

American Society of Human Genetics
Ad Hoc Committee on Consumer Issues

COMMITTEE REPORT

Genetic Lay Advocacy Groups: Significant Others in the Conduct of
Human Genetics Research

October, 1999

(Revised 11/10/00)

(Revised 10/18/01)

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Introduction

The Boards of the American Society of Human Genetics (ASHG) and the Genetic Alliance established the Ad Hoc Committee on Consumer Issues in 1998. The main purpose of the Committee is to open a dialogue between the communities of human geneticists and genetic support groups. The first activity of the Committee consisted of co-sponsoring a workshop designed to examine the conduct of human genetics research. The workshop participants included former and present members of the Board of Directors of both organizations. The workshop was held in Baltimore on June 4-6, 1999. The participants identified various themes related to the conduct of genetics research and proposed that a new *model of relationship* between the two communities has been emerging in recent years. The participants encouraged the advancement of this model in coming years. The essence of this model is encompassed in the following *consensus statement* developed during the meeting by the workshop participants:

Genetic Lay Advocacy Groups: Significant Others In the Conduct Of Human Genetics Research

Research in Human Genetics is a shared enterprise that involves investigators, human participants, and ancillary agencies, including funding organizations and regulatory boards. The cultures of these groups and organizations often differ, and these differences create a dynamic tension between shared and divergent interests. When divergent interests interfere with recruitment of subjects to research and compromise mutual understanding of the goals of both parties, it is important to identify mechanisms to create or recognize shared goals. **Genetic lay advocacy groups are an important agency in this enterprise.** These groups have diverse objectives: creation of community, advancement of education for members and professionals, and sponsorship and encouragement of research. To the extent that successful lay advocacy groups have created active involvement in research, their activities provide models for future collaborative relationships. The key activities at the level of the lay advocacy group include education of members about expectations for research, elements of the consent process, and the nature of research as well as education of investigators with respect to many of these same elements. Lay advocacy groups can best broker the successful relationships between investigators and participants where the *cultural differences* are clearly delineated. As the Human Genome Project reaches its objective of a complete sequence and the emphasis in Human Genetics research evolves from gene finding to characterization of disease mechanisms, the role of lay advocacy groups in recruitment of participants will be increasingly important. The next change i.e. to search for successful therapies, will require an even more active interaction of investigators, participants, lay advocacy groups, and other agencies to recruit the participants and to explain the objectives of the research process. **Active collaboration** among investigators, participants, and lay advocacy groups will be the catalyst for future research productivity in human genetic disorders.

Historical Background

In 1997 at the ASHG meetings in Baltimore Maryland, selected members of the ASHG Board of Directors and of the Genetic Alliance of (herein referred to as the Alliance) met with the purpose of opening a dialogue between the two communities. This particular meeting was chaired by Diane Baker, then a member of the Board of Directors of both organizations. While the agenda of the meeting was open ended by design, a specific and recurring theme regarding the conduct of human genetics research emerged. In particular, there appeared to be significant concern from the genetic support group community regarding participation in human genetics research. Issues such as the ownership and availability of tissue samples and less than optimal communication between researchers and subjects in general came up as universal concerns among groups. While a number of positive interactions were also emphasized, there was a sense from the lay advocacy group leadership that there existed a lack of partnership in this enterprise. (The minutes from that particular meeting are available in the Appendix of this document.) There was a sense from the participants of the meeting that this lack of partnership represents a problem. The meeting led to a number of suggested action items and on the top of that list was the recommendation for the ASHG and the Alliance to jointly sponsor a workshop whose purpose was to address this issue. The theme of the ASHG and the Alliance **working together** came out of this meeting and the document that followed. The initial charge of that joint committee was for the workshop participants to generate a statement of *principles* of conduct for researchers and participants.

From a historical background point of view, it also is important to acknowledge that in 1998 at the Denver ASHG meeting the ASHG Board formally recognized this joint committee as an official Ad Hoc committee of ASHG. The co- chairs of the committee (and of the workshop) are John Carey of ASHG and Jannine Cody of the Alliance (also a member of ASHG).

The workshop, held in Baltimore of June 4-6, 1999, addressed this charge. Participants included a number of present ASHG board members, some past members, and the Executive Director and standing board members of the Alliance. In addition, three guests were invited as participants of the meeting because of their unique expertise. These included Allan Sandler, an authority at the NIH on Institutional Review Boards, Benjamin Wilfond, a medical geneticist and bioethicist, and Deborah Heath, a cultural anthropologist (list of the participants is included in the Appendix).

The original schedule of the workshop is also included in the Appendix. Originally the plan was to break the participants into small groups whose purpose was to identify and articulate the key issues surrounding the conduct of human research. However, by the first evening of the Workshop, it was evident that the group was small enough and was working together cohesively, and thus the plan was altered so that the entire committee dealt with the identification of issues together.

The content of the weekend workshop was divided into three areas:

- 1) A presentation of historical overviews of three disciplines (medical genetics research, the ethics of human genetics research, including protection of human subjects and informed consent, and genetic lay advocacy groups).
- 2) The identification of the key themes and issues.
- 3) The development of goals and objectives as a basis for planned action items.

The interesting and exciting discussions that occurred over the two days will be summarized in the remainder of this document. The essence of the proposed model is captured in the consensus statement above.

Issues And Themes Surrounding The Conduct Of Human Genetic Research

During the discussions that followed the presentation of historical overviews, a number of issues and themes surrounding the conduct of human genetics research emerged. These included: the cultural aspects of the research enterprise, including the culture of science, the psychology of why people participate in human research, and the culture of self-help groups. Another significant issue was the unrecognized role of certain lay advocacy groups who have been highly successful in developing collaborations and partnerships with scientists that subsequently led to successful outcomes. An important premise that ensued from the discussion of these themes was the importance of *collaboration* between subjects, including lay advocacy groups, and the genetics research community. The utilization of the term *collaboration* as the center of this relationship underscores the notion that this idea represents a new model for the enterprise of genetic research. **This model emphasizes the role of genetics lay advocacy groups in the human genetics research endeavor and stresses the collaborative nature of the relationship.** It suggests that development of a collaborative relationship will lead to increased productivity. In this way the enterprise of research in human genetics could be depicted as a five point star with the participants at one point and the research scientists, the regulatory agencies, the funding agencies, and genetics lay advocacy groups at the other points.

Other themes that emerged during the discussions included the need to clarify the goals of lay advocacy groups regarding research, and the importance of research participants and lay advocacy group representatives on IRBs and grant review panels. Also, the significance of teaching communication skills to scientists, the need to define the research process to subjects, and the goal of imparting the essence of this model to all genetic professionals were identified.

The four goals and outcome objectives identified by the workshop participants are designed to acknowledge this model and integrate it into the communities of human genetics and support groups. The outcome objectives listed under these goals led to *action items* many of which are currently being carried out by committee members.

Goals and Outcome Objectives

Goal #1: *Promote the collaborative dimensions of the relationship between researchers and subjects.*

Outcome Objective A: Education of Human Genetics researchers about the ways in which lay advocacy groups can facilitate and enhance the research process.

Action Item 1: Write paper regarding success of selected lay advocacy groups. (see below)

Action Item 2: Propose a workshop at the American Society of Human Genetics 2000 on this model.

Action Item 3: Look into sponsoring a workshop at the Clinical Genetics Society March 2000. (not accomplished)

Action Item 4: Write paper on the cultural aspects of the genetics research enterprise. (see below)

Action Item 5: Submit abstract to the American Society of Human Genetics summarizing the consensus statement of this workshop (this abstract was a poster presentation #1051, 1999).

Outcome Objective B: Education of lay advocacy groups about the research process and culture.

Action Item 1: Perform a survey of Alliance members as to the present connections between lay advocacy groups and researchers. (see below)

Action Item 2: Provide training for lay advocacy groups and to present regarding the process of research, including goals and IRB process.

Outcome Objective C: Identify effective approaches to research collaboration and advance the knowledge of the model.

Action Item 1: Provide technical assistance to lay advocacy groups.

Action Item 2: Survey and document existing models (see survey above).

Outcome Objective D: Promote awareness of the Alliance.

Goal #2: *Devise effective means to involve support organizations in the process of translation of research protocols.*

Outcome Objective A: Assess involvement of lay advocacy groups and informed consumers to the genetics research process.

Action Item 1: Above-mentioned survey to genetic lay advocacy groups.

Action Item 2: Survey to ASHG members about their awareness of and their connections with lay advocacy groups.

Action Item 3: Provide technical assistance as mentioned above.

Goal #3: *Emphasize the education and communication components of the informed consent process.*

Outcome Objective A: Make available the interactive CD from the NIH.

Outcome Objective B: Identify similar resources and publicize them.

Outcome Objective C: Develop effective ways of communicating the intent of research to participants, before, during and after process.

Action Item 1: Write an article for publication in the genetics literature focusing on the complexities of relationships in the research process, i.e. the culture of research. (See Action Item under Goal #1 above.)

Goal #4: *Integrate these concepts and this model into the training of those involved in human genetics and research.*

Outcome Objective A: Incorporate this knowledge base into the guidelines for training programs *for all* medical genetics professionals.

Action Item 1: Contact the RRC, the American Board of Medical Genetics, the American Board of Genetic Counseling, the Canadian Colleges, and the Professors of Human and Medical Genetics groups regarding this recommendation.

Action Item 2: Incorporate this into the NIH guidelines for training of junior scientists with NIH funding.

Summary of Action Items Planned By Workshop

In summary, the most crucial action items that were recommended and are currently being carried out include:

- 1) Survey to Alliance members in regards to existing participation of lay advocacy groups in research. (completed 1999; new survey planned for 2002)
- 2) Survey of ASHG members regarding their utilization of lay advocacy groups in carrying out research (completed November 2000, summary presented to Board October 2001; data currently being analyzed and this will be the subject of another report).
- 3) A peer reviewed paper possibly in the American Journal of Human Genetics on the successful lay advocacy groups that have sponsored and led research (paper published in American Journal of Medical Genetics, Seminar Series 2001, Researching the biology of PXE: partnering in the process.106:177-184.)
- 4) A paper on the cultural aspects of the conduct of human research. (planned 2001-2002)
- 5) Abstracts presented to the ASHG meeting in San Francisco on this model (accomplished at ASHG meetings 1999, 2000, 2001 poster presentations).
- 6) A presentation of a workshop at ASHG Annual Meetings 2000-2002 on this model (submitted for 1999, 2000 – not accepted as an invited presentation; a proposal was submitted for 2002).

Conclusions and Recommendations

The meetings of Board members of the Society and the Alliance over recent years led to the conclusion that there exists a perceived problem by the genetic lay advocacy group community regarding the conduct of human genetics research. The principal aspect of this identified problem consists of the view that there exists a lack of partnership between researchers, participants and lay advocacy groups.

The various players in the research enterprise came from different cultures and commonly have different agendas. Acknowledgement of their differences could help in resolving tensions. Further investigation into the psychology and anthropology of research participants, particularly human genetic research, is needed.

We are proposing a new model of relationship. This proposal promotes a partnership between researcher, research participants, and genetic lay advocacy groups. The support groups are in the position to help foster a productive relationship between research participants and scientist.

We would suggest an educational process that involves both communities: education of the lay advocacy group community in the research process and education of the research community on the benefits of having a connection with support groups.

Acknowledgment

The committee participants and chairs would like to gratefully acknowledge and express their sincere appreciation to Jane Salomon for her incredible support and resourcefulness in orchestrating the meetings, as well as all of the follow up items. In particular, typing the transcript deserves much more than a simple thanks.