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Genetics' Influence on Patient Experiences with a Rare Chronic Disorder: A Photovoice Study of Living With Alpha-1 Antitrypsin Deficiency

Pamela Holtzclaw Williams, JD, PhD, RN^{a,b,*}, Lucinda Shore, MS^b, Marvin Sineath^b, Jim Quill^b, Barbara Warner^b, Jamila Keith^b, Deirdre Walker^b, Sara Wienke, MS, CGC^b, Susan Flavin, MSN, RN^b, and Charlie Strange, MD^b

^aCollege of Nursing, University of Arkansas for Medical Sciences, 4301 West Markham Street, #529, Little Rock, AR 72205-7199, USA

^bAlpha-1 Community Research Partnership, Medical University of South Carolina, 96 Jonathan Lucas St, MSC 630, Charleston, SC 29425, USA

Keywords

Photovoice; Chronic diseases; Genetics

Introduction

Patients with rare chronic disorders and their caregivers increasingly form communities to support and exchange social experiences. Because up to 10% of the United States population is affected by one of 5000 to 6000 rare disorders, efforts to understand both the individuals and affected communities are important. The current study was conducted using community-based participatory research (CBPR) approaches within a community of patients and caregivers living with alpha-1 antitrypsin deficiency (AATD). This relatively rare inherited condition creates a chronic protein deficiency that predisposes to obstructive lung or liver disease. Patient populations at some risk for lung transplant include individuals who smoked cigarettes and patients who underwent liver transplant in infancy and later adulthood due to accumulation of misfolded alpha-1 antitrypsin (AAT) within hepatocytes. The approaches, methods, and conclusions described here have implications for future research in other rare genetic disease communities that deal with disabling disorders.^{1,2}

CBPR partnerships historically emerged to address socially driven health-related needs and policy.³ The needs are framed by social experiences. For purposes of this article, “social experiences” refer to events not driven by physiologic symptoms. These experiences are influenced by public and social systems, health care infrastructure, and societal norms. Examples of social experiences include those shaped by public resource allocation, health care delivery systems and providers, public awareness and attitudes, and health related

policy. Therefore, the social themes in this rare disease community potentially generate universal messages.

The participants in the present study were asked to use their digital cameras and audio recorders to describe the social experiences related to living with ATTD. The aims focused on, but were not limited to, the patients' social experiences related to the genetic component of the disorder. The content of the photographs, narratives, and focus groups reflects themes of burden and resiliency that hold promise to advance knowledge about ATTD and for other rare genetic disorder communities.

Methods

Community identity framed around a shared genetic risk can generate CBPR approach challenges. The approaches require partnership and community members to share a wide range of perspectives with each other. However, rare disease patients are often geographically dispersed. Some community members protect themselves from identification related to genetic test results because of perceived threats to employment or insurance challenges.^{4,5} Therefore, this rare disorder community partnership found it necessary to tailor special strategies to systematically gather community members' input.

The described strategies and methods hold equal interest to the themes that emerged from this CBPR initiative. Few publications describe successful strategies for tailoring collection of community input when there is geographic isolation, disabilities that restrict mobility, or privacy concerns that limit public self-identification. This report describes use and results of mixed methods and technologies to engage community participation. This CBPR partnership, run by and for persons living with AATD and their caregivers, was established with an early goal to prioritize problems to address by research.

Formative Steps of CBPR

Under a memorandum of understanding, 5 patients and community members joined 5 academic clinicians and scientific personnel and became the Medical University of South Carolina (MUSC) Alpha-1 Community Research Partnership ("the partnership"). The MUSC Clinical and Translational Science Award (CTSA) was leveraged for resources to define research priorities. Community leaders expressed interest in using photovoice methodology found in the literature.⁶ Traditional focus groups were added to the design to enhance depth of the inquiry.⁷ The MUSC institutional review board (IRB) approved this study to identify and describe the social experiences within the AATD community.

Recruitment

Acting as a community partner, the Alpha-1 Foundation Research Registry is a rare disease registry located at MUSC supported by the nonprofit Alpha-1 Foundation. All recruitment of community member participation in the methods described subsequently was conducted through the e-mail or mailed invitations sent by this registry. This recruitment process assured that the identity of persons living with AATD was protected until he or she made the choice to participate and interface with the partnership's process. Throughout the process,

the choice was given to participants whether they wanted to share their identity with other participants, or participate to the extent possible under an anonymous status.

Data Collection

The data was collected during several phases.

Photovoice methods—Twenty-nine enrolled participants attended an online webinar that included a power point presentation and interactive time for questions and discussion. The webinar communicated steps to using cell phone photographic technology, digital cameras, and audio recordings to describe experiences and burdens associated with AATD. Participants could choose to participate in the webinar anonymously. Specific instructions included privacy and confidentiality safeguards for photographic and online communication and content. In addition to contributing photographic images, participants also were instructed to record a digital audio file to further describe the meaning of the photograph's content. The content of the audio files was transcribed for thematic content analysis. The photographs were sorted by theme for dissemination and also imported into NVivo software for content thematic analysis.

The focus group—After all the photographs and audio files were collected, the partners examined the content to select 5 participants for additional communication. Selection was designed to assure diversity of age and geography and included a caregiver. An online focus group was held by anonymous webinar to explore the perceptions invoked by the entire set of photographs and audio files. For example, some participants took turns responding to probes through the webinar microphone, while others used the chat box to type in their responses to probes. The audio portion of the session was transcribed. The transcript and text box narrative were imported into qualitative analysis software for thematic content analysis.

Data Analysis

The coding and analysis of the transcripts and material were imported into NVivo software to support the qualitative analysis. The photos were also imported into NVivo in coordination with the participants' comments about them, to assure the context of the transcribed statements remained intact and paired with the subjects' composed photographs. An inductive approach was applied to assembling a coding framework and coding for conceptual themes.⁸ Themes emerged from the iterative steps of coding and text review.⁹ The partners met to audit the coding, and an audit trail was maintained in NVivo.

In addition, NVivo supports analysis of word content in a data set by creating a visual graphic referred to as a word cloud. This visual graphic shows key word frequency. Such graphic images add visual support for drawing conclusions and communicating the context of content. Fig. 1 reflects the word cloud derived by using the transcribed text from the audio files and focus group content.

Results

The enrolled participants (n = 29) represented a wide range of geographic locations, multiple AATD-related genotypes, and ages ranging from early to late adulthood. A diverse community experience was captured by individuals with lung and/or liver symptoms, asymptomatic individuals who were carriers of an abnormal allele with affected young children, individuals who learned of their genetics from affected parents, and individuals disabled at a young age by the condition.

Fifty-five photographs and audio files were submitted. One participant asked permission to write his photo content explanations because of lack of knowledge and access to digital recording hardware. The photographs were taken by a diverse range of cell phone applications and digital cameras. Table 1 describes the themes that emerged and their meanings and provides exemplar quotes from the qualitative thematic analysis of the content from the audio narratives and focus group discussion.

Discussion of Resulting Themes

Genetic Etiology Influences Family Dynamics

A wide range of images and narrative comments focused on family dynamics affected by the shared genetic risks. Issues around the extent of genetic disclosure to extended family, family planning for future generations, and numbers of family members currently affected by symptoms and disability were prominent. The study of impact of genetic conditions on relationships, activities, and behaviors within generations of families is in its infancy, with the most study within the inherited cancers.^{10,11} The dominance of this theme from most of this study's participants highlights this priority topic for future research.

Genetic Susceptibility to Environmental Exposures

AATD is a disease in which genetic risk is linked to environmental exposures to cause clinical disease. The genetic interaction with environmental exposure risks is best understood in the context of cigarette smoke. However, some data suggest other particulates and fumes may be harmful.¹² Very little study has been done on patients' perceptions and accuracy in understanding inhalant-related health risks associated with any positive genetic test result.¹³ Genetic susceptibility to environmental inhalants such as secondhand smoke, pet dander, perfumes, and cleaning products has not been well defined. Therefore, in the absence of data, variability in lifestyles is extreme. Although avoidance of smoking cigarettes is universally advocated, research is needed to clarify which airway irritants cause future lung damage and if these risks are universal in the community.

Genetics Impose Testing Decisions for Immediate, Extended, and Future Family

Families are burdened by the impact of genetics on reproductive decisions, individuals' genetic testing decisions, sibling disclosures and testing decision-making, and parental decisions whether to test children. This decisional burden is common to many genetic disorders, particularly when a genetic test is technologically available to predict future risk of symptoms. Testing decision burden within families with genetic risks and carrier status

for inherited chronic conditions has been studied in inherited cardiac conditions,¹⁴ including Fragile X¹⁵ and sickle cell.¹⁶ The emergence of this theme in this population is consistent with those descriptive studies and warrants future research to develop interventions that mitigate the decisional burdens.

Perceiving Judgment by Others

Diseases that cause disability sometimes generate secondary social experiences. Examples of behaviors in response to perceived judgment included choosing to use/not use disability parking, hiding or avoiding medical equipment use to avoid others feeling uncomfortable, and choosing not to disclose prenatal in vitro services for family planning. For lung disease that lacks outward signs of disability, the invisibility can contribute to lack of public support and awareness. This phenomenon is not unique to genetic conditions, as it has been reported by persons living with chronic obstructive pulmonary disease (COPD) unrelated to AATD.¹⁷

Resilience

Finding the theme of resiliency in the content is consistent with study of other patient communities.^{18,19} This community uses physical activity to improve self-efficacy in symptom management and social support through pulmonary rehabilitation, an evidence-based outlet for organized patient mobility that generates quality of life improvement. The community also embraces advocacy and adaptation as personalized health behaviors in response to genetic information.²⁰

Limitations of Study

Although the data were rich in embedded themes, it must be acknowledged that the sample size was small, and there was no randomization in selection. The sample cannot be represented as representative of the entire population of persons with AATD. Ongoing directions of this partnership toward identifying a wider range of the experiences with the AATD community include 2 ongoing studies with significantly larger sample sizes, geographic distribution, and range of social experiences.

Summary

The MUSC Alpha-1 Community Research Partnership succeeded in data collection and analysis because of the mixed methods and use of technology in addressing the needs of this geographically distant rare disease community. The process of infusing the study with online technology and communication, including private webinars, secured drop boxes, e-mails, digital cell phone and camera photos, and digital audio narratives that can be sent via online technology kept the study exciting for the participants and allowed remote individuals to feel empowered. The authors have been able to demonstrate the feasibility of collecting qualitative research using voices and images of patients, caregivers, and community members living with this genetically inherited chronic condition. Importantly, the authors were able to simultaneously address the challenges of geographic distance and privacy in communications between each other.

Additionally, the authors' partnership was able to develop themes from the narrative and photographic content that help prioritize future CBPR initiatives. The CBPR partnership will use these themes to shape future research and disseminate these conclusions to other populations living with a genetically inherited chronic health condition. Finding 5 themes from this relatively small study indicates more research is warranted to advance clinical and public health interventions that mitigate social burden and support resiliency for living with a chronic condition that is genetically inherited and rare.

Implications for Nurses, Genetic Counselors, Providers, and Clinical Settings

The results and conclusions from this study imply suggestions for improved public health services and clinical delivery of care for persons living with chronic disorders with inherited genetic etiology.

One should recognize the social impact of living with an inherited genetic-based disorder in developing public health services, clinical care, and treatment plans; maintaining a referral resource list to community support options is also important. Additionally, should consider partnering with patient support or research advocacy groups to:

- Maintain social engagement and support networks

- Contribute to and/or maintain a genetic disorder patient registry that supports communication of research developments and support future recruitment in research participation

- Facilitate research to identify and describe²¹ patient-centered priorities in advancing intervention development responding to the patient community's psychosocial needs

- Promote public awareness and tolerance and educate regarding the need for mobility and social support among patients with chronic illness

One should additionally adapt traditional support group communication, patient education, and research methodology with Web-based technology to overcome geographic distances, privacy concerns, and mobility limitations.

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Key Points

- The Medical University of South Carolina Alpha-1 Community Research Partnership succeeded in data collection and analysis because of the mixed methods and use of technology in addressing the needs of this geographically distant rare disease community.
- The process of infusing the study with online technology and communication, including private webinars, secured drop boxes, e-mails, digital cell phone and camera photos, and digital audio narratives that can be sent via online technology kept the study exciting for the participants and allowed remote individuals to feel empowered.
- The authors have been able to demonstrate the feasibility of collecting qualitative research using voices and images of patients, caregivers, and community members living with this genetically inherited chronic condition.
- The partnership was able to develop themes from the narrative and photographic content that help prioritize future community-based participatory research initiatives, which will be used to shape future research and disseminate these conclusions to other populations living with a genetically inherited chronic health condition.

Fig. 1.

Word cloud derived by using the transcribed text from the audio files and focus group content.

Table 1
Results of thematic analysis of photovoice data: photographs, audio narratives, and focus groups

Theme and Its Abstracted Meaning	Representative Quotations from Narrative Data	Representative Photograph (www.alphaoneregistry.org)
Genetic Etiology Influences Family Dynamics Learning the chronic condition is based on genetic influences, emotions, and relational dynamics between siblings, parent and child, and spouses/partners; the genetic component can mean that many members of the same family, across generations, are affected at the same time, creating stressful health care costs to family households that affect family budget dynamics	<ul style="list-style-type: none"> • "I believe my parents felt guilty when we found out that I had alpha (ATTD)." • "The disease has 11 persons affected in my family so far, my sister had double lung transplant." • "My husband and I wonder when we have more children how many of them will have alpha one. It is a definite stressor in family planning." • "The suggestion to my extended family to get tested caused great strife among my family and ended up splitting some of the extended family apart from my immediate family."
Perceived Genetic Susceptibility to Environmental Exposures in Social Settings Participants understand ATTD as a genetic condition creating susceptibility to risks and triggers from environmental exposures; the heightened awareness modifies their social behavior and raises awareness that past behaviors (eg, smoking) contributed to outcomes	<ul style="list-style-type: none"> • "I won't go to workout centers just because of the exposure. We bought an exercise bike for home." • "Haven't had to modify too much yet, but in the future I see going to one floor home." • "This (picture of ashtray) represents 2 packs of cigarettes I smoked for over 30 years until I quit."
Genetic Inheritance Carries Decision Burden for Immediate, Extended and Future Family The ATTD family history or genetic test result places burdens to decide how and when to disclose to family members, family planning, testing recommendations, and exposure and living environment decisions	<ul style="list-style-type: none"> • "Decisions to keep your pets are ones alpha patients have to make on their own." • "My 18-month-old daughter is the one with the AATD genotype. My mother smokes and refuses to quit, so I had to make the hard decision to limit my daughter's time with her, and when they see each other it is at places she cannot smoke." • "Some of my family members chose not to get tested (for AATD genes)."
Judged By Others <ul style="list-style-type: none"> • Content reflects perceptions of being judged by others, and this influences their social behaviors • Content reflects apprehension that the invisibility of their disability contributes to lack of public support and awareness 	<ul style="list-style-type: none"> • "I get so embarrassed when I have to use my handicapped parking placard. People look at me like what's the matter with you." • "I feel like I have to hide the supplies when we have company so nobody feels uncomfortable." • "After our first son was born we learned we were both carriers of the alpha (AATD) gene. We decided that we did not want to risk having another baby born with the condition. So, we decided to do in vitro fertilization. We usually choose not to discuss our treatment with anyone besides family members, because it can be controversial."

Theme and Its Abstracted Meaning	Representative Quotations from Narrative Data	Representative Photograph (www.alphaoneregistry.org)
Resilience Strategies Modify Social Impact Collectively, content indicates actions to reduce psychosocial burden and risk associated with patients' genetic-based condition <ul style="list-style-type: none">Physical activity with othersAdvocacy with othersAdaptations to reduce isolation	<ul style="list-style-type: none">"I feel it (gathering with group for physical therapy) is necessary to my physical well-being, and it also stimulates my emotional well-being.""Research is so important to alpha-1 (AATD) patients. It is very important to my family and me to participate in fund raisers, research studies and donate to the alpha-1 foundation.""Backpack (oxygen tank in backpack picture) makes it easier for dad to maneuver through grocery store."