknown and Fig. 3o was used by some counselors to represent adoption. A spontaneous abortion of a fetus of unknown sex was symbolized 12 different ways (Fig. 4).

Adoption was also symbolized in numerous ways (Fig. 5). Adopted-in (where the nonbiological family is shown) and adopted-out (where the biological family is drawn) were usually symbolized differently although some-

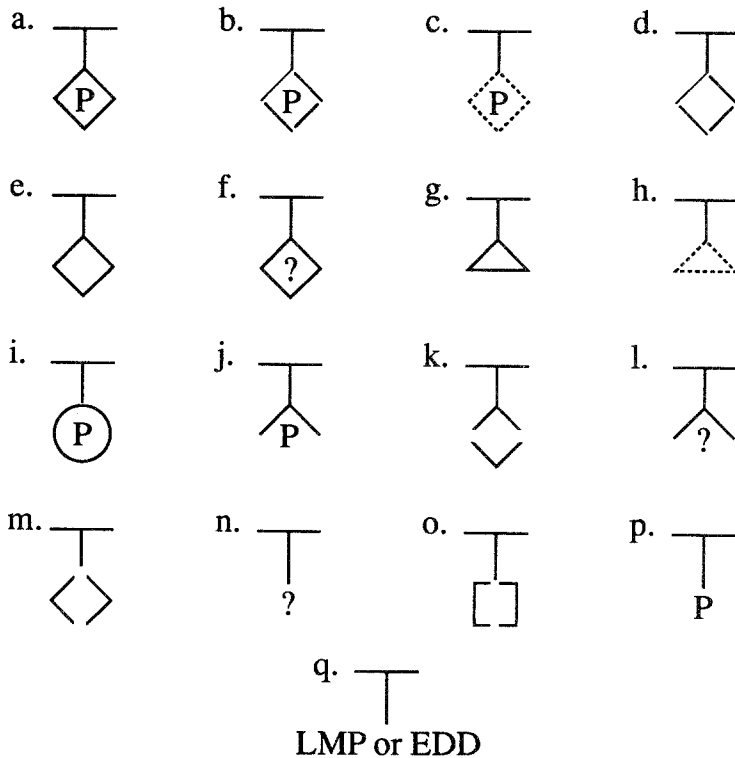


Fig. 3. Symbolization of pregnancy.

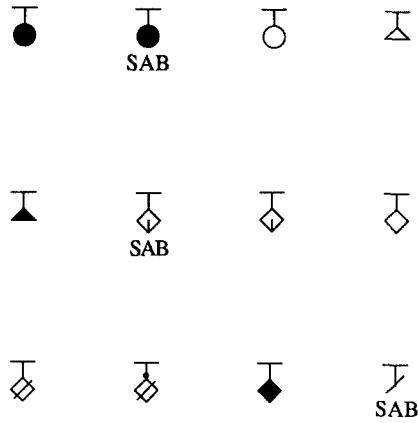
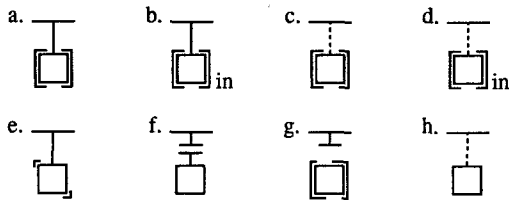


Fig. 4. Symbolization of spontaneous abortion — sex unknown.

times a distinction was not made between the two as illustrated in Figs. 5a and 5i. Many respondents used a dotted or broken line to denote the non-biological relationship for an individual who is adopted into a family as shown in Figs. 5c, 5d, 5f-h.

IN (non-biologic family shown)



OUT (biologic family shown)

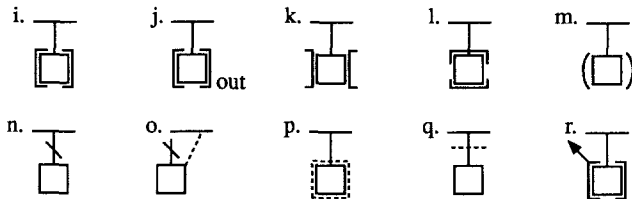


Fig. 5. Symbolization of adoption.

Abbreviations were not standardized. For example ET, ET.P, MIP, VIP, ab-e, EAB, elab, VTOP, TOP, TA were some examples of abbreviations used for a voluntary termination of pregnancy. Spontaneous abortion was abbreviated a number of ways including: misc, s.a., SMC, spab, sponab, sp abort, SAB and SAb.

New Reproductive Technologies

The responses for symbolization of scenarios using new reproductive technologies were extremely variable. A major question arises in choosing symbols for these alternative forms of reproduction; should nonbiological information be recorded on the pedigree? For example, in the instance of artificial insemination by a donor should the pregnancy line be between the biological unit (such as the woman and the sperm donor) or between the social couple (such as the woman and her partner)? Regarding artificial insemination, the survey respondents chose symbols which varied along a continuum of emphasis from biological to social relationships (Fig. 6). Some survey respondents did not record the sperm donor on the pedigree at all as in Figs. 6g and 6h. Some respondents felt only biological relationships should be shown and the male or female partner in the relationship (the nonbiologic parent) was not included on the pedigree as illustrated in Figs. 6a-d. The respondents who stressed social relationships included all social relationships, including same sex relationships, and put the pregnancy line between the social couple as in Figs. 6e-i. Other survey respondents did not record the sperm donor on the pedigree at all as in Figs. 6g and 6h. Some respondents felt only biological relationships should be shown and the male or female partner in the relationship (the nonbiologic parent) was not included on the pedigree as illustrated in Figs. 6a-d. Many respondents included both biological and social relationships (Figs. 6j-m) and usually put the pregnancy line between the biological couple (the woman and sperm donor) but recorded the social relationship as well.

DISCUSSION

From our results it seems obvious that standardization of family pedigree symbols is necessary much as it was for cytogenetic nomenclature in the 1970s (Paris Conference, 1971). In the process of choosing standardized pedigree symbols, should symbols be chosen because they represent the usage of the majority of genetic professionals, or should symbols be chosen which make the most sense? For example (Fig. 7), perhaps a diamond

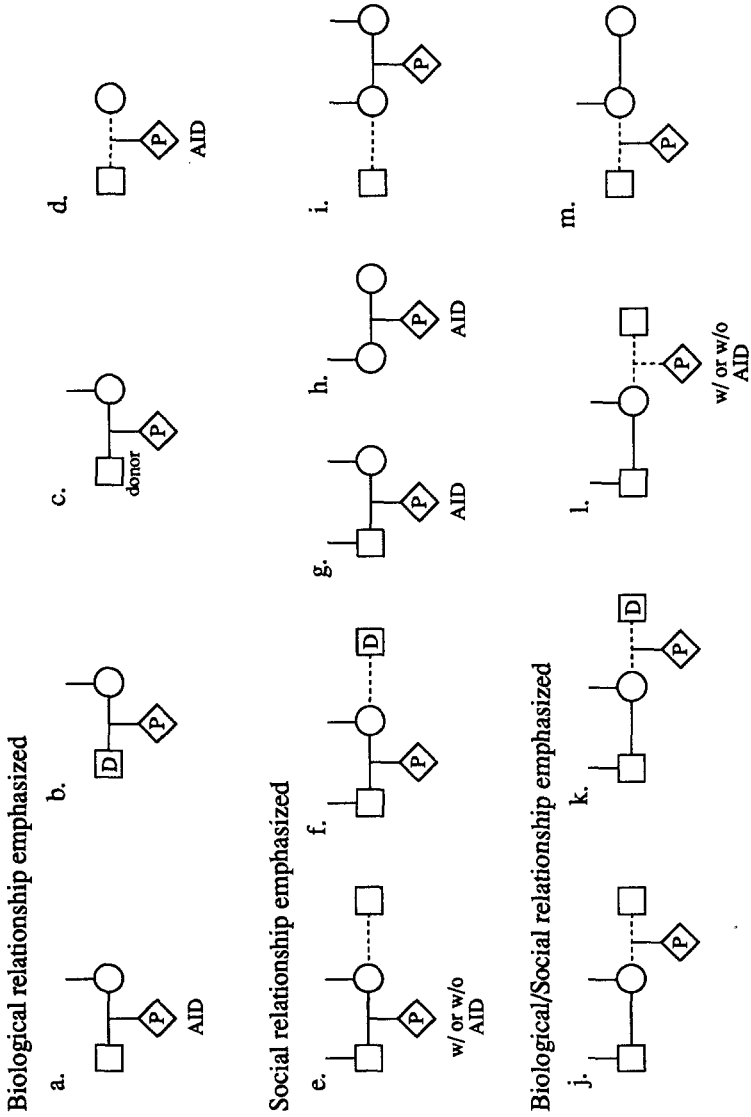
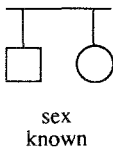
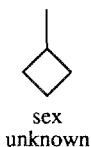
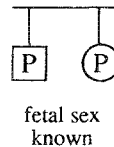


Fig. 6. Symbolization of artificial insemination with an undetermined sperm donor.

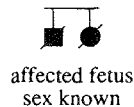
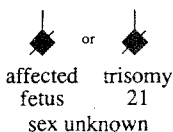
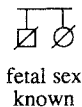
Individual



Pregnancy



Termination of Pregnancy



Spontaneous Abortion

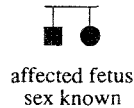
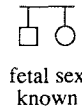


Fig. 7. Sample of possible pedigree symbols.

should represent an individual with sex unknown, a “P” could be placed inside it for a pregnancy, and a “P” placed inside a square or circle would represent the pregnancy when the fetal sex is known. A small diamond with a slash through it could represent a termination of pregnancy and a filled in diamond with a slash through it could represent the termination of pregnancy of a known affected fetus with an explanation of the diagnosis (such as trisomy 21) below the symbol. If the sex of the affected fetus were known, the symbol would be a small square or circle with a slash through it. A small diamond could represent a spontaneous abortion where the sex of the fetus is unknown, whereas a small square or circle would represent a spontaneous abortion of a fetus of known sex. These suggestions may not represent what the majority of genetic professionals are currently using for symbolization, but we believe that the time has come to examine the rationale for using a particular pedigree symbol.

As standardized pedigree symbols are developed, it is important that this information be distributed. Programs which train health professionals, such as genetic counseling training programs, M.D. and Ph.D. human and

molecular genetic training programs and schools of nursing and social work should be provided with curricula using standardized pedigree symbols. Adoption workers should be trained to use the same pedigree symbols in taking a family history. Science text books for high school students and genetic text books for college and graduate students should have standardized symbols. The various computer software programs which are being developed to record genetic family histories should use universal symbols. A recommendation should be made that standard pedigree symbols be part of the board examination for genetic counselors, medical geneticists and related genetic professionals. Editors of the human genetics journals should require uniform pedigree symbolization in submitted papers.

CONCLUSION

Standardization of the family genetic pedigree has the potential to improve the quality of care provided by genetic professionals. Such symbolization is not meant to create a foreign language but a universal language. In developing standard symbols it is important to keep in mind the ethical issues involved with recording a genetic family history. What types of information should be recorded (i.e., social information such as marital status, full names of family members, infectious diseases such as HIV)? The type of information recorded may depend on where the pedigree containing social information is stored (i.e., the genetic professional's personal files or the patient's medical record). Who should have access to the family history? As advances in carrier detection, presymptomatic testing, prenatal diagnosis and gene therapy become available for more and more genetic disorders, the demand for genetic services will increase, creating the need for more individuals trained in the methods of obtaining accurate family history information. The pedigrees constructed by these health professionals should be easily interpreted by others within their profession as well as among other medical disciplines.

The Professional Issues Committee of the NSGC has begun to address the development of standardized pedigree symbols by forming a Task Force of genetic professionals to recommend pedigree nomenclature. This quote from an anonymous questionnaire respondent nicely illustrates the challenges ahead:

"Genetic counselors often sit right on the boundary of medicine and counseling. As a result we must alternate our role with clients. However, we must not blur the distinctions between genetic and social responsibilities through the use of confusing pedigrees or ones that mix social and bio-

logical ties. Even as I completed this exercise I realized how difficult it became to follow my own advice!”

ACKNOWLEDGMENTS

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