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# The Role of Patient Advocacy Organizations in Shaping Genomic Science

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## Abstract

Patient advocacy organizations (PAOs) are nonprofit groups that represent patients and families affected by a significant medical condition or disease. We review some of the different approaches that humanities and social researchers use to study PAOs. Drawing on this recent scholarship, we describe some contemporary patient groups and explore how PAOs can collaborate with biomedical researchers to advance genomic science. We highlight research that aims to describe how PAOs are contributing to multiple aspects of biomedical research, including study design, definition of research goals, data collection and analysis, dissemination of results, and research funding. We also describe several challenges that genomic researchers may encounter in collaborations with PAOs. Throughout our review, we focus on the manner in which new PAO roles challenge traditional boundaries between researchers and subjects, thereby redefining the relationship of patients to science. We consider how this shift may affect our view of scientific collaborations and impact genomic researchers in the future.

## INTRODUCTION

Patient advocacy organizations (PAOs) have emerged as key actors in biomedical science and pharmaceutical research and development (R&D). With advances in genetic sequencing technologies and growing federal commitments to translational research on rare diseases, PAOs will likely play increasingly significant roles in genomic science. In this evolving environment, traditional relationships between PAOs and scientists are being reconceptualized and redefined through a variety of partnerships and collaborations. Through these new relationships, PAOs work to advance science not only by raising public awareness of rare diseases and supporting research through philanthropic efforts, but also by participating in the design and conduct of biomedical research, including translational research that aims to develop new diagnostic tests and therapeutic products.

Many PAOs characterize their efforts as attempts to give patients a greater voice and ensure that patients' interests are acknowledged by those in positions of power. It is in this spirit that many patient advocates sought to participate more directly in biomedical research. Over the past three decades, PAOs have made inroads into all aspects of biomedical research, and there are numerous examples where patient groups have played significant, multidimensional roles in research design and execution, most notably in studies of breast cancer (4), rare diseases (1), and HIV/AIDS (30).

One way to conceptualize the contributions that PAOs make to biomedical research, and to genomic science in particular, is to consider their impact on the relationships between researchers and those who are subjects of research. For researchers, clinicians, patients, and the organizations that represent patients, there are many ways to collaborate. Although some of these collaborative efforts have been studied, much work remains to systematically characterize these collaborations and situate them within the larger contexts of medical institutions, civil society, and organizational theory.

In this review, we provide a map of recent social research on PAOs, honing in on diverse research examining PAOs as a phenomenon, and discuss several issues that are particularly relevant for genomic science. Throughout, we highlight some of the key issues and challenges that genomic researchers and others face in structuring research collaborations that include PAOs. In particular, we focus on the manner in which new PAO roles are blurring traditional boundaries between scientists and subjects, thereby redefining our understanding of the relationship of patients to science. We suggest that the evolving view of this relationship will have a significant impact on how genomic scientists engage patient communities. We also suggest a research agenda that can help to elucidate the nature of these collaborations.

## BACKGROUND: PATIENT ADVOCACY ORGANIZATION CONTRIBUTIONS TO BIOMEDICAL RESEARCH

### Terms and Definitions

There is no universally accepted definition or criterion for determining what groups are PAOs. Although the term is used widely in multiple scholarly literatures, its meaning is often context dependent. Complicating matters, multiple related terms are used for the same organizations, including patient groups and/or associations, consumer groups and/or associations, voluntary health organizations, health consumer groups, and disease advocacy organizations. Although these terms are sometimes used interchangeably, they have different histories, connotations, and associations.

Social scientists consider this lack of a consistent definition to be a major conceptual and methodological problem (2, 7, 14, 87, 92). The decision to use certain terminology and orienting concepts depends on the goals of the specific study. For example, Allsop and colleagues (2) have

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explicitly rejected the term patient group in favor of health consumer group, which they defined as “a voluntary sector organization that is concerned with promoting and/or representing the interests of users and/or carers in the health arena at a national level” (p. 57). Given the substantial diversity of both formal organizations and informal patient networks, there may be no choice but to use ad hoc, study-specific definitions of the relevant groups.

An unfortunate result of this terminological plurality is the difficulty of comparing studies of PAOs with one another. In this review, we use the terms PAO, patient group, and patient association interchangeably except where noted. Our review is also limited to PAOs that focus on single diseases (or a small set of closely related diseases). Many of these organizations are led by a patient, family member, or caregiver. PAO leaders often emphasize the grassroots nature of these organizations and their close ties to patients’ values and needs. Leaders of these organizations frequently cite respect, self-determination, and empowerment in describing their work. Although this appeal to grassroots empowerment is often highlighted in depictions of PAOs, social researchers and historians provide a much more complex and nuanced picture of these organizations. As shown in the case studies and discussion that follow, the past several decades have witnessed not only exponential growth in the number of patient groups and their involvement in biomedical research, but also numerous efforts to “democratize science” and increase its availability to nonspecialists who wish to use it as a tool for effecting social or political change.

Most broadly, PAOs are often understood as nonprofit groups that represent patients and families who are personally managing a significant medical condition or disease (38). Reviewing the history of a specific PAO can provide a better understanding of its mission and structure, but the diversity of these organizations limits the ability to provide generalizations about them as a collective category. Although for analytical purposes it may be possible to create clear demarcations between types of PAOs, the dynamic nature of these organizations may be obscured by simple classifications. For example, some PAOs were formed in partnership with professional organizations and later become wholly patient-run (53), whereas others may have been initiated by the private sector. Attention to historical origins and contextualization aids our understanding of their shifting identities, missions, strategies, organizational structures, and cultures.

### **The Current Landscape as Described by Historians, Social Scientists, and Health Researchers**

Although the contributions of PAOs to science, health policy, and medicine have increased significantly over the past three decades, historians and social scientists have only more recently begun to study their development and impact. Researchers who study PAOs come from a wide array of disciplinary traditions, with some overlap, which necessitates that a review of this literature include studies in bioethics, science and technology studies (STS), medical sociology, and health policy research.

Studies of PAOs have employed a multitude of research methods: single- and multisite ethnographies (19, 26, 41, 63, 65, 67, 71), focus groups (6, 12), semistructured stakeholder interviews (21, 60), in-depth case studies (55, 57), surveys (9, 10, 42), content analysis (69), textual and narrative analysis (28), historical approaches (40), network analysis (15, 62), comparative studies (7, 92), and mixed quantitative and qualitative methods (6, 92). In addition to research, there are also a growing number of personal memoirs and biographical accounts of the work of patient advocates, many of whom have founded PAOs (3, 23, 78, 89). Here, we limit our review to analytic, empirical, and historical studies of PAOs and their contributions to biomedical research.

PAOs vary considerably in size, age, and financial capacity as well as in their missions and goals. Historians and social scientists have demonstrated that national contexts are important to

understanding the development of PAOs and their capacity to contribute to research (13, 26, 59). These scholars have also shown how PAOs in the United States, the United Kingdom, and some western European countries grew out of a complex mix of grassroots health organizations and sociocultural movements that emerged in the 1960s, including environmentalist (16, 17, 31, 43, 56) and consumer-rights (52, 80, 81) movements. Other historians have traced the origins of contemporary PAOs back to the mid-twentieth century and connected them to self-help groups, in which self-determination and patient empowerment were important social drivers. In doing so, these historians showed that some “patient movements” and PAOs—e.g., those related to mental health—were started with the explicit aim of being run by patients committed to helping other patients and their caregivers (39). These historical studies help us understand contemporary patient groups and their evolution over time. In addition, by highlighting the evolution of PAOs in national, institutional, and cultural contexts, these studies help us understand how contemporary stakeholders and PAOs engage each other and approach research collaborations. They also help us anticipate how changes in biomedical research practices may impact patient groups.

Several recent social and health research studies have demonstrated the diversity of PAOs and their contributions to biomedical research (2, 69, 75, 92). Political scientist Brian Wood (92) conducted one of the largest comparative studies of PAOs, or what he referred to as “disease-related patients’ associations.” He found that patient associations are increasing in number and that many PAOs were established after 1980, and also that PAOs are highly diverse in both their organizational structures and their goals. Allsop and colleagues (2) surveyed patient groups in the United Kingdom and found similar variation in these organizations’ structures, missions, and financial capacity.

A 2009 study by researchers in partnership with PAOs in France examined rare-disease associations in Europe and found that many of them ( $n = 309$ , 37% of their sample) had funded research in the preceding five years (75). Of these PAO-supported studies, 70% were initiated by researchers interested in conducting basic research on rare diseases. Although the ways in which PAOs contribute to genetic research in particular have not been well characterized, a recent study by Landy and colleagues (42) highlighted the diversity of roles played by these organizations. In that study, which examined 124 disease advocacy organizations focused on genetic conditions, the authors found that PAO contributions to clinical research included assisting researchers with subject recruitment (91%), collecting research data (75%), assisting with study design (56%), and supporting a research registry or biobank (45%). This survey was also conducted in partnership with a PAO. These surveys suggest that most PAOs are engaged in research and that their contributions extend well beyond mere financing.

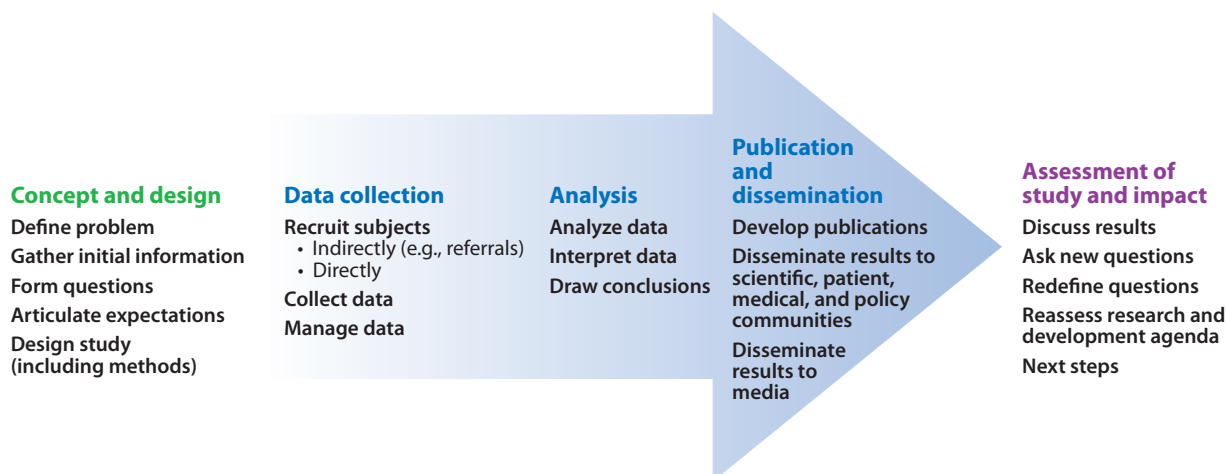
### Social Researchers Examine the Contributions of PAOs to Research

Patient groups have made major contributions to all phases and components of biomedical research (Figure 1). These groups sometimes seek to contribute to state and federal research policies (4) as well as legislation—e.g., the US Orphan Drug Act of 1983 and its amendments (51). By partnering with corporations, PAOs may also contribute to the development of new diagnostic technologies and therapeutics (46). Various social researchers have shown how PAOs contribute to scientific and technological developments through research agenda setting (21, 30, 61). In that sense, PAOs can act as either codevelopers or users of new technologies. In these ways, they can influence the direction of biomedical research and technological development.

Sociologists have shown that PAOs can influence clinical practice and patients’ lived illness experiences, e.g., by shaping illness-management strategies. In her study of HIV/AIDS patients,

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**Figure 1**

Contexts in which patient advocacy organizations (PAOs) can contribute to biomedical research. PAOs can contribute to multiple phases and components of research, including study design, definition of research goals, data collection and analysis, dissemination of results, and research funding. The figure highlights several of these contexts in which these organizations can contribute either through independent research or in collaboration with biomedical researchers. In developing new collaborations with PAOs, partners should clarify mutual expectations about the distributions of these tasks and be prepared to periodically revisit these roles.

Barbot (11) described four patient archetypes that are generated and reinforced by PAO interactions with patients: the patient as illness manager, the empowered patient, the science-wise patient, and the experimenter. Social researchers have also shown that PAOs can shape the attitudes and practices of health professionals. Through direct interactions with these professionals, PAOs can help motivate them to pursue particular social and medical agendas. For example, sociologist of science Aaron Panofsky, who interviewed PAO leaders and scientists, showed that PAOs may hold workshop-style conferences to connect potential biomedical researchers with PAOs to get them to focus on concerns important to the PAOs (57). Less directly, PAOs may work to bring about changes in clinical practice by participating in the development of clinical practice guidelines or by providing continuing medical education to health professionals. Social researchers have also shown that PAOs can impact how physicians and laypeople understand the etiology of a disease [e.g., see Silverman's work on autism (70)].

Social researchers are increasingly interested in studying patient groups that attempt to participate directly in biomedical research and pharmaceutical R&D. Some do this to help develop workable models of participatory research that can unite laypeople with experts (26, 29, 32). Understanding how patient advocates understand research collaborations, including the respective roles and distribution of responsibilities among collaborators, may help biomedical researchers develop better working relationships with patient groups. Although comparatively few PAOs become co-investigators in research (50), some social researchers contend that a major contribution of PAOs is to act as research catalysts, mobilizing networks of academic researchers, companies, and patients in pursuit of a common cause (1, 57).

Later, we present several case studies that illustrate some of the contributions that PAOs have made in rare-disease research, focusing primarily on the US context, but first we examine what we consider to be a key conceptual issue in understanding PAOs—namely, drawing boundaries between lay and certified forms of expertise.

## REPRESENTATION, POWER, AND CONTESTED EXPERTISE

### Challenging Experts and Expertise

Patient advocates have challenged traditional ideas about expertise, with many becoming experts in specific areas of research, such as breast cancer (40) and HIV/AIDS (30). Early commentators on PAOs tended to distinguish between certified experts and lay experts, noting that patient advocates were engaged in activities that blurred those traditional boundaries (30). Today, social scientists suggest that this conceptual distinction may no longer be very robust, as patient advocates and others have transformed the research landscape and generated new ideas about expertise (34). Greater emphasis on translational research and public participation in science are compelling both certified experts and PAOs to develop new ways of collaborating with one another in biomedical research and pharmaceutical R&D. These new collaborations not only challenge traditional social boundaries between experts and laypeople but also complicate the very notion of expertise itself.

Rare-disease research provides insight into the ways in which PAOs pose challenges to received views of biomedical expertise. In the context of this research, patients and their families have occupied social roles that closely parallel those typically reserved for certified experts in medicine and science. Historians and sociologists have shown that one major factor that has enabled PAOs to flourish in this context has been the comparatively small amount of research funding available to study rare conditions and the resulting absence of strong drivers of research competition (13, 73). They argue that these factors may give outsiders more opportunities to enter the realms of science and medicine than might be the case in other research contexts. By examining the work of PAOs that serve individuals with rare diseases, sociologists and others have nicely documented important tensions that can arise in partnerships between PAOs and biomedical researchers (13, 20, 64, 73, 84, 94).

### Representing the Roles of Patient Advocacy Organizations in Health Research: Advocate Voices

Patient advocates have described multiple benefits of PAO involvement in research (5, 83, 91). As noted above, many advocates have chosen to share their personal stories, which helps to elucidate some of these perceived benefits.

Over the past three decades, many patient advocates have demanded a seat at the political decision-making table. Today, advocates not only create PAOs and advocate for more patient-centric research agendas, but also are vocal in demanding the right to participate as investigators in biomedical research (78). In the mid-1990s, Weiss and Mackta (47, 88) described what they called “genetic support groups,” which expanded traditional self-help and support group activities to include the promotion of clinical research studies. Similar visions of a more expansive forum in which to explore an array of patient interests have since been articulated by many others, as increasing numbers of patient advocates and PAOs stress the importance of advancing science through patient-driven research (33, 76–78).

PAO leaders use multiple informational technologies and venues to share their experiences, views, and expertise, including websites and blogs, personal memoirs and biographies (3, 89), editorials and commentaries (77, 82), town-hall meetings, academic articles (33, 76), and other forms of social media (48, 49). Familiarity with these perspectives can provide biomedical researchers with insights into the motivations, missions, and expectations that PAOs bring to research collaborations. Combined with a better understanding of the unique histories of individual PAOs, advocate narratives can help researchers understand what PAO leaders may expect from research

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collaborations. At the very least, these narratives allow biomedical researchers to see patient advocates and the organizations that they have created as multidimensional entities that function within complex socio-organizational networks and institutional contexts. That richer understanding can highlight some of the constraints and opportunities that may be associated with PAO involvement in research, ultimately resulting in a deeper appreciation of how PAOs and biomedical researchers can work together.

As discussed above, qualitative researchers have attempted to characterize key features of collaborations between researchers and PAOs, often drawing on specific case studies. These researchers seek to describe how patient groups have forged relationships with scientists and initiated collaborations with various types with clinical researchers, academic or otherwise. Drawing on these ethnographically rich case studies, historians and social researchers may then develop conceptual frameworks or typologies for understanding these collaborations and assessing their significance over time, which we discuss in the sections that follow.

### PATIENT ADVOCACY ORGANIZATIONS AND GENOMIC SCIENCE

Many patient groups, especially rare-disease organizations, engage with scientific research into the causes and possible therapies of their respective diseases of interest. The interactions between these organizations and biomedicine are complex, particularly with regard to the organizations' relationships with biopharmaceutical companies (37, 46, 72). Relationships with academic researchers are complex as well; there may be tensions between researchers and PAO leaders, for instance, regarding research priorities, their respective understandings of disease, and their stances with regard to genetic testing and/or screening.

PAOs are often represented by historians and social researchers as playing a vital role in driving research and treatment for genetic conditions. Since the 1980s, there have been a growing number of partnerships between PAOs and genetic researchers in both the public and private arena. In the new millennium, there has been a resurgence of interest in rare genetic diseases and with it a corresponding increase in interest among social scientists and historians seeking to describe patient groups. Much of the resulting empirical research has been descriptive, attempting to explain these phenomena more systematically and to tease out the diversity of meanings that patient activism has in our society. Other research is much more action oriented and aims to improve collaborations between research stakeholders (29, 32).

Social researchers and patient advocates point out that genetic research can be a double-edged sword for both people living with genetic conditions and the organizations that represent them. Leaders of these organizations must resolve several tensions: Do we (PAOs) focus on diagnostics or therapeutics? Basic or clinical/translational research? Genetic or nongenetic contributors to disease? Advancing research or providing core services for our members? Preventive, pediatric, or adult-oriented research? Many genetic PAOs were founded in the 1990s in the midst of the biotechnology bloom, the Human Genome Project, and the emergence of the Internet. These developments have been important in creating an environment that promotes collaboration among PAOs, academic researchers, and industry. However, whereas earlier social research on PAOs focused on issues of patient empowerment and challenging expertise, new studies are documenting how personal narratives about citizen participation in biomedical science also suggest a new discursive shift to "collaboration" from discourses of empowerment and challenges to expertise (P.P. Koay, manuscript in preparation). In the following, we show examples and then discuss how increased boundary blurring among stakeholders has created additional challenges for patients, PAOs, policy makers, biomedical researchers, and researchers who are studying PAOs as a social phenomenon.

## AN EXAMPLE OF HOW FAMILIES CAN PROMOTE RESEARCH THROUGH PATIENT ADVOCACY ORGANIZATIONS

Jannine Cody is the mother of a child with a chromosome 18 syndrome. Her daughter Elizabeth was born in the mid-1980s with a severe bilateral cleft palate and metatarsus adductus. Genetic testing showed that Elizabeth was missing a segment of the long arm of chromosome 18. In a personal narrative published in *Genetics in Medicine* (23), she recounted that she was given a book about the disease and little more. Her reaction was that she and her family were “alone, negotiating the maze blindly and without a map” (p. 797).

Already having an MS in biology, Jannine enrolled in a PhD program in human genetics. She went on to receive her PhD and later became director of the Chromosome 18 Clinical Research Center at the University of Texas Health Science Center in San Antonio. She also founded the Chromosome 18 Registry and Research Society, which has flourished and now includes more than 1,800 affected children and their families. It also supports a large clinical research program studying these syndromes.

Other patient advocacy organizations for rare diseases have similar origins. As Cody wrote, “There are hundreds of similar organizations each dedicated to a specific genetic condition or group of conditions. . . . The most amazing thing about virtually all of these organizations is the fact that families of affected individuals started them. They were started without a business plan and without venture capital” (pp. 797–98). By blurring traditional boundaries between researchers and advocates, leaders of these organizations have challenged traditional understandings of scientific expertise.

### Blurring Boundaries: Parent, Advocate, and Researcher

In her case study of familial dysautonomia, Lindee (45) showed how families can be key drivers of biomedical research, particularly in drawing on their experiential knowledge in the process of caregiving. Sometimes PAOs are catalysts for parent-advocate-researchers; other times they are a consequence. In another context, Silverman (71) conducted an ethnographic study of autism in the United States and examined the role of caregivers, especially parents. Silverman’s work focused on Cure Autism Now (CAN), a PAO founded in 1995 by two Hollywood insiders, Portia Iversen and Jonathan Shestack, shortly after their son Dov was diagnosed with autism. Iversen and Shestack recognized that, by leveraging control over genetic materials and mobilizing online social networks, they could influence researchers’ perspectives on autism. Another interesting example of PAOs blurring boundaries between parents, researchers, and advocates is the Chromosome 18 Registry and Research Society (see sidebar An Example of How Families Can Promote Research Through Patient Advocacy Organizations).

Silverman’s study highlights the importance of several factors that influence the leadership of PAOs. For example, she noted the importance of advocates being inspired by and learning from other PAOs. She also noted that Iversen and Shestack drew inspiration from the Los Angeles–based Pediatric AIDS Foundation, founded in 1988 by Elizabeth Glaser. Glaser argued that in the milieu of funding neglect for HIV/AIDS research at the time, scientists had ignored the possibility that the virus could affect children in ways that differ from the way it affects adults. Silverman further noted that CAN followed a number of groups organizing their research activities around the aim of gene discovery, including PXE International, the Dystonia Medical Research Foundation, and the Hereditary Disease Foundation. These also provide cases of blurred boundaries between parent/caregiver, researcher, and advocate (45, 74, 90).

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### Blurring Boundaries: Patient Advocacy Groups and Biotechnology

In 1993, Brad Margus, the father of two boys with ataxia telangiectasia (A-T), founded the A-T Children's Project, a nonprofit research organization. He additionally cofounded Perlegen Sciences, a biotechnology company based in California; this company focuses on developing commercial products to identify genetic variations that predict patient response to pharmaceutical drugs, and on developing diagnostic tests that utilize genetic technologies. Although Margus has since moved into a board-member position as vice chairman of Perlegen Sciences, his involvement in both patient advocacy and commercial biotechnology illustrates the conflicts of interest that may arise for PAOs, particularly those that support individuals with very rare conditions (68). Recently, together with three scientists at Rockefeller University, Margus started a new company, Envoy Therapeutics.

### CONCEPTUAL FRAMES FOR EXPLORING RELATIONSHIPS BETWEEN PATIENT ADVOCACY ORGANIZATIONS AND RESEARCHERS

The case studies above illustrate how patient groups are transforming researcher-researched (expert-layperson) relationships. Although boundaries between scientists and laypeople are increasingly blurred, many genomic scientists and advocates seek to establish clear expectations in their individual collaborations. This can be viewed as an additional challenge to thinking about collaborations and how they can best be structured and made to work for participants. Thinking about conceptual frameworks can help clarify these boundaries and help explain them, and thinking about new conceptual frameworks may help to develop better collaborations.

One way that stakeholders and policy makers describe influences on science is to use the categories of external and internal influences (24). In this review, we use these categories because they are important in stakeholder and public dialogues about patient group involvement, not because we endorse this framing. As we have brought forward through some of the studies and literatures discussed above, society's views of biomedical R&D have changed significantly over the past 40 years, although these transformations are far from complete. What we highlight in this review are social efforts to study complex dynamics—e.g., to show how researcher-researched relationships have changed, how they are continuing to evolve, and how they will likely continue to change, especially in the context of genomic science. Here, we describe some efforts by sociologists of science to develop new conceptual frameworks.

Today's increasingly blurred boundaries mean that individuals can cross many traditional research divides—between laypeople and experts, between users and producers, across different professional groups, and so on. The distinctions between individual identities become further blurred in areas of venture philanthropy and biotechnology. One case that nicely highlights some of these developments involves John Crowley. Crowley is a lawyer who had only high school training in biology when he became the head of a biotechnology company seeking to develop a therapy for Pompe disease, a very rare, terminal illness that affected two of his children. Crowley's experiences were described in the book *The Cure: How a Father Raised \$100 Million—and Bucked the Medical Establishment—in a Quest to Save His Children* (3). As the subtitle hints, Crowley's efforts are often recounted as a challenge to medical experts, namely, to conduct bold research studies that seek to cure disease. Social researchers have analyzed Crowley's story with more nuance. In his analysis of this case, sociologist of science Carlos Nova (55) focused on the role of mass and corporate media in representing Crowley's narrative and his activities as an example of "American heroism" (the book served as the inspiration for the movie *Extraordinary Measures*). Novas also



argues that developments in commerce, manufacturing, communication, and marketing mutually reinforce one another and are implicated in this and other cases of PAOs (54).

Although many researchers and stakeholders acknowledge that patient groups continue to challenge certified expertise, they are also challenging different professional and group identities. Social researchers highlight the limits of policy frameworks to explain these phenomena (31, 35). Some suggest using the framework of user groups (22). Others suggest that reconfiguring our understanding of expertise may help us move beyond polarized frames of reference (e.g., lay expertise versus certified expertise) to describe the distribution of stakeholder contributions. As highlighted in our examples above, the boundaries between patients, advocates, and researchers are often blurred. This is also seen at the organizational level, in the blurring of boundaries between advocacy organizations, self-help, research institutes, and so on. Dividing expertise between laypeople (experiential) and experts (certified, technical) in some respects has become much less robust as a framing tool to explain these phenomena or facilitate discussion, and become more of a rhetorical device to include or exclude individuals and different groups. In his study of complementary and alternative medicines, anthropologist of science David Hess (34) suggested, for example, that analysts take into account what he calls the “coalition” or “hybridization” of knowledge: experiential expertise, lay expertise, and counter-expertise.

As we have seen, for good reasons, PAOs and their roles in biomedical and health research are increasingly of interest to health researchers, social researchers, bioethicists, policy makers, and the stakeholders themselves. However, limited data are available on how health researchers, policy makers, and clinicians understand the roles that PAOs play in research (42, 69).

Scholars from different fields of STS, bioethics, and health policy tend to focus on different questions and use different methodologies and bodies of knowledge. Researchers in the field of STS often highlight issues of identity crossings—e.g., from patient to community member to citizen or from advocate to parent to researcher. They do so to describe how stakeholders negotiate these identities, which are often constrained by different organizational forms and institutional norms, and to explore the impact of these efforts on the individuals involved. They may also study such boundaries to examine not only issues of identity and representation but also the roles and rights associated with these identities. Many STS researchers are concerned with including analyses that go beyond the (rights of) individual patients and other actors and their relationships (e.g., doctor–patient relationships) to look at societal rights and issues of equity. Examinations of patient activism in this light differ from the work of bioethicists and health policy researchers, who tend to focus more narrowly on the individual as patient and/or research subject. Legal studies in these fields have dealt with issues of informed consent, benefits sharing, and ownership issues, but have often still done so at the level of the individual (8, 27, 58) rather than the group. When bioethicists have considered larger organizational issues related to PAOs, they have tended to highlight issues related to the management of conflicts of interest (68). Although issues of legitimacy, authority, and organizational autonomy are at stake, they have not been subjected to enough scrutiny. Some researchers who study PAOs as a phenomenon are finding themselves in situations similar to those of academic and biomedical researchers and clinicians. These are situations with potential for productive partnership with PAOs but bring with them professional challenges. Some of these researchers have engaged directly in work that involves PAOs, and have reflected on both the professional importance of these activities and their challenges (71, 93).

The plurality of research methods and questions provides a rich and multidimensional resource of examination, exploration, and accounts of particular experiences and/or the phenomena themselves. There is a growing view that patients, consumers, and/or citizens should be more actively involved—that is, should be participants—in biomedical and health research, and some patients/consumers are demanding that inclusion. State, regional, and international bodies have

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declared or, in some cases, developed policy (e.g., the UK National Health Service) based on rhetoric that calls for health research and decision making to be more inclusive of patients/consumers (in the UK literature, the term consumers is inclusive of patient/consumer organizations). The media often present consumer and patient groups as driving this trend, portraying these individuals as tired of the slow progress made by academic researchers (37). PAO leaders also present themselves and their groups as collaborators in biomedical research who not only can help speed the process but also can provide important forms of expertise that are vital to the goal of developing new therapies.

Conceptual work in this arena becomes more important as this phenomenon—that is, increasing patient/nonexpert participation at all levels of research and/or its decision-making processes—becomes of greater concern to multiple interests, such as industry (especially biotechnology companies), regulators, and states. These groups, which at first glance may seem to have conflicting goals, are coming together, either voluntarily or because they have been compelled to, in several areas of health and biomedical enterprises, including genetics and genomics. Although the fact that patient groups partner with commercial entities, professional groups, and researchers may look like a completely new phenomenon, we stress again that PAOs, more generally, have varied historical origins. However, the more active role of patient groups as “cocollaborators” in biomedical research specifically, especially genetic and genomic research, has emerged much more recently (18, 44).

## **FUTURE COLLABORATIONS BETWEEN PATIENT ADVOCACY ORGANIZATIONS AND GENOMIC SCIENTISTS**

In this review, we have highlighted methodological and conceptual difficulties that challenge understandings of PAOs and their relationships to the biomedical enterprise. We now present an agenda for further research, aiming to clarify the multiple ways in which PAOs impact biomedical research and genomic science.

### **Historicizing and Contextualizing Collaborations in Genomic Science**

Historical and ethnographic researchers, such as the authors of the studies reviewed above, can trace how specific groups and PAOs come to work with or resist partnering with biomedical institutions. These and other social researchers have highlighted the importance of contextualizing and historicizing patient groups and their work. Through their scholarship, these social researchers identified areas that are significant for understanding PAOs, including (*a*) the natural history of the disease itself (the type and severity of the condition have a profound influence on the focus of the organization), (*b*) leadership (lay advocacy groups’ founders have a great influence on the organization’s vision and focus), (*c*) evolutionary and organizational aspects (the organization’s stage of development has a great effect on the services offered), and (*d*) the lived and social disease (the incidence of the condition—i.e., its rarity—and its relationship with R&D and the society has some effect on research conducted). In addition, to understand the relationships between PAOs and researchers, we must understand and explain (*e*) institutional considerations, (*f*) the national context, and (*g*) various sociocultural aspects of science.

Recent interest in promoting patient participation in research, particularly from the media, governments, and industry, is another double-edged sword for patient groups and researchers. Although “translational” and “patient centered” are key concepts in contemporary research, especially genomics, the rhetoric of collaboration is also a reflection of our times. Leaders of patient groups appear to move effortlessly across traditional borders between academia and industry, but,

as the recent ethnographic work discussed above suggests, the situation is more complex than it seems. Although some stakeholders claim PAOs are driving this rhetoric, we should note that there are mutually reinforcing developments in communication technologies (social media and the Internet), science (the Human Genome Project and genomic medicine), and industry (the demise of blockbuster drugs and patent expirations) that are also helping to drive the rhetoric of collaboration and that may strongly encourage the pursuit of these goals.

It is also important that we neither exaggerate nor understate the effects of PAOs on research. These organizations possess certain abilities to mobilize researchers, industry professionals, politicians, policy makers, regulators, and health professionals, and these capacities warrant further research (25). Some social researchers have found that patients may not have the same understanding of partnership and collaboration as leaders of PAOs promoting biomedical genomics research do (20). Further, as some researchers and commentators have noted, the PAOs that are often cited as successful models of partnership tend to focus on the needs of relatively privileged patients who have the social means to pursue research at academic medical centers. More studies are needed that examine how wealth and privilege impact the work of patient groups, especially community-based health groups (66). The case for such research has been strengthened because social research on diseases that have been associated with racial and ethnic groups has highlighted the important roles of community-based organizations, rather than of PAOs (as we describe), in the conduct of research (85, 86).

Historical and social science research has shown just how context specific these partnerships are and that there can be no single model for partnerships between PAOs and researchers in genomics or any other area of biomedical research. Nonetheless, individual case studies are more than singular stories: They can provide researchers with key insights into how the public engages science and how public understanding of science changes over time.

### Normative Considerations

The involvement of PAOs in genomic research has also raised normative questions, the most basic of which is why we should support more expansive forms of patient involvement at all (see sidebar Arguments for Expanding Patient Advocacy Organization Involvement in Biomedical Research). Currently, with efforts by most industrial nations to boost capacity in the area of genomic medicine, many patient groups have been able to align their organizational goals with those of researchers, funding agencies, and industry. This is an important development for those concerned with the future of health advocacy organizations more generally. As these and other collaborations expand, genomic scientists should consider whether certain areas of research may provide additional opportunities for partnership, e.g., relating to biobanking, recruitment for clinical trials, and health policy legislation.

Leaders of PAOs should also consider how collaborations with for-profit companies may impact how these organizations are perceived, potentially limiting public support or closing off certain opportunities for collaboration. Another issue concerns research. Several studies in which biomedical researchers were interviewed have found that, although scientists were not opposed to including the patients and patient advocates in their work, they had some reservations. Some of these concerns are related to the quality of research and potential for bias (57, 79, 87). However, as one group of social researchers found by conducting in-depth interviews with biomedical researchers, although scientists may have concerns about patient involvement, they believe they have an obligation to include patients in their work (21). These researchers also conducted interviews with PAO leaders and found that they acknowledged multiple challenges similar to those identified by scientists. Other concerns may come from patient advocates themselves, including

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## ARGUMENTS FOR EXPANDING PATIENT ADVOCACY ORGANIZATION INVOLVEMENT IN BIOMEDICAL RESEARCH

Social researchers have identified several arguments for including patient advocates and advocacy organizations in biomedical research. Building off earlier work from other researchers (30), Thompson and colleagues (79) conducted qualitative research in Britain with patients and health researchers and found that arguments for greater advocate involvement in research can be divided into two broad categories: those that appeal to the political advantages of mandating lay involvement, and those that cite specific research benefits resulting from advocate involvement.

Versions of the first argument—political justification—emphasize links between lay participation in research and democratic values. Prevailing notions of democracy suggest that members of the public, as the “owners” of publicly funded research, should have a say in the selection of research topics and the conduct of scientific studies. These arguments also appeal to the idea that research funding allocation should take into account the needs of those with legitimate interests. When viewed through this lens, lay participation in research becomes a political imperative—a noble pursuit in its own right, independent of study outcomes. The process of research decision making is the focus of these appeals, and concern for the quality of the resulting decisions is secondary. Advocates who employ the rhetoric of patient empowerment tend to evoke these political ideals in support of expanding advocacy organization involvement in biomedical research.

Versions of the second argument—specific research benefits—begin with the truism that patients and their caregivers have insights about disease that complement the medical expertise of health care professionals and biomedical researchers. Leaders of patient advocacy organizations maintain that the involvement of such organizations leverages those insights in a way that adds value to biomedical research. The tangible benefits of this involvement might include greater success in raising funds, identification of patient-centered study outcomes, more effective recruitment of patients for clinical trials, and success in disseminating research findings to interested patients. Patient groups argue that their involvement in generating scientific knowledge may also increase the perceived relevance and acceptance of research findings.

In combination with personal narratives by patient advocates, these two arguments for expanding advocacy organization involvement in biomedical research are useful in clarifying some of the advantages that patient advocates associate with their contributions to biomedical research.

issues of co-optation not only by industry but also by professional groups or others (36, 57). These aspects have not been well explored in prior studies and are particularly important to examine in order to clarify how patient groups and researchers negotiate tensions in their collaborations.

## CONCLUSIONS

PAOs have become significant partners in biomedical research. In genomic science, they have been important catalysts for the creation of biobanks and disease registries. Historians and social scientists have sought to describe PAO involvement in research, situating these evolving organizational roles within a broader historical and sociocultural context. It will be important to assess how patient groups have shaped genomic research; doing so will require examining their impact in a multifaceted way that is sensitive to the specific contexts in which PAOs function.

At the same time, researchers examining PAO contributions to research need to better understand one another’s methods and evidentiary standards and possibly develop more interdisciplinary approaches that reflect the multidimensionality of these phenomena, which cross scientific, social, cultural, and ethical boundaries. We hope that we have provided genomic researchers and others

with a useful review of recent scholarship in the medical humanities and socially oriented research that attempts to understand the roles of PAOs in biomedical research. Historical research can help us understand how PAOs have evolved over time, and contemporary social research provides examples and models of meaning-making among various stakeholders. Conceptual work can help us develop better ways of describing and explaining relationships among noncertified experts/researchers and more certified scientists. Policy makers should also be attentive to the growing body of research examining how the inclusion of organized patient voices can improve health care by making it more effective and equitable. Although the global effects of PAOs cannot be predicted, it is clear that genomic scientists will have to situate their work in relation to these and other forms of patient advocacy.

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