The Role of the Jewish Genealogist In Medical and Genetic Family History

by Stanley M. Diamond

U.S. Surgeon General Richard H. Carmona notes that "Knowing your family history can save your life. Millions of dollars in medical research, equipment and knowledge can't give us the information provided by this simple tool. When a health care professional is equipped with a patient's family health history, he or she can easily assess the inherent risk factors and, in some instances, begin tests or treatment even before any disease is evident."*

Genealogists not only have been documenting their family histories, but have become the repository of vital medical and genetic history for their families. With the advent of widely available genetic testing, the giant leaps in disease identification, the dramatic growth of DNA databanks, the introduction of umbilical cord blood storage and the new science of gene replacement therapy, genealogists have been given an increased opportunity—and responsibility—to contribute to both their own family's personal health and that of future generations.

The growing mass of genealogically related data available online has expanded and enriched our ability to find and connect with previously unknown family members. With family history research easier than ever, we genealogists now have previously unimagined opportunities to compile our medical and genetic data and, in tandem with our family trees, use this information to enhance the health of the people in our families. We can now use our genealogy and genetics to make a difference in the world.

Jewish Genetic Diseases: Reality and Responsibility

Thousands of known genetic diseases afflict the world's population. In almost every ethnic or racial group, however, certain genetic diseases occur at higher frequencies among their members than in the general population. Such is the case for the Jewish people. Many of these diseases are severely incapacitating and some are tragically debilitating, leading to death in infancy or early childhood. Tay-Sachs is one of the most notorious of the lot, but other diseases and genetic predispositions to diseases, just as prevalent and just as devastating, shatter the lives of Jewish families.

If called upon, all genealogists have an obligation to play an investigative and advisory role in their families, and when a genetic trait is discovered, we have an obligation to reach out and warn extended family members that they may be at risk. I hope my 15-year quest to document the incidence of the gene for Beta-Thalassemia disease, an inherited blood disorder causing mild to severe anemia, in my extended family will be a model for all genealogists and family historians dedicated to recording their own families' genetic and medical history.

Even after archive doors open wide for research with life-saving potential, the challenge remains to persuade previously unknown family members to be documented and to convince close family members to cooperate with research. We must combat resistance of whatever cause—embarrassment, fear, laziness or ignorance.

Combining genetic, medical and genealogical research involves different methods and special responsibilities. Defining this philosophy, formulating the message and honing sensitivities are unusual challenges for casual genealogists.

Role of Genetic/Medical Research

Some family historians want to know about all the generations of musicians or scholars, and some want to track every family story that has been passed down through the generations. But today there is growing recognition among genealogists of the need also to provide detailed information in the medical field in your genealogical computer program or use specialized family medical history software such as *Geneweaver*, produced by Genes & Things, Inc.

The American Medical Association recommends that every family maintain a family health history. Recording one's personal and family medical history is becoming the norm rather than the exception. The ability to provide ready access to this information to an extended family can be of great and often unanticipated benefit. This is particularly so in the case of recessive disorders, where a single altered copy of a gene inherited from *both* parents may result in devastating consequences for the next generation, such as Tay Sachs disease and Thalassemia Major, to name two examples.

When a recessive disorder first occurs in a family, it appears to come from nowhere—but that typically is not the case. More likely, the recessive gene had been passed down silently for many generations. What makes recessive conditions appear so obscure is that inheriting a single copy of an altered recessive gene rarely causes medical complications. It typically remains quiet and does not reveal itself until two people, who both carry a single copy of the same altered gene, happen to have a child together. Consequently, once any genetic disorder—particularly a recessive condition is identified, genealogists have an *obligation* to reach out and caution family members that this gene is hiding in their family.

Fortunately, because of the joint efforts of the medical, genetics and religious communities and the ease and speed of modern-day communications, it now is possible to identify individuals and even pregnancies at risk for a number of recessive conditions. On the other hand, since the majority of recessive conditions are rare, not as well publicized, and are not associated with any specific ethnic group, a

routine public health genetic screening network usually does not exist. In this situation, typically vigilant genetic counselors and doctors have no red flag to alert them to the potential danger of a rare recessive gene that, unbeknownst to the family, has been passed down from generation to generation. Therefore, populations not considered at risk for certain disorders do not benefit from routine screening and discovery as is found with the Ashkenazic genetic conditions mentioned above.

In my case, family members either were unaware they carried a single copy of the altered beta-thalassemia gene, also referred to as "trait," or only learned about it serendipitously later in life, usually when undergoing exhaustive tests for other medical conditions. This has been the norm rather than the exception, and, unfortunately the knowledge often came too late to provide much-needed information to other family members, who in turn might have had a child with the devastating Beta-Thalassemia disease. Carriers often were misdiagnosed as being just plain anemic, without specific differentiation. As a result, these people often inadvertently were prescribed ineffective medication, typically iron, suitable only for other forms of anemia. Recognizing the potential existence of hundreds of unsuspecting carriers in distant branches—and that this trait is virtually unknown among Ashkenazim—is what drives my genealogical/genetic research project.

Genealogists must be aware that genetic science is only approaching the threshold of the re-engineering of disease-causing genes. That is why genetic counseling and prenatal testing have played such an important role in raising awareness and helping people understand their chances of being affected with genetic diseases as well as how to deal with this knowledge. Directing family members to medical professionals who are trained to communicate the appropriate information is the responsibility of every genealogist who charts his or her family's history for medical or genetic reasons.

Researching Your Family's Medical/Genetic History

Genealogists who ask medical history-related questions—whether of long-known or recently discovered relatives—soon realize they may be treading on delicate ground. Whether the information sought is general (i.e., just to fill in the cause-of-death field in a genealogical software program) or very specific, one often hears, "Why do you want to know?" While the question "why" may be the first one heard, the researcher's response must also address "who," "when," "where" and "how"—all the while being both cognizant of the sensitive issues and prepared to allay the concerns of a reluctant relative.

Why Do You Want to Know?

Whether it is simply asking for names, dates and places—the staples of genealogical research or medical-related questions, your family members will ask, "Why are you doing this?" Many excellent articles and hundreds of posts on the JewishGen mailing list detail reasons why individuals become fascinated with family history research. Answering the question "why?" when it involves medical matters presents special challenges. It has been said, "Ask the family gossip a medical question and the answer may be never-ending or dead silence." Therefore, be prepared to give a direct, carefully crafted answer—one that invites cooperation as opposed to a "never bother me again."

Family historians should:

• Define the objectives of your medical/genetic research project (your "mission statement")

• Be able to clearly explain what you are doing and why you are asking questions.

• Understand and be able to communicate the basic facts of the medical condition or genetic trait that is the basis for your reaching out. If you are gathering general information, ask questions about common concerns, such as heart disease, stroke, diabetes or Alzheimer's. Prepare clear and concise documentation that can be used to follow up verbal communications. Provide references to reading material and/or Internet websites for those who want to learn more.

• Outline the benefits of your research to all members of your family and their future generations. Use terms they understand—"life-saving" or "early diagnosis."

• Detail what you expect to do with the information you gather and how it would be communicated to family members or shared with the medical community which might find the data of scientific value. Explain what you will *not* do, such as making the information generally available on a website.

Whom To Talk To

Every member of each branch of a target family should be tapped for relevant information. "The left hand doesn't know what the right hand is doing" may aptly describe what one nuclear family knows and another does not about the circumstances of Zayde's death. When it involves a genetic trait, not every family member will be comfortable sharing all the details—or even mentioning it. Therefore, researching your family's medical history and making a genetic tree requires talking to everyone, frequently more than once. Researchers must:

• Focus on those branches and individuals who may be able to provide the key leads for expanding the search.

• Never assume anything! People often do not know or cannot remember their exact medical condition. Check and double-check. Ask for permission to talk to family doctors or anyone who has been involved with the health of the family. This is particularly relevant when tracking genetic traits that can be a potential disaster for future generations, because not everyone will recognize the implications of the trait they carry: Two carriers of a recessive gene have a one-in-four chance of producing an affected child.

• Enlist others in the family, particularly doctors and other medical professionals who understand and support the aims of the research. Ask them to join your team.

• Keep the family up-to-date on your research, breakthroughs and plans. This will keep them involved and encourage them to help.

When to Start; When to Push

We are told time and again, "Interview the living! The documents will be here forever." The two words genealogists dislike intensely are "if only." If only I had listened to Bubbe when she talked about her youth…If only I had written down the endless stories my father used to tell about his grandparents…If only mother hadn't thrown out Zayde's old address book or diary or _____ (fill in the blank!). It is no different for the family historian who seeks to record his or her family's medical history. Remember when Mom or Dad came home from visiting Uncle Sam at the hospital and described his strange condition. I wasn't listening, were you? Talk to the older generations *now!* Even if a death certificate states "arterial sclerosis," you should be asking questions: "Was this a heart attack? When did Zayde first get sick? Was it his first heart attack? Did he die suddenly or did he go to the hospital?" Ask about and record the circumstances.

How to Find the Answers

Face-to-face meetings are always best. They inspire confidence. The expressions on your face and the sound of your voice show that you really care. However, genealogists know researching family history entails more than a drive around town. Our ancestors settled all over the world, and our modern families have spread with the winds. It is true that Internet resources, the advent of e-mail and low-cost long-distance rates have significantly simplified the search process and facilitated communications, but unless we are exceptionally skilled, the printed word can seem unfeeling when asking sensitive medical questions. Because your genuine concern and interest may not come through in written material, the telephone call is indeed "the next best thing to being there."

The first goal must be to gain the confidence of the person you are calling, often someone who may never have heard of you or your branch of the family. Even the words to be left on an answering machine should be considered carefully in advance. The response can be all the way from a demanding "how did you find me?" to "I am so glad you called!" Establishing credibility with someone you are calling or writing for the first time is a must. And after you have spoken to an older member of the family, follow up immediately with a son or daughter. Children are usually protective of the elderly parents and may be suspicious of strangers asking mom and dad many seemingly personal questions.

When phone calls are impractical (because of old family feuds, language barriers, etc.), the reaching-out letter must be clear, concise and effective; having it co-signed by other family members and/or a doctor can enhance its credibility and is recommended. When you have someone make a call for you—to speak to your new-found cousin in his or her native language —try to be next to that person so you can give immediate follow-up answers. This approach makes the call more personal and helps preclude the feeling that the call has left you with more questions instead of the answers you sought.

Short Guide to Interviewing for Medical/Genetic Family History

• Explain who you are, where you live and how you obtained your family member's name.

• Convey in a few short sentences why you have an interest in the family's history.

• Describe your exact relationship or what you think the relationship may be. Articulate it in terms that a non-genealogist can understand. "Third cousin, once removed" is likely to bring silence. But, "My grandfather and your great-grandmother were sister and brother" is far easier to grasp.

•Share your family history: Tell the story of your branch and show a general interest in theirs, where they live and how they got there. Offer to send a "family tree," but avoid providing details as to whether it will be a graphic tree, a descendant's list or other report; that can be confusing to non-genealogists. Share a vignette about a common ancestor or living relative, one that will make a person proud or provide a laugh.

• Avoid the turn-off: Most people are flattered to be asked non-leading questions about their history and unique accomplishments, but it could be counterproductive to rush into discussions about college degrees or well-kept family secrets about mental illnesses or suicides. Allow the conversation to evolve. Avoid applying pressure. Do not try to get all the information in one telephone conversation.

• Define your role as the family (medical/genetic) historian: If you are the first person to call about the family, then—in their eyes—you become a special person to be befriended *or* feared. You will be *the* family historian by default. People want to be cast in a favorable light. Listen, take notes, ask questions, take more notes!

• Elaborate about yourself as an individual: Describe where your family history studies have taken you, whether it be to ancestral towns, the Family History Library in Salt Lake City or visits with branches you just discovered. Your deep interest and sincere effort will be recognized. If articles about your research have appeared in magazines and newspapers, send copies. If you have a website, suggest that the person look at it and provide the URL; but refrain from put-downs if they are not computerized or don't have ready access to the Internet!

• Carefully pose the medical/genetic question: How you say it and what you say should be tailored to your own comfort level and the nature of the reaction. One example might be: "You know, because of my study of *our* family, I

hear as many questions as I ask. It seems everyone is curious about one thing or another, and I now seem to be the one with some answers. Health preoccupies all of our older relatives, and that has taught me a lot. For instance, were you aware that Grandma and almost all her siblings had heart disease? That made me curious, and I found that their father's death certificate showed heart disease, too. I guess that's a signal for us. What's the heart situation in your family?"

The question I usually pose is: "Has there been any sign of mild chronic anemia in your family?" By way of follow-up, the comment is: "Well, we seem to be rather special. We are one of only 15 Ashkenazic families carrying a genetic trait called beta-thalassemia or Mediterranean anemia." Often, by the time I get that out of my mouth, the questions come rapidly: "What does that mean?" or "Is it dangerous?" or "How do I know if I am a carrier?" That's when the calming words and clear statements are needed. My reply is: "It has no effect on carriers. I know because I am one. But there is a significance to future generations, because two carriers have a one-in-four chance of having an affected child." The discussion goes on from there, and I quickly point out that I am not a medical person, but merely someone with a deep interest in the medical and genetic history of our family

How a Medical/Genetic Focus Differs From Typical Family History Project

Several important aspects of genealogical research with a medical/genetic focus set it apart from typical family history projects.

• Potential for networking. There are more receptive ears—everywhere, both within and outside the genealogical community, particularly when it involves potential life-saving situations.

• Response of the genealogical community. Genealogists probably are the most generous individuals one can find in any walk of life. When humanitarian activities are involved, the level of response from fellow researchers eager to help can be astounding.

• Reaction of archival resources. Whether at home or abroad, archivists' attitudes can vary from being highly cooperative to passionately supportive. Invite the archivist to be part of your research team! As executive director of Jewish Records Indexing-Poland, I am learning that many of our fellow genealogists are researching for medical/genetic reasons. In almost every case, support from the Polish State Archives and managers of civil records offices in Polish towns has been exemplary.

•Foundations or organizations with an interest in the medical or genetic condition in your family have the experience and materials to help you convey effectively the importance of your mission. (Example: National Organization for Rare Disorders [NORD], WWW.RAREDISEASES.ORG).

• Support from the non-genealogical community. Newspapers want stories; doctors and scientists welcome the opportunity to share their expertise or learn from unique studies; universities seek projects that address the need of students to learn while at the same time making meaningful contributions to the outside world. The Jewish Genetic Disease mailing list HTTP://HEALTH.GROUPS.YAHOO.COM/GROUP/GAUCHERDISEASE/ is a forum for networking with both medical professionals and those at risk or living with genetic diseases.

• Credibility factor: Because it involves the health of both living family and future generations, your family history project should rightly give your research an enhanced level of credibility. This is not automatic. It takes time, effort and patience to bring all the pieces together.

Documentation, Confidentiality, Perpetuity

Whatever the reasons for charting your family's medical and genetic history, confidentiality must be respected; permission is necessary to share information. In the U.S., where it often seems that medical insurers are looking over everyone's shoulder, there is a particular need for prudence. I maintain a separate confidential family tree of carriers of the trait. Finally, decide to whom you will pass on your valuable research and under what conditions.

Recommended Further Reading.

Bennett, Robin L. and M.S. Bennett, *The Practical Guide to the Genetic Family History,* New York: John Wiley and Sons, Inc., 1999.

McNabb, Luanne, Curtis, Elizabeth Curtis and B.A. Barclay-Rowley, *Family Health Trees*, Toronto: Ontario Genealogical Society, 1997.

Nelson-Anderson, Danette L., R.N., B.S.N., and Waters, Cynthia V., *Genetic Connections: A Guide to Documenting Your Individual and Family Health History,* Sonters Publishing, P.O. Box 109, Washington, MO 63090-0109, 1995.

Willard, Jim and Terry, with Jane Wilson, *Ancestors: A Beginner's Guide to Family History and Genealogy,* Houghton Mifflin, Boston, 1997 (see chapter 8: Your Medical Heritage, pages 89-102.).

Note

* Carmona, Richard H, *M.D., M.P.H., F.A.C.S.,* Surgeon General of the United States Public Health Service, *U.S. Medicine, January, 2005:* "PHS Uses Education As Preventive Medicine."

Stanley M. Diamond, winner of the 2002 IAJGS Lifetime Achievement Award, is founding president of the Jewish Genealogical Society of Montreal and executive director of Jewish Records Indexing-Poland (JRI-Poland). His interest in genealogical research related to genetics ultimately led to the creation of JRI-Poland.