J Genet Counsel (2008) 17:424–433
DOI 10.1007/s10897-008-9169-9

PROFESSIONAL ISSUES

Standardized Human Pedigree Nomenclature: Update and Assessment of the Recommendations of the National Society of Genetic Counselors

Robin L. Bennett · Kathryn Steinhaus French · Robert G. Resta · Debra Lochner Doyle

Received: 12 April 2008 / Accepted: 29 May 2008 / Published online: 16 September 2008
© National Society of Genetic Counselors, Inc. 2008

Abstract In 1995, the Pedigree Standardization Task Force (PSTF) of the National Society of Genetic Counselors (NSGC) proposed a system of pedigree nomenclature. Recently, the PSTF (now called the Pedigree Standardization Work Group or PSWG) sought evidence that the published symbols met the needs of health professionals, were incorporated into health professional training and were utilized in publications. We searched PubMed and reference lists of select publications, reviewed the Instructions for Authors of several journals, searched the websites of professional societies, sought comment from the membership of the NSGC, and looked at recommendations and training practices of various health professional organizations. Many journals still do not cite specific standards for pedigrees, but those found cited the PSTF nomenclature. We did not find significant objections or alternatives to the 1995 nomenclature. Based on our review, we propose only a few minor stylistic changes to the pedigree symbols. The pedigree nomenclature of the NSGC is the only consistently acknowledged standard for drawing a family health history. We recommend regular and continued review of these pedigree standards to determine if additional symbols are needed to accommodate changes in clinical practice to ensure that the symbols continue to meet the needs of health professionals and researchers as well as adhere to evolving ethical and privacy standards. All health professionals, trainees, and researchers should be made aware of the utility of using a common pedigree nomenclature in clinical practice and publication. This will become particularly important as electronic medical records become more widely utilized.

Keywords Electronic medical record · Family history · Genetic counseling · National Society of Genetic Counselors · Pedigree symbols · Pedigree standardization

R. L. Bennett (✉)
Division of Medical Genetics, Department of Medicine,
University of Washington Medical Center,
Box 357720, Seattle, WA 98195-7720, USA
e-mail: robinb@u.washington.edu

K. S. French
Division of Genetics and Metabolism, University of California, Irvine,
Orange, CA, USA

R. G. Resta
Hereditary Cancer Clinic, Swedish Cancer Institute,
Seattle, WA, USA

D. L. Doyle
Genetic Services Section,
Washington State Department of Health,
Seattle, WA, USA

Introduction

A medical pedigree is a graphic presentation of a family's health history and genetic relationships and it has been a pivotal tool in the practice of medical genetics for nearly a century (Bennett 1999; Resta 1993). With the mapping of the human genome and the increasing role of genetics in daily medical practice, recording and interpreting a pedigree should be a standard competency of all health professionals (Bennett 1999; Center for Disease Control 2008; HHS 2008; NCHPEG 1995).

In the early 1990s, the National Society of Genetic Counselors Pedigree Standardization Task Force (NSGC PSTF) documented that even genetics professionals were using an inconsistent array of symbols and nomenclature to record pedigrees (Bennett et al. 1993; Steinhaus et al. 1995).

The PSTF worked with professional genetic societies, prominent medical genetics professionals, genetics educators, and patient advocacy groups to develop standardized pedigree nomenclature (Bennett et al. 1995). In this review, we assess the utilization and acceptance of the 1995 pedigree nomenclature by health professionals, educators, and in publications. A systematic review of clinical practice was not performed, although a literature review was conducted. We propose some minor stylistic changes to the nomenclature and provide suggestions for future development.

Pedigree Nomenclature in Publications

The NSGC Pedigree Standardization Work Group (PSWG) employed several strategies to determine if there were challenges to the standardized pedigree nomenclature published in 1995. To look for published references to the nomenclature we searched PubMed using the key words *pedigree nomenclature, pedigree standards, pedigree standardization, family history standards, family history standardization, and family history nomenclature* over the period of 1994 to August 2007. Using SCOPUS Preview (2007), we searched for journal articles in English that cited the nomenclature. The *Instructions for Authors* of English language journals with a focus on clinical genetics that have a history of publishing pedigrees were reviewed to see if PSTF nomenclature was recommended. The websites of several professional societies of journal editors were searched for recommendations for publishing pedigrees.

The PSTF nomenclature is referred to in a variety of classical human genetics texts (referenced in Table 1). The *AMA Manual of Style* (2007) and several human and medical genetics journals cite the PSTF recommendations in their Instructions for Authors (*American Journal of Human Genetics* (http://images.cell.com), *American Journal of Medical Genetics* (http://www3.interscience.wiley.com), *Genetics in Medicine* (http://edmgr.ovid.com), and *Journal of Genetic Counseling* (www.springer.com)). Other genetics journals with a significant focus on clinical genetics did not have any recommendations in their Instructions to Authors regarding the publication of pedigrees or specific standards for drawing pedigrees (such as *Clinical Genetics* (http://blackwellpublishing.com), *European Journal of Human Genetics* (http://www.nature.com/ejhg), *European Journal of Medical Genetics* (http://www.elsevier.com), *New England Journal of Medicine* (www.nejm.org), *Nature Genetics* (www.nature.com/ng), and *Lancet* (www.thelancet.com)). We are not aware of any clinical genetic journals that expound a different pedigree nomenclature to use in publications than that proposed by the PSTF. The websites for the Council of Biology Editors (http://writing.colostate.edu), the European Association of Science Editors (http:www.ease.org.uk), the International Committee of Medical Journal Editors (http://www.icmje.org), and the World Association of Medical Editors (http://www.wame.org) do not include guidance for publication of pedigrees.

Table 1 Examples of Journals, Text and Reference Books Which Include the National Society of Genetic Counselors Standard Pedigree Nomenclature

General reference
AMA Manual of Style. A Guide for Authors and Editors, 10th Edition (2007)
Journals
American Journal of Medical Genetics
American Journal of Human Genetics
Genetics in Medicine
Journal of Genetic Counseling
Text books
A Guide to Genetic Counseling (Schuette and Bennett 1998)
ASCO Curriculum: Cancer Genetics and Cancer Predisposition, 2nd edition (2004)
Emery's Elements of Medical Genetics, 13th edition (Turnpenny and Ellard 2007)
Emery and Rimoin's Principles and Practice of Medical Genetics, 5th Edition (Kingston 2007)
Neurogenetics: Scientific and Clinical Advances (Bennett 2006a)
Oxford Desk Reference: Clinical Genetics (Firth et al. 2005)
Principles of Molecular Medicine, 2nd Edition (Bennett 2006b)
The Genetic Basis of Common Disease, 2nd Edition (LeRoy and Walker 2002)
Thompson & Thompson, Genetics in Medicine, 7th Edition (Nussbaum et al. 2007)

Scientific and medical journals as well as professional societies of journal editors should adopt standardized pedigree nomenclature. As we documented previously (Steinhaus et al. 1995), wide variation in pedigree nomenclature (even within the same journal) can potentially lead to misinterpretation of data and errant scientific conclusions based on the publication of the pedigree.

Publication of Pedigrees and Confidentiality

When pedigrees are published, journals should assure that standardized nomenclature is followed. Care should be taken to preserve patient and family confidentiality, particularly since the widespread availability of professional journals and scientific reporting makes it more likely that a pedigree will be recognized by a family member or by individuals familiar with the family. Researchers should consider obtaining consent from research participants to allow publication of a pedigree when enrolling subjects in a study, and to show participants a sample of a generic pedigree that is similar to one that might appear in a publication (Bennett 2000; Byers and Ashkenas 1998). A

pedigree should not contain information about which a subject had no prior knowledge. In other words, a person who had presymptomatic or susceptibility genetic testing through research should not find out about increased or decreased disease risk status from a publication. Likewise, health professionals have a duty to warn a patient about potential genetic risks to the patient's relatives (Offitt et al. 2004); such conversations should occur before the publication of a pedigree (Bennett 2000; Botkin et al. 1998). To preserve confidentiality, the minimum amount of information needed to convey the scientific finding should be included on the published pedigree. Authors should ask themselves how they would feel if their own pedigrees were being published and if the information would make them or their families uncomfortable (Bennett 2000).

The practice of masking or altering pedigrees should be addressed by the various societies and councils of journal editors along with the use of standard pedigree nomenclature. We discourage masking or altering pedigrees (Bennett 2000). Masking, which is usually evident to the reader, refers to an obvious change in the pedigree such as using diamonds to hide gender on a pedigree. Alteration involves changing information such as birth order or gender. A survey of 177 investigators who published pedigrees in peer reviewed journals indicated that 19% had altered the pedigree and 45% had not disclosed their alterations to the journal editor (Botkin et al. 1998). Although the minimal amount of information needed to provide scientific documentation should be used in a published pedigree, it is also essential to maintain the integrity of the pedigree with proper birth order, ages, and affected status as this information may be vital to pedigree interpretation. Alteration may hinder the recognition of important genetic paradigms such as anticipation, parent of origin effects, sex-linked or sex-limited expression, or in utero lethality. In addition, genetic and environmental factors that alter disease expression, or confounding effects of having common ancestors (as may be noted in the offspring of consanguineous unions or in populations with a high coefficient of inbreeding), may be missed if pedigrees are altered or masked (Bennett 2000).

Usage of the PSTF Nomenclature in Health Professional Training, Certification and Credentialing

We contacted the Transnational Alliance of Genetic Counselors (http://igce.med.sc.edu) and the directors of genetic counseling training programs that are accredited by the American Board of Genetic Counseling (http://www.abgc.net) (ABGC) to determine if the NSGC PSTF nomenclature was used in training of genetic counselors worldwide. We also communicated with the executive directors of the certifying or credentialing organizations for genetic counselors (ABGC), medical geneticists (American Board of Medical Genetics (http://www.abmg.org)), and advanced practice genetic nurses (the Genetic Nurse Credentialing Committee (http://www.geneticnurse.org)) to document whether the PSTF nomenclature was used in examination questions or in the credentialing process.

All genetic counseling training programs in North America accredited by the ABGC use the nomenclature. A query of the Transnational Alliance for Genetic Counseling notes use of the PSTF nomenclature in genetics professional training programs in Australia, China, South Africa, the United Kingdom, France, Israel, Spain, and Japan. As evidenced by educational resources on their websites.

Many professional societies have introduced the nomenclature to their membership as part of genetics/genomics family history initiatives, including The National Coalition for Health Professional Education in Genetics (NCHPEG 1995) (which includes representatives from over 100 diverse health organizations), the American Medical Association (http://www.ama-assn.org), the American Academy of Family Physicians (http://www.aafp.org), the American Academy of Physicians Assistants (http://pa.nchpeg.org), the American Society of Clinical Oncologists (ASCO 2004), the United Kingdom's Clinical Genetics Society (http://www.clingensoc.org), and the China Genetic Counseling Network (www.gcnet.org.cn). Jenkins and Calzone (2007) document a coalition of nursing societies that advocate for use of the PSTF nomenclature as a component of taking and interpreting a family history as a core nursing competency.

Changes Proposed to the Pedigree Nomenclature

We found no comprehensive alternative pedigree nomenclatures or recurring criticisms of the PSTF's recommendations. In June of 2002 and again in June of 2007, we solicited open-ended comments regarding use of the pedigree nomenclature from members of the NSGC through its general listserv. Comments collected from the NSGC membership regarding use of the pedigree nomenclature were mostly related to stylistic preferences as compared to changes in the actual symbols, and there were no new comments that had not already been addressed in 1995, in the development of the original nomenclature. Figures 1, 2, 3 and 4 reflect minor changes from the original 1995 nomenclature. The following changes and the respective rationale are noted:

1. A diamond for the "individual symbol" can be used to reflect *persons with gender not specified* and can be used for persons with *congenital disorders of sex developments (DSD)* and also with *transgendered individuals* (Fig. 1.1). The concept of gender identity

Fig. 1 Common Pedigree Symbols, Definitions, and Abbreviations.

Instructions:
— Key should contain all information relevant to interpretation of pedigree (e.g., define fill/shading)
— For clinical (non-published) pedigrees include:
  a) name of proband/consultand
  b) family names/initials of relatives for identification, as appropriate
  c) name and title of person recording pedigree
  d) historian (person relaying family history information)
  e) date of intake/update
  f) reason for taking pedigree (e.g., abnormal ultrasound, familial cancer, developmental delay, etc.)
  g) ancestry of both sides of family
— Recommended order of information placed below symbol (or to lower right)
  a) age; can note year of birth (e.g., b.1978) and/or death (e.g., d. 2007)
  b) evaluation (see Figure 4)
  c) pedigree number (e.g., I-1, I-2, I-3)
— Limit identifying information to maintain confidentiality and privacy

	Male	Female	Gender not specified	Comments
1. Individual	b. 1925	30y	4 mo	Assign gender by phenotype (see text for disorders of sex development, etc.). Do not write age in symbol.
2. Affected individual				Key/legend used to define shading or other fill (e.g., hatches, dots, etc.). Use only when individual is clinically affected.
				With ≥2 conditions, the individual's symbol can be partitioned accordingly, each segment shaded with a different fill and defined in legend.
3. Multiple individuals, number known	5	5	5	Number of siblings written inside symbol. (Affected individuals should not be grouped).
4. Multiple individuals, number unknown or unstated	n	n	n	"n" used in place of "?".
5. Deceased individual	d. 35	d. 4 mo	d. 60's	Indicate cause of death if known. Do not use a cross (†)to indicate death to avoid confusion with evaluation positive (+).
6. Consultand				Individual(s) seeking genetic counseling/ testing.
7. Proband	P	P		An affected family member coming to medical attention independent of other family members.
8.Stillbirth (SB)	SB 28 wk	SB 30 wk	SB 34 wk	Include gestational age and karyotype, if known.
9. Pregnancy (P)	P LMP: 7/1/2007 47,XY,+21	P 20 wk 46,XX	P	Gestational age and karyotype below symbol. Light shading can be used for affected; define in key/legend.

Pregnancies not carried to term	Affected	Unaffected	
10. Spontaneous abortion (SAB)	17 wks female cystic hygroma	< 10 wks	If gestational age/gender known, write below symbol. Key/legend used to define shading.
11. Termination of pregnancy (TOP)	18 wks 47,XY,+18		Other abbreviations (e.g., TAB, VTOP) not used for sake of consistency.
12. Ectopic pregnancy (ECT)	ECT		Write ECT below symbol.

has evolved over the last decade. Current practice is to avoid early assignment of gender for people with disorders of sex development (defined by Vilain and colleagues as 'congenital conditions in which development of chromosomal, gonadal, or anatomic sex is atypical') (Vilain et al. 2007). In addition, some individuals identify themselves somewhere along the spectrum between the opposites of male and female (Adultsociety.com 2007). The Pedigree Standardization Work Group continues to recommend that the male or female symbol be used to define the phenotypic gender, with the karyotype noted below the symbol, when known. This might be important, for example, in identifying the cancer risks faced by a *BRCA* mutation carrier who has

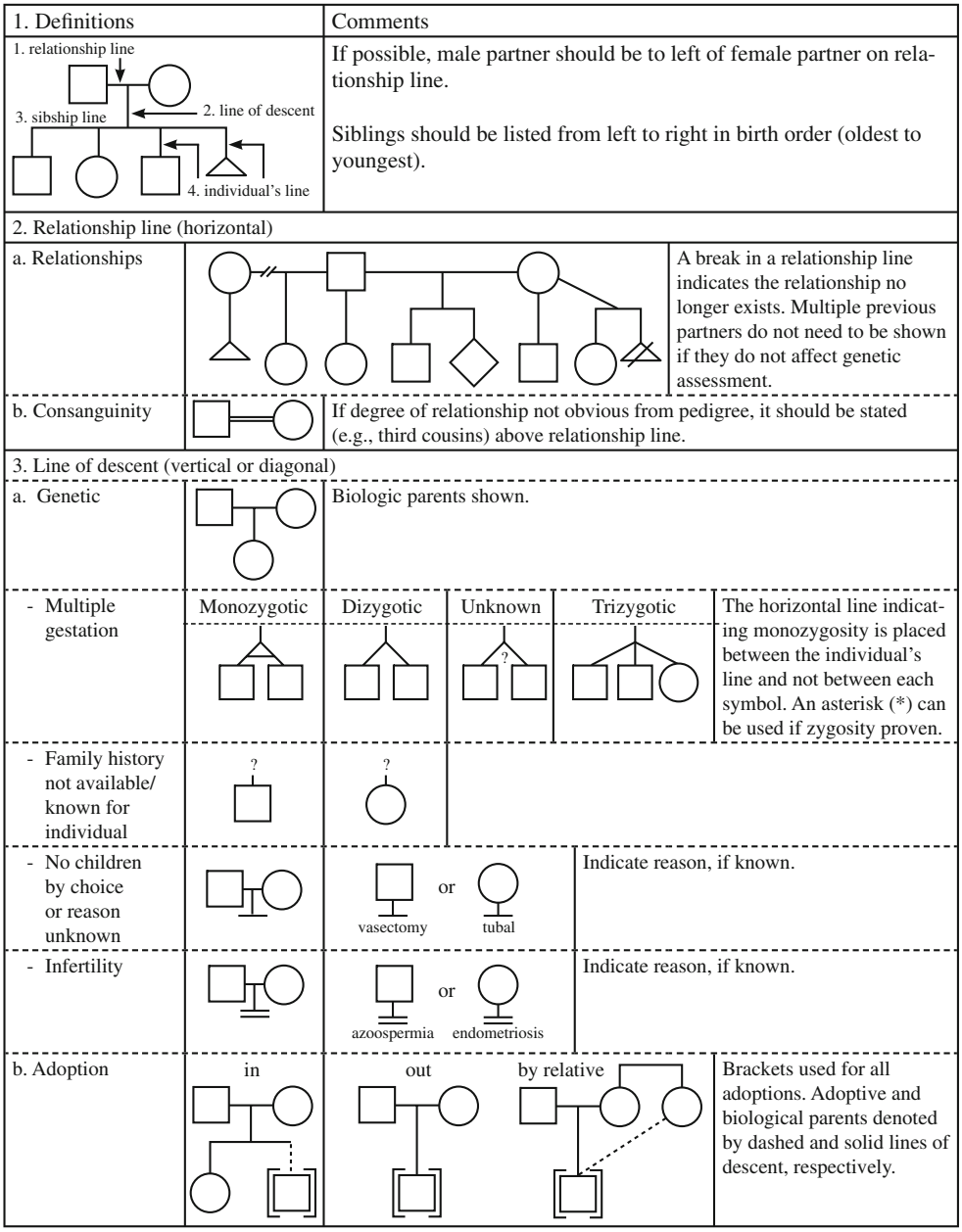


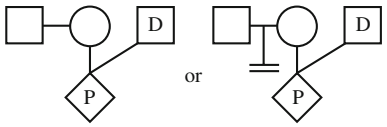
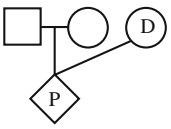
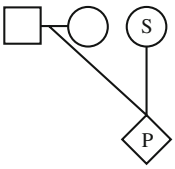
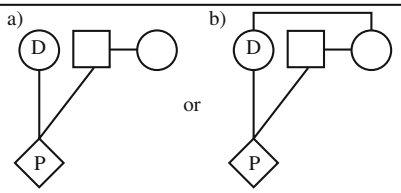
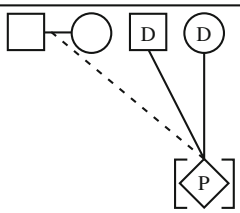
Fig. 2 Pedigree Line Definitions.

undergone female-to-male transgender surgery. A diamond can be used when it is not clinically relevant to assign gender (Fig. 1.1)

2. We no longer use a shorter "individual's line" for *pregnancies not carried to term* (Figs. 2.1–2.2). A pregnancy not carried to term is distinguished from other symbols by the unique use of a triangle, thereby making the shorter individual's line redundant graphically (Figs. 1.10–1.12).
3. We recommend including on the pedigree the *indication for referral* (reason for taking the pedigree) such as "abnormal ultrasound," "familial cancer," etc (see Fig. 1 Instructions). This helps to clarify the orientation of the information collected on the pedigree, because all pedigrees document information from directed health queries recording the consultand's personal medical and family history (Bennett 1999).
4. The Instructions (Fig. 1) were changed for the recorder to include *less identifying information* on the pedigree to be compliant with the Health Information Privacy, Access and Accountability Act (HIPAA) standards in the United States (http://www.hipaa.org). Initials or first names (in lieu of full names) may be enough to identify persons where medical records are documented and for orientation of discussions with the patient. Use of birth year or age, year of death or age at death, rather than birth date or date of death, would be more compliant with HIPAA guidelines since a full

Fig. 3 Assisted Reproductive Technology Symbols and Definitions.

Instructions:
— D represents egg or sperm donor
— S represents surrogate (gestational carrier)
— If the woman is both the ovum donor and a surrogate, in the interest of genetic assessment, she will only be referred to as a donor (e.g., 4 and 5)); the pregnancy symbol and its line of descent are positioned below the woman who is carrying the pregnancy
— Available family history should be noted on the gamete donor and/or gestational carrier

Possible Reproductive Scenarios		Comments
1. Sperm donor		Couple in which woman is carrying pregnancy using donor sperm. No relationship line is shown between the woman carrying the pregnancy and the sperm donor.
2. Ovum donor		Couple in which woman is carrying pregnancy using a donor egg and partner's sperm. The line of descent from the birth mother is solid because there is a biologic relationship that may affect the fetus (e.g., teratogens).
3. Surrogate only		Couple whose gametes are used to impregnate a woman (surrogate) who carries the pregnancy. The line of descent from the surrogate is solid because there is a biological relationship that may affect the fetus (e.g., teratogens).
4. Surrogate ovum donor		Couple in which male partner's sperm is used to inseminate a) an unrelated woman or b) a sister who is carrying the pregnancy for the couple.
5. Planned adoption		Couple contracts with a woman to carry a pregnancy using ovum of the woman carrying the pregnancy and donor sperm.

birth date or date of death is considered private and protected information.

Recording Pedigrees for Persons and Pregnancies in Relation to Assisted Reproductive Technologies

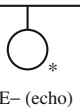
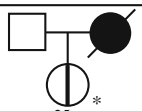
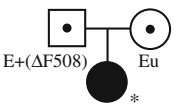
The symbolization for depicting pregnancies achieved through assisted reproductive technologies (Fig. 3) has been widely adopted. A dashed line of descent, used to identify a non-biological parental relationship (Fig. 2.3b), is particularly useful for documenting parental gamete donor, and gestational carrier relationships to a pregnancy. However, the nomenclature does not show documentation of frozen embryos or transferred embryos. The convention of "E" for evaluation (Fig. 4) could be used to track genetic testing of transferred embryos, but this has not been used extensively by the community of professionals involved with assisted reproductive technologies. Further review is needed to consider the complexities of pedigree documentation of the number of embryos conceived, frozen, and implanted, along with their genetic testing history, particularly given that embryos may be "adopted" from "donor parents," and the high frequency of multiple gestations associated with assisted reproduction.

Tracking Genetic Evaluation and Testing on a Pedigree

NSGC members reported that the symbolization for Genetic Evaluation and Testing (Fig. 4) is not widely used, particularly the use of "E" for evaluation. Figure 4.3 has been generally accepted as the symbol for a person who is

Fig. 4 Pedigree Symbols of Genetic Evaluation/Testing Information.

Instructions:
— E is used for evaluation to represent clinical and/or test information on the pedigree
  a. E is to be defined in key/legend
  b. If more than one evaluation, use subscript (E_1, E_2, E_3) and define in key
  c. Test results should be put in parentheses or defined in key/legend
— A symbol is shaded only when an individual is clinically symptomatic
— For linkage studies, haplotype information is written below the individual. The haplotype of interest should be on left and appropriately highlighted
— Repetitive sequences, trinucleotides and expansion numbers are written with affected allele first and placed in parentheses
— If mutation known, identify in parentheses

Definition	Symbol	Scenario
1. Documented evaluation (*) Use only if examined/evaluated by you or your research/clinical team or if the outside evaluation has been reviewed and verified.		Woman with negative echocardiogram.  E– (echo)
2. Carrier—not likely to manifest disease regardless of inheritance pattern		Male carrier of Tay-Sachs disease by patient report (* not used because results not verified). 
3. Asymptomatic/presymptomatic carrier—clinically unaffected at this time but could later exhibit symptoms		Woman age 25 with negative mammogram and positive BRCA1 DNA test.  25 y E_1– (mammogram) E_2+(5385insC BRCA1)
4. Uninformative study (u)	 Eu	Man age 25 with normal physical exam and uninformative DNA test for Huntington disease (E_2).  25 y E_1– (physical exam) E_2u (36n/18n)
5. Affected individual with positive evaluation (E+)	 E+	Individual with cystic fibrosis and positive mutation study; only one mutation has currently been identified.  E+(ΔF508) Eu E+(ΔF508/u)
		10 week male fetus with a trisomy 18 karyotype.  10wk E+(CVS) 47, XY,+18

currently unaffected and tested positive for a condition and who can develop the disease (e.g., a person testing positive for a BRCA2 gene mutation who has no cancer at the time of evaluation). Some practitioners use a dot in the middle of the symbol for a heterozygous carrier of an x-linked disorder and a symbol shaded in half for a heterozygous carrier of an autosomal recessive disorder. We continue to support denoting carrier status based on phenotypic expression of the disease and not based on pattern of inheritance.

The Challenge of Incorporating the Pedigree in the Electronic Medical Record (EMR)

The NSGC PSTF recommendations provide a clear and concise pedigree nomenclature that is widely but not uniformly used. No alternative comprehensive symbolization recommendations have been proposed nor did we identify literature or professional provider organizations refuting the NSGC PSTF recommendations. With this in mind, we would encourage manufacturers developing EMR systems to consider the inclusion of the standardized pedigree nomenclature within their products.

The family health information contained within EMRs gives rise to new challenges because medical records may be accessed to varying degrees by a variety of different people (e.g., the patient, health care providers, hospital administration, etc.). Therefore, protocols should be developed to identify who can access pedigree information, and what information to include on the pedigree. During the spring of 2007, the Washington State Department of Health Genetic Services Section and the University of Washington Medical Center's Division of Medical Genetics convened a series of meetings designed to explore these issues (Bennett et al. 2007). The meeting participants included medical

geneticists and genetic counselors, consumers, researchers, information technology staff, primary care providers, HIPAA compliance officers, and health administrators. Samples of anonymous pedigrees taken from the regional genetics clinics in Washington State were examined to determine the content of information on the pedigree and why it was obtained. While few regional genetic clinics were utilizing electronic medical records, this exercise set the stage for discussing the potential risks, benefits and limitations of incorporating pedigrees within EMRs.

The conclusions from these meetings were that the elements of a pedigree are consistently recorded for the overall purposes of: 1. *orientation*, 2. *risk assessment*, 3. *ease of reading by multiple users*, 4. *validation* 5. *accountability* 6. *education/health promotion and intervention*, and 7. *communication* (Bennett 1999; Bennett et al. 2007). To expand on the above categories, pedigree elements related to issues of *orientation* refer to the user's understanding of who is the consultand or proband, understanding the genetic and biologic relationships within the documented family, knowing when the pedigree was obtained or the information updated, who provided and recorded the information, and why the pedigree was being constructed. Pedigree elements relevant for *risk assessment* include knowing who on the pedigree is affected or unaffected, relationship lines depicting the biologic relationships and degrees of relatedness (including consanguinity), ancestry, ages of relatives including age at disease onset, and other pertinent healthcare information. Elements such as using the standardized symbols and including legends or keys are included for *ease of reading by multiple users*. Knowing whether the information provided was actually *validated*, for example, by reviewing medical records or death certificates, is important for *risk assessment* and *health intervention*. Information is also recorded on a pedigree to meet *documentation and accountability demands* for medical-legal standards. Finally, information on a pedigree is used for *education* of the patient and to develop plans for health interventions and *health promotion*.

An over-riding principle is the use of a pedigree for *communication* between the health professional and the patient, communication with other health professionals, and potentially sharing the pedigree with relatives (Bennett 1999; Bennett et al. 2007). There is a delicate balance between recording enough information to make a pedigree useful and including so much information that the pedigree can no longer be quickly and concisely interpreted. The pedigree's utility lies in its ability to simply and graphically depict complex information so that disease patterns, risks, and biological relationships are immediately and obviously apparent. If the pedigree becomes cluttered with irrelevant information, its usefulness is diminished.

In a health care environment with EMRs, one can imagine that a pedigree will be revised throughout the lifetime of a patient when he or she faces new age-related health risks. For example, the familial disorders that should be tracked for a newborn are different than for an adolescent, a pregnant woman, or a male over age 60 (Bennett 1999). As different providers obtain or update the pedigree, they may direct their inquiries about the family health history in targeted ways. For example, an obstetrician may ask questions to determine risk factors for the fetus, while the same patient in later years may see a cardiologist whose questions of her family health history would clearly be focused on cardiovascular disease risks. The fill patterns in a pedigree that are identified in the key are likely to change as different disorders arise in the person's family and as the number of relatives increase in size and age. Ideally, to reduce such pedigree "clutter," an EMR would allow the viewer with access to the pedigree information to eliminate or filter excess information not currently relevant to the patient's care (e.g., the cardiologist could omit elements of the pedigree obtained during the obstetrical visit that are not relevant to the patient's current concern of cardiovascular disease).

Privacy and Confidentiality

It's also important to recognize that pedigrees document sensitive information that should be collected and maintained with the utmost protection for privacy and confidentiality. Individual pedigree symbols have low information content, but when they are formatted into a pedigree, there is a new representation of the family and the individual's relation to it. The previously independent data become a collective work making "invisible knowledge" visible to the patient, family, and potentially the scientific community (Bennett 2000; Nukaga and Cambrosio 1997). The PSWG suggests that in non-published pedigrees, initials or first names (in lieu of full names) may be enough to identify persons where medical records are documented and for orientation of discussions with the patient. Use of birth year or age, year of death or age at death, rather than birth date or date of death, would be compliant with HIPAA guidelines where exact dates are viewed as private and protected information.

Summary

The NSGC PSTF recommendations provide a clear and concise pedigree nomenclature that is widely but not uniformly used. No alternative comprehensive symbolization recommendations have been proposed. Although we did not perform an exhaustive search of the literature regarding the

use of the PSTF nomenclature, or look at the website of every professional society whose members might use a pedigree in clinical practice; we found no evidence of a trend to refute the NSGC PSTF recommendations.

The pedigree nomenclature published by the National Society of Genetic Counselors in 1995 is becoming an international standard. Upon review, we propose a few stylistic changes only. The century-old adage of Francis Galton (1889) that "there are many methods of drawing pedigrees and describing kinship, but for my own purposes I still prefer those that I designed myself," remains an issue today. Emphasis on using standardized pedigree nomenclature should continue in genetic education, documentation of medical records, genetic research, and in publication. Use of this nomenclature has the potential to reduce medical error and ease communication among health professionals, patients and their families regarding genetic diagnosis and testing. It also has the potential to save money by documenting prior testing so that additional diagnostic or susceptibility genetic testing can be approached in a cost-efficient manner (e.g., testing for a known mutation instead of full sequencing, or offering an expanded panel of genetic testing that might not have existed at the time a relative was tested, etc.). The use of this nomenclature should be reviewed periodically to assure that it is sensitive to changing legal requirements, responsive to changing medical and testing technologies, successfully incorporated into electronic medical records, and always respectful of patient confidentiality. A simple litmus test in documentation of a pedigree should be: would you feel comfortable handing this pedigree to your patient, or would you be willing to have this pedigree published if it was your family?

Acknowledgements The pedigree symbols and abbreviations are revised with permission from Cell Press and were originally published in Bennett, R. L. et al. (1995). Recommendations for standardized pedigree nomenclature. American Journal of Human Genetics, 56, 745–752. Meetings and conference calls of the PSWG were supported by grants from the National Society of Genetic Counselors, the University of Washington Division of Medical Genetics. The Mamafest gift fund of University of Washington Division of Medical Genetics provided support for graphic artist Christine Symonds. The Board of Directors of the National Society of Genetic Counselors reviewed and approved the revisions to the pedigree nomenclature on October 16, 2007. The pedigree symbols in the figures were formatted using Adobe InDesign CS2. None of the authors received financial compensation for their work. The meetings regarding Core Elements of the Pedigree in the Electronic Medical Record were supported in part by Project # H61MC00219 from the Maternal and Child Health Bureau (Title V, Social Security Act), Health Resources and Services Administration, Department of Health and Human Services.

References

Adult Society.com. Retrieved 10-16-2007, from http://www.adult-society.com/transgender.html.

American Medical Association (2007). *AMA manual of style: a guide for authors and editors* (pp. 660–666, 10th ed.). New York: Oxford University Press.

American Society of Clinical Oncologists (2004). *ASCO curriculum: Cancer genetics and cancer predisposition* (pp. 3–7, 2nd ed.). Alexandria, VA: ASCO.

Bennett, R. L. (1999). *The practical guide to the genetic family history*. New York: Wiley.

Bennett, R. L. (2000). Pedigree parables. *Clinical Genetics*, *58*, 241–249 doi:10.1034/j.1399-0004.2000.580401.x.

Bennett, R. L. (2006a). Genetic counseling. In M. S. Runge, & W. C. Patterson (Eds.), *Principles of molecular medicine* (pp. 46–52, 2nd ed.). Totowa, New Jersey: Humana Press.

Bennett, R. L. (2006b). Genetic counseling. In D. R. Lynch (Ed.). *Neurogenetics: scientific and clinical advances* (pp. 32–34). New York: Taylor Francis.

Bennett, R., Lochner Doyle, D., Harrison, T., Byers, P. (2007). Core elements of the pedigree in the era of electronic medical records. *Journal of Genetic Counseling, 16,* 687 Cooper, S. and Austin, J., Presented Abstracts from the twenty-sixth annual education conference of the National Society of Genetic Counselors, Kansas City, Missouri, October 2007. doi:10.1007/s10897-007-9100-9

Bennett, R. L., Steinhaus, K. A., Uhrich, S. B., & O'Sullivan, C. (1993). The need for developing standardized family pedigree nomenclature. *Journal of Genetic Counseling*, *2*, 261–273. doi:10.1007/BF00961575.

Bennett, R. L., Steinhaus, K. A., Uhrich, S. B., O'Sullivan, C., Resta, R. G., Doyle, D. L., et al. (1995). Recommendations for standardized pedigree nomenclature. *American Journal of Human Genetics*, *56*, 745–752.

Botkin, J. R., McMahon, W. M., Smith, K. R., & Nash, J. E. (1998). Privacy and confidentiality in the publication of pedigrees: a survey of investigators and biomedical journals. *Journal of the American Medical Association*, *279*, 1808–1812 doi:10.1001/jama.279.22.1808.

Byers, P. H., & Ashkenas, J. (1998). Pedigree—publish? Or perish the thought? *American Journal of Human Genetics*, *63*, 678–681 doi:10.1086/302033.

Firth, H. V., Hurst, J. A., & Hall, J. G. (2005). *Oxford desk reference: clinical genetics* (p. 674). New York: Oxford University Press.

Galton, F. (1889). *Natural inheritance* (p. 249). London: Macmillian.

Genetic Nursing Credentialing Commission. Retrieved 10-16-2007, from http://www.geneticnurse.org [home].

Jenkins, J., & Calzone, K. A. (2007). Establishing the essential nursing competencies for genetics and genomics. *Journal of Nursing Scholarship*, *39*, 10–16 doi:10.1111/j.1547-5069.2007.00137.x.

Kingston, H. M. (2007). Genetic Assessment and pedigree analysis. In D. L. Rimoin, J. M. Connor, R. E. Pyeritz, & B. R. Korf (Eds.), *Emery and Rimoin's principles and practice of medical genetics* (pp. 520–522, 5th ed.). Philadelphia: Elsevier.

LeRoy, B. S., & Walker, A. P. (2002). Genetic counseling: history, risk assessment, strategies and ethical considerations. In R. A. King, J. Rotter, & A. H. Motulsky (Eds.), *The genetic bases of common disease* (pp. 87–101, 2nd ed.). New York: Oxford University Press.

National Coalition of Health Professionals Education in Genetics (NCHPEG) (1995). Core competencies in genetics for all health professionals. Retrieved 10-16-2007, from http://www.nchpeg.org/core/corecomps2005.pdf.

Nukaga, Y., & Cambrosio, A. (1997). Medical pedigrees and the visual production of family disease in Canadian and Japanese genetic counselling practice. In M. Elston (Ed.). *The sociology of medical science & technology* (pp. 29–55). Malden, MA: Blackwell.

Nussbaum, R. L., McInnes, R. R., & Willard, H. F. (2007). *Thompson and Thompson, genetics in medicine* (pp. 117–118, 7th ed.). Philadelphia: Saunders Elsevier.

Offitt, K., Groeger, E., Turner, S., Wadsworth, B. A., & Weiser, M. A. (2004). The "duty to warn" a patient's family members about hereditary disease risk. *Journal of the American Medical Association*, *292*, 1469–1473 doi:10.1001/jama.292.12.1469.

Preview, S.C.O.P.U.S. Retrieved 10-16-2007, from http://www.scopus.com.

Resta, R. G. (1993). The crane's foot—the rise of the pedigree in human genetics. *Journal of Genetic Counseling*, *2*, 235–260. doi:10.1007/BF00961574.

Schuette, J. L., & Bennett, R. L. (1998). Lessons in history: obtaining the family history and constructing a pedigree. In D. L. Baker, J. L. Schuette, & W. R. Uhlmann (Eds.), Lessons in history: obtaining the family history and constructing a pedigree *A guide to genetic counseling* (pp. 27–54). New York: Wiley–Liss.

Steinhaus, K. A., Bennett, R. L., Uhrich, S. B., Resta, R. G., Doyle, D. L., Markel, D., et al. (1995). Inconsistencies in pedigree nomenclature in human genetics publications: a need for standardization. *American Journal of Medical Genetics*, *56*, 291–295 doi:10.1002/ajmg.1320560314.

Turnpenny, P., & Ellard, S. (2007). *Emery's elements of medical genetics* (p. 104, 13th ed.). Churchill Livingston: Elsevier.

Vilain, E., Achermann, J. C., Eugster, E. A., Harley, V. R., Morel, Y., Wilson, J. D., et al. (2007). We used to call them hermaphrodites. *Genetics in Medicine*, *9*, 67–73 doi:10.1097/GIM.0b013e31802cffcf.