Assessing Community Engagement Factors To Support Individuals Impacted By A Rare Disease: Alpha-1 Antitrypsin Deficiency

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Abstract
Aims: to assess collective identity and capacity for community engagement approaches to intervention development; supporting individuals impacted by a rare genetic disorder, alpha-1 antitrypsin deficiency (Alpha-1).
Methods: case study methodology with mixed methods framed study of 6 data sources of communication between persons affected with Alpha-1. Analysis involved triangulation to assure completeness, convergence and depth. Conceptual frameworks for collective identity and community capacity guided analyses.
Results: Survey responses (n=694) showed high proportions of Internet use (86.9%), interaction with each other (67.7%), and belief that individuals can help someone newly diagnosed (76.4%). Thematic analysis identified all elements of a collective identity in the Internet chat room content. Triangulation showed convergence of all sources showing indicators of behavioral involvement that reflects community identity. All sources converged regarding community engagement capacity indicators: leadership and communication networks as resources.
Conclusion: Persons living with the rare genetic condition Alpha-1 express a collective identity using Internet resources. Through online technology this virtual community navigates a wide range of resources to support their diverse collective, with capacity to support intervention development to address patients’ social needs.

INTRODUCTION
Challenges of living with a rare disease are an important public-health issue.1 Millions of individuals are affected by an estimated 6000-8000 rare diseases.2 Persons with rare disease, the majority having genetic etiology, often experience different barriers to equitable healthcare delivery than persons with common diseases such as diabetes or hypertension. Persons with rare genetic based disorders face barriers to accessing: knowledgeable primary care and, in many cases, knowledgeable specialty providers, evidence-based psychotherapeutic support, and affordable medications or infusion products. Persons with rare diseases often experience diagnostic delays that affect health outcomes, no cure or symptom management medications, multiple family members in multiple generations with disabling symptoms due to genetic etiology, inherited genetic risks that create testing and information sharing decisional burdens; and perceptions of stigma, discrimination, and public apathy.1,3-10

Alpha-1 is a genetic disorder that causes susceptibility to chronic obstructive pulmonary disease (COPD) and cirrhosis with high rates of liver or lung transplantation and disability. The age of symptom onset is widely varied, from newborns to elder individuals.11,12 Several persons in an immediate or extended family pedigree must evaluate and expect genetic risk for symptoms, decisions concerning testing, and/or illness symptoms at the same time. Over recent decades, individuals, and families that share this genetic disorder formed organizations to support online delivery of self-management strategies, research recruitment, and communication in dealing with the social expectations of living with Alpha-1. The strategies include an Internet accessible research registry, websites and support group organizations that host public access online forums and networks.13

Few studies examine social networks created as a response to shared genetic information, test results, or disease risks.14 Likewise, the dynamics of how genetic knowledge and shared genetic information may lead to a collective identity
is also sparse.15 Field observations from this nurse investigator’s work with persons living with rare genetic disorders led to selecting the social networks of individuals with alpha-1 antitrypsin deficiency (Alpha-1) as a prototype to conduct a community assessment of whether a collective identity existed that had capacity for community engagement approaches to intervention development, delivery, and research. The Alpha-1 social networks were selected because the Internet reflected they had readily accessible data sources for case study analysis: a research registry with self-reported data from community members, online chat room interactive communications, website announcements of national and regional educational activities and conferences, and regularly published online community organizational reports to their membership. Two questions framed this study: 1) how do communications within the virtual social networks of persons with Alpha-1 demonstrate that this geographically distributed population of individuals shares a collective identity that constitutes a community?; and 2) if a community with a collective identity exists, does it have capacity to engage in community engagement approaches to intervention development, delivery, and research? Conceptual frameworks of collective identity16 and capacity17 were identified in the literature to guide the analysis.

AIMS AND OBJECTIVES
An advanced practice nurse employed in the role of nurse scientist needed to assess a community of individuals living with a rare genetic disease, specifically their collective identity and capacity for community engaged research approaches to intervention development for social burdens.17-19 Community engagement for intervention or research development requires formative work that begins with observing social contexts of the population of interest. The process involves iterative community assessments identifying community/patient members’ preferences that can shape desirable outcomes.20 The long range objective of this ongoing community engagement described here is to empower persons with the rare genetic condition, Alpha-1, to participate in shaping their own disease self-management strategies; promoting intervention development shaped by patient-centered outcomes from the perspective of persons living with this rare genetic disorder. This report describes nursing steps in assessing whether persons living with Alpha-1 have a collective identity that functions as a community, and whether the community has the capacity to collectively engage its voice in intervention development.

This report concludes by recommending to all advanced practice nurses the potential for engaging in similar assessments of other collectives living with rare genetic disorders. The Internet is a resource that allows online community assessments to support a variety of advanced practice nursing care roles for patients living with rare genetic conditions.

METHODS
The units of analysis were individuals’ and organizational communications. The 6 sources of data21 were:
• A set of individuals’ survey item responses (n=694) from the Alpha-1 Foundation Research Registry;
• Three reports containing leadership’s statements to persons living with Alpha-1 found within two organizational websites (Alpha-1 Foundation and Alpha-1 Association websites: http://alpha-1foundation.org/ and http://www.alpha1.org/ respectively)
• Two separate online chat rooms where individuals affected by Alpha-1 are in dialogue with each other.

The survey responses and chat room statements contained no personal identifying information. The content within the publicly accessible chat room comments and online reports were the units of study, not the individuals making them. This study method received Institutional Review Board approval from the Office of Research Integrity at the Medical University of South Carolina to proceed with exempt status from Human Research Subject regulations.

Sources
Survey Responses
The Alpha-1 Foundation registry (the Registry) represents a social network of persons with severe deficiency of AAT or the carrier status who consent to future re-contact for research recruitment.22 The Registry facilitated a 2000 person survey study regarding perceptions of persons with Alpha-1.23 The Registry provided access for secondary analysis of parts of the survey data to study conceptual framework elements of collective identity and community capacity.16,17 The survey items selected for analysis asked survey respondents to report various behaviors, interactions, beliefs, and communications regarding other persons living with Alpha-1. Survey data from convenience samples is a recognized data source to inform formative stages of community engagement.20
Leadership Reports

In addition to the Alpha-1 Registry, other extensions of the Alpha-1 organizational infrastructure offer online educational meetings and an online website for persons affected by Alpha-1. This online website publishes a newsletter containing leadership statements and reports to interested readers.

Three organizational reports were selected from 2009, 2010, and 2011. Reports were found through examining the organizational websites for the Alpha-1 Association and Foundation. Organizational representatives including the executive director of the Alpha-1 Association and the board director of the Alpha-1 Foundation prepared the report content for publication. This source yielded descriptions of organizational infrastructure and resources. The websites describe attendance levels at sponsored interactive events held, which provides information on the behavioral element of collective identity. Content in the websites and reports describe the Alpha-1 infrastructures’ resources; not available through chat room or survey data and in this way enriched the analysis.

Chat Rooms

Two public access online chat rooms provided access to observe online interactions. The chat rooms were supported by nonprofit entities: the Alpha-1 Association and a host calling itself “Spiderspun, an Alpha-1 Haven”. The postings were anonymous; no registration was required to post or observe the threads related to discussion topics and were posted over a period of two years. Chat room content offers opportunities to observe self-reported concerns, and thoughts without the investigator manipulating the setting.

Analyses

Each source was independently analyzed in the sequence listed in Table 3, and then the data and findings were triangulated to confirm depth and completeness.

Analysis for Survey Responses

Methods for Chat Room and Organizational Reports

The coding framework for the thematic analysis of observed statements in chat rooms and organizational reports was guided by a priori elements comprising the conceptual frameworks of collective identity and capacity for community engagement approaches. Chat room postings were entered into Excel spreadsheets and then imported into NVivo9 software, so that each posting became a unit of analysis for coding. Text from online reports was copied and pasted into Word documents, and also imported into NVivo software. The coding of content was deductive, coding chat room and organizational report statements as they related to the framework elements of collective identity: self-categorization, evaluation, importance, attachment/sense of interdependence, social embeddedness, behavioral involvement, ideological content and meaning. Where content did not address collective identity elements or indicators of community engagement capacity, new coding categories suggested by the content were developed using an inductive approach. The framework elements or “key indicators” of community engagement capacity that guided coding were: effective leadership, inclusive membership, complementary competencies and adequate resources.

Triangulation

The data and analyses were triangulated for completeness and convergence. The process included triangulation of findings from the 3 different categories (survey response, chat room posting and infrastructure website organizational reports) of sources. Table 3 summarizes data features considered for the triangulation process.

DISCUSSION OF RESULTS

Survey Responses

Table 1 summarizes results from statistical analysis of the survey. The first 4 variables describe the characteristics of the survey population. The majority of the population was female, Internet users, had clinical symptoms and their genetic test result indicated the genotype associated with the most severe Alpha-1 symptoms.
Collective Identity Elements

The majority of respondents reported reaching out to others with Alpha-1 after diagnosis. Forty-five percent of respondents reported organization interactions; 22% reported their family was interested in the Alpha-1 support groups or organization. Seventy-six percent of respondents engage in the belief that they could help a fellow newly diagnosed person with Alpha-1, indicating a strong indicator of a “sense of interdependence felt with others in the group”, a dimension of the sense of collective identity.16

Community Engagement Capacity Indicators

Leadership among a collective of individuals that reflects “skill, experience, cooperation”17 is an indicator of capacity for community engagement initiatives. Although the survey items and data did not directly address leadership, the high proportion of respondents reporting a belief in ability to help another newly diagnosed person suggests self-perceptions of requisite skill and experience to lead peers. Another skill revealed in the survey items was the experience and knowledge of Internet communication; 87% reported using the Internet for communication.

Chat Rooms Postings

The chat room postings offered observations of discussion among individuals with Alpha-1 in a naturalistic setting where they exchange interactive statements in conversation threads.27,28 Chat room statements were anonymously posted which offered a degree of privacy and freedom to express thoughts and concerns without judgment and guilt. As one posting stated “I can talk about my concerns without burdening my family”. The contexts of the postings were free from errors that can occur in survey method such as: poorly articulated questions influencing results, responses bias, and inaccuracies due to poor recall. In chat rooms the observed statements reflected real time perceptions. However, privacy and anonymous formats prevent confirming reliability of the source or collect corresponding demographics.

Collective Identity Elements (Table 2)

Table 2a

<table>
<thead>
<tr>
<th>Table 2a</th>
<th>ALPHA 1 CHAT ROOM POSTINGS REFLECT ELEMENTS OF COLLECTIVE IDENTITY: EXEMPLARS</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>ELEMENT: ATTACHMENT/DEGREE OF INTERDEPENDENCE, EMOTIONAL INVOVLEMENT WITH GROUP</strong></td>
<td></td>
</tr>
<tr>
<td>I have no other family for support besides my kids and my parents, but they have always been there for me. When I feel the need for emotional support, a chat room is my first choice. (11/10/2023 8:34 PM)</td>
<td></td>
</tr>
<tr>
<td><strong>ELEMENT: ATTACHMENT/DEGREE OF INTERDEPENDENCE, EMOTIONAL INVOVLEMENT WITH GROUP</strong></td>
<td></td>
</tr>
<tr>
<td>I don’t think I could ask for more support than I do here. I feel supported and valued by the community. (11/10/2023 8:34 PM)</td>
<td></td>
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Table 1

<table>
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<tr>
<th>Table 1</th>
<th>Characteristics of Population(s) and Proportions of Reported Interactions</th>
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</thead>
<tbody>
<tr>
<td>Variable</td>
<td>Proportion (n/N*)</td>
</tr>
<tr>
<td>Gender (female)</td>
<td>56.1% (359/654)</td>
</tr>
<tr>
<td>Internet user</td>
<td>69.9% (459/657)</td>
</tr>
<tr>
<td>Symptomatic</td>
<td>73.9% (516/690)</td>
</tr>
<tr>
<td>Severely deficient genotype: PiZZ, PiSZ or PiZZ</td>
<td>68.2% (471/691)</td>
</tr>
<tr>
<td>Heard about Alpha-1 through a national organization or support group</td>
<td>8.4% (58/691)</td>
</tr>
<tr>
<td>Interacted with organizations for explanations, questions and concerns about diagnosis</td>
<td>45.2% (308/681)</td>
</tr>
<tr>
<td>Believed they could help a newly diagnosed ‘Alpha’</td>
<td>76.4% (527/690)</td>
</tr>
<tr>
<td>Family member did want to get involved in support groups and the Alpha-1 community</td>
<td>22.4% (149/666)</td>
</tr>
</tbody>
</table>

*Variations in total N due to missing data
All elements of the conceptual framework of “collective identity” were found in the thematic analysis of chat room postings, as described and exemplified in Table 2. The theme arising most frequently from the postings was self-categorization, including by description of their particular genotype.

Capacity Indicators

Membership inclusivity, as a community engagement capacity indicator, was demonstrated by the reciprocity and mutual responsiveness to the postings. The postings all received responses indicating support or with referrals to knowledgeable leaders within organizations for answers. Many chat room postings expressed specific emotions of support and validation towards other discussion participants, reflecting a collective emotional state of mutual support.

Other Themes

Themes about the social burdens experienced as a result of an Alpha-1 diagnosis emerged from the inductive coding approach applied to content that did not fit the deductive coding framework, instead indirectly informing and confirming the need for this community’s voice to be empowered by community engagement approaches. For example, a theme of “lost trust” in providers was suggested by the chat room content. Examples suggesting this theme were references to delays in diagnosis and testing for Alpha-1 and provider lack of knowledge regarding infusion and medication approaches. A theme of “isolation” and “powerlessness” emerged, suggesting motivation for using the chat room. “Isolation” was also expressed by their descriptions of frustration that friends and even family are not familiar with this rare disease.

Organizational Reports

The organizational leader’s reports all contained references that demonstrated collective identity and community engagement capacity indicators.

Collective Identity and Community Capacity Indicators

All 3 reports referred to membership activities that indicate the collective identity framework’s behavioral element. All 3 reports refer to leadership experience and cooperation taking place at the support organizational level. The reports also described resources available to persons living with Alpha-1, through organizational frameworks. Leadership experience, cooperation, and resource availability are all community engagement capacity indicators.

Triangulation, Completeness & Depth Analysis

Table 3 summarizes data features considered for the triangulation process. Thematic analysis identified all elements of a collective identity in the chat room content. Triangulation showed convergence of the behavioral involvement element of community identity. All sources converged regarding community engagement capacity indicators: leadership and adequate communication networks as community resources to support research partnership endeavors.

Table 3

Summary of Method, Data and Analysis Triangulation

The 6 sources of data offered a complete range of
perspectives and demographic characteristics of persons living with Alpha-1. The range included persons with verified genotype status (survey participants), persons not sure of their genotype but knew they were at risk (chat room content), caregivers and family members (chat room, websites, and reports content). Chat room postings (Table 2) included self-identified brothers, sisters, parents, and spouses of persons affected by Alpha-1 test results or symptoms. The range of diverse relationships to Alpha-1 experiences offered completeness to the holistic approach to searching for a collective identity. Persons with Alpha-1 from many US states, and from other countries participated in the chat rooms. The naturalistic context of the chat rooms and organizational reports supported the depth of this descriptive analysis.

CONCLUSIONS

A collective identity exists among persons living with variable experiences with Alpha-1. Linked by their strategic use of information technology such as chat rooms, webinars, and organizational websites, this collective identity functions as a virtual community. The number of chat room postings, the online Foundation and Association websites, and proportion of survey respondents reporting Internet usage all reflect this community’s strategic use of information technology to maintain their linkage to one another. The conclusion that information technology facilitates this community’s expression of collective identity is consistent with other studies’ findings that persons with rare diseases outpace other patient populations in seeking out the company, perspectives and advice of their peer network using the Internet.

Regarding community capacity, the sources reflect that the Alpha-1 virtual community has capacity and need for community engagement approaches in intervention development to address the social burdens discussed in chat room content. All sources converged on the community capacity indicators of peer leadership, skills and experience. Inclusive membership and adequate resources were demonstrated in the content of chat rooms and the organizational newsletters.

The survey responses that served as one source of data (Table 1) for this study were from a convenience sample, limiting representativeness of the overall population of persons with Alpha-1. However, since the survey data served as only 1 out of 6 data sources, the limitation does not substantially threaten the completeness or depth of the findings.

Implications for Practice

Genetic information and conditions can create expectations of being different or the same as others, and may define communities. Recent advancements in personalized medicine technology support genetic information use to identify health factors. However, individuals’ responses to knowing their genetic information and collective identities based on shared genetic information are not well understood. More nursing inquiry is needed to understand how disease-specific social networks share their genetic information as part of their self-regard and how it is socially embedded in relationships contributing to health outcomes.

The analyses determined that individuals living with shared genetic risk for, or confirmed testing results of Alpha-1, indeed generate a collective identity using the Internet as their community neighborhood. Through strategic information technology use, this Alpha-1 community shares its genetic information, and collectively addresses its social consequences. The communications express a collective identity with community capacity for engaging in intervention development to resolve unjust social expectations and burdens of healthcare marginalization. The Alpha-1 collective identity and its community engagement capacity serve as a traditional model to support nursing inquiry into other geographically dispersed, rare disease collectives of individuals at similar stages of resource development.

The findings from this study imply future potential in nursing leadership to assess and facilitate research based community partnerships to study intervention development. The analysis described here was followed by formation of a community based research partnership. The partnership include two Alpha-1 support group leaders, a leader from a national support organizational infrastructure, and four community members (Alpha-1 patients) that represent the wide age and range of interests of the patient population. The partnership went through formative stages of organizing and identifying community needs for research, and was recently awarded research funding and community partnership training from the Medical University of South Carolina’s Center for Community Health Partnerships and the Patient Centered Outcomes Research Institute (PCORI). The partnership’s long range plans are to develop high impact community based interventions to reduce perceived
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powerlessness, to increase health provider and community awareness and to empower participation in improving healthcare delivery and rare disease health policy. Findings have multiple implications for advanced practice nursing. Advanced practice nurses (APNs) in clinical settings caring for patients with rare genetic conditions such as Alpha-1 can assess Internet and social media sources to support patients’ engagement in shaping the research that affects their health outcomes through community based research partnerships. APNs in community health settings can support persons living with rare genetic conditions by maintaining knowledge and competency in using Internet and social media resources for empowering and engaging persons living with rare genetic conditions in education and social support intervention development and delivery. APNs in the role of nurse scientists can develop community engaged research where they assess a capacity for conducting studies in virtual rare disease communities. All APNs, in their advocate roles, can serve as advisory partners in community based participatory research partnerships and contribute their expertise to serving communities of persons living with rare genetic conditions.

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