
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-1.²⁴ This online website publishes a newsletter containing leadership statements and reports to interested readers. 13,25

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

Descriptive statistics were used to report survey results. Frequencies and proportions were obtained for demographic characteristics, Internet use, and self-report behaviors. Although these data had strengths in a large sample size and participant characteristic data, they were limited in their range of perspectives by the item format. Other perspectives in natural settings without research manipulation and limitations were pursued to add completeness to the data.

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 identity¹⁶ and capacity for community engagement approaches.¹⁷ Chat room postings were entered into Excel spread sheets 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.¹⁶ 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.¹⁷

Triangulation

The data and analyses were triangulated for completeness and convergence.^{21,26} 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.

Table 1

Characteristics of Population(s) and Proportions of Reported Interactions

Variable	Proportion (n/N*)
Gender (female)	56.1% (389/694)
Internet user	86.9% (589/678)
Symptomatic	73.9% (510/690)
Severely deficient genotype: PiZZ, PiSZ or ZNull	68.2% (471/691)
Heard about Alpha-1 through a national organization or support group	8.4 % (58/691)
Talked to other 'Alphas' after diagnosis	67.7% (469/693)
Interacted with organizations for explanations, questions and concerns about diagnosis	45.2% (308/681)
Believed they could help a newly diagnosed 'Alpha'	76.4% (527/690)
Family member did want to get involved in support groups and the Alpha-1 community	22.4% (149/666)

*Variations in total N due to missing data

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 methods 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

ALPHA 1 CHAT ROOM POSTINGS REFLECT ELEMENTS OF COLLECTIVE IDENTITY: EXEMPLARS

TABLE 2 ALPHA 1 CHAT ROOM POSTINGS REFLECT ELEMENTS OF COLLECTIVE IDENTITY: EXEMPLARS	
ELEMENT: SELF CATEGORIZATION: IDENTIFYING ONESELF IN TERMS OF SOCIAL GROUPING	
<ul style="list-style-type: none"> > My son is a ZZ Alpha. > I'm so sorry that you lost your brother to Alpha-1. We have far too many Alpha singles in heaven right now. > I am new to the Alpha aspect of life. I haven't found out what category I fit into. 	<ul style="list-style-type: none"> > "Hi I'm an alpha." > I'm a ZZ with COPD. > I am a ZZ both liver and lung affected. > I too am a ZZ. > My husband and I are both MZ's
ELEMENT: EVALUATION: REFLECTING POSITIVE/NEGATIVE ATTITUDE TOWARD SOCIAL CATEGORY	
<ul style="list-style-type: none"> > My husband and I deal pretty well with Alpha-1 on most days. I have an occasional "why us" day, but on most days, I can find the silver lining <i>somewhere!</i> > My husband and I have not been tested. Partly, due to insurance discrimination issues and partly because we are not sure we want to know. 	<ul style="list-style-type: none"> > Unfortunately, our kids also lost the genetic lottery. Both are ZZ's and are liver affected. With that said, I would not change a thing and we are thrilled with our two kids.
ELEMENT: IMPORTANCE: REFLECTING DEGREE COLLECTIVE IS PART OF SELF CONCEPT	
<ul style="list-style-type: none"> > We are so relieved to meet other parents who have "been down this road" or who are currently traveling with us. > All I ever wanted was to be like the other kids and be able to run all the laps in gym without having to stop because I could not breathe. To be able to walk up steps without getting winded. Alpha children feel different but long to be normal and at times may lash out because of their frustration. 	
ELEMENT: ATTACHMENT/SENSE OF INTERDEPENDENCE: EMOTIONAL INVOLVEMENT FELT WITH GROUP	
<ul style="list-style-type: none"> > This is the really great part of the Internet...meeting people going through similar situations that I never would have met otherwise. > I am now part of a new family of loving, caring and understanding people. > Thank you for giving me the WANT to go to the conference, and the time and love you put into alpha kids I have so much info on what it is I need to look out for and do for my MZ kids. And lastly, thank you members/congrats for making me feel welcome and a part of my new extended family. > I hope you are all doing well and you are all in my prayers. > All this is new to me so would be nice to have the support of other Alphas. > Believe me when I say, "I know what you are going through." > So, anyway, I cope by helping others, praying for strength and the health of my children, and by meeting other parents just like you even if it is only in cyberspace. 	<ul style="list-style-type: none"> > Thank you again for sharing with me. I just met you and yet you have been like a rock in a storm. > I have a great support group of friends and family, but it is a different (good) kind of support to talk to people who are in the exact same boat! > Isn't it strange how one gene can link all of us strangers together? > I just want to say, "God Bless" to all the parents who have experienced the heartache and fear with having a child, or children, been diagnosed with alpha-1. I am glad that this community is here. > I can relate. I'll be praying for your entire family. HUGS, and please come back here and ask questions as they arise. We love to help. > Welcome to "our little corner of the Internet" But most of all, Welcome to your new extended family. > I just wanted to thank everyone for this website. It has helped me a great deal over the last few months.

Table 2b

ALPHA 1 CHAT ROOM POSTINGS REFLECT ELEMENTS OF COLLECTIVE IDENTITY: EXEMPLARS

TABLE 2 ALPHA 1 CHAT ROOM POSTINGS REFLECT ELEMENTS OF COLLECTIVE IDENTITY: EXEMPLARS	
ELEMENT: SOCIAL EMBEDDEDNESS: COLLECTIVE EMBEDDED IN EVERYDAY SOCIAL RELATIONSHIPS	
<ul style="list-style-type: none"> ➤ We no longer live in the fear of "what if" mostly because we feel well informed. Our kids understand their health concerns and have established healthy living patterns. We also understand that things could change at any time and accept that. ➤ My life will certainly be different from this point. I have read many of the postings on this site and listing, to others and how they cope, gives me some hope. 	
ELEMENT: BEHAVIORAL INVOLVEMENT: ACTIONS DIRECTLY IMPLICATING IDENTIFICATION WITH COLLECTIVE	
<ul style="list-style-type: none"> ➤ We need to get involved. ➤ (Speaking about attending a conference) Not only did I meet my first alpha (besides my brother), but I got an education of a lifetime! ➤ I don't really have any questions at the moment. I don't think, I guess I'm just looking to connect with people who understand what I'm going thru. Thanks for listening. 	<ul style="list-style-type: none"> ➤ As I said I don't know much about much right now, but if I can help anyone or if anyone wants to talk. ➤ I've been to the last two national conferences and they were great. The association is a great way to get educated and find some support. ➤ Our first few months of dealing with the Alpha-1 diagnosis are thanks to the mentoring program through the Alpha-1 Association. We cannot say enough about the support and information this organization provided us.
CONTENT & MEANING: DEFINITION: IDENTITY IS INTERNALIZED & REFLECTED IN IDEOLOGY	
<ul style="list-style-type: none"> ➤ I have 2 girls who are both ZZ alphas. It isn't what I would have chosen for them, but we live it and move on. After all, Alpha-1 doesn't define who we are and how "wonderful" of life we can have. ➤ Little by little, I came to accept Alpha-1 in our lives, and now I can see one positive effect it has had on my family. We don't take our time together for granted anymore. Each day is a blessing. 	<ul style="list-style-type: none"> ➤ We must educate ourselves as much as possible so we can take the appropriate steps to overcome this war we are fighting within ourselves. Think of it as "we have Alpha, Alpha doesn't have us". We may not be able to stop the war but every day we exist is a battle we have won. If you think of it as Alpha has you, you've lost the fight.

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; included 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.²⁹

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.¹⁶ 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.¹⁷

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 Triangulation³⁵

Sources in Order of Analysis	Completeness: Factors that explain, limit or promote understanding of collective identity &/or community capacity?	Context: Setting, Approach, Methods	Evidence of Collective Identity ¹⁶ &/or Community Capacity ¹⁷ (CICC Elements)
1 Set of Survey Responses (n=64) Dates of responses: 09/28/2009 – 10/28/2009	<ul style="list-style-type: none"> • Large sample size with verified Alpha-1 testing results • Includes respondents' demographics not available in other sources • closed-ended item format limits completeness • Sample limited to individuals with positive genetic testing results, i.e. limited range of perspectives 	<ul style="list-style-type: none"> • Multiple choice formatted responses to printed questionnaire format • Secondary analysis of responses looking for indicators of collective identity & community capacity • Verified relationship to Alpha 1 part of survey inclusion criteria: 	<ul style="list-style-type: none"> • Self-reported Behavioral Involvement (CI) • Evidence of Leadership self-efficacy (LCC) • Internet usage rates indicate communication competency (CC)
2 Sets of Chat Room Postings (n=979) Dates of postings: 2010-2011	<ul style="list-style-type: none"> • Large sample without verification of Alpha-1 experience • Wide range of age, geography and Alpha-1 experiences not available in other sources • Individual participant's characteristics & demographics not always included 	<ul style="list-style-type: none"> • Naturalistic, virtual setting with no investigator manipulation • Real time expression of perceptions with minimal limitations to expression • Direct observation method of collection from public access webpages 	<ul style="list-style-type: none"> • All collective identity elements: • Capacity elements: Leadership, resources & inclusiveness, internet communication competency (CC)
3 Organizational Messages/Reports: a) 2009 Alpha-1 Foundation Annual Report b) 2010 Letter from Alpha-1 Association Director c) 2011 Message from Alpha-1 Foundation Chairman of the Board	<ul style="list-style-type: none"> • Websites offer most complete source of organization infrastructure, resources & meeting descriptions not available in other sources • Content refers to wide range of Alpha-1 experience: tested, untested, caregiver, parents, siblings • Limited by unilateral format • http://www.alpha1foundation.org/about/?c=01:Message_From_The_Chair • http://archive.constantcontact.com/s/0901103657014e20arch/e/1030557791.html_alpha-1foundation.org/file_download/723 	<ul style="list-style-type: none"> • Naturalistic, virtual setting with no investigator manipulation • Infrastructure operations & events, attendance levels & resource descriptions available to enrich contextual meaning of survey & chat room posting content • Direct observation method of (unilateral) communication from organization leaders to individuals living with Alpha-1. 	<ul style="list-style-type: none"> • Behavioral Involvement element of collective identity • Leadership, resources & internet communication competency as Capacity elements

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.³⁰

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 these 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.³¹ 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-regard³² and how it is socially embedded in relationships contributing to health outcomes.³³

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 model³⁴ 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. Community partners 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

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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References

1. Schieppati A, Henter JI, Daina E, Aperia A. Why rare diseases are an important medical and social issue. *Lancet*. Jun 14 2008;371(9629):2039-2041.
2. Institute of Medicine. Rare Diseases and Orphan Products: Accelerating Research and Development. National Academies Press. Report; 2010. <http://www.ncbi.nlm.nih.gov/books/NBK56189/pdf/TOC.pdf>. Accessed Jan 10, 2011.
3. Kole A, Faurisson F. Rare diseases social epidemiology: analysis of inequalities rare diseases epidemiology. In: Posada de la Paz M, Groft SC, eds. *Rare Diseases Epidemiology*. Netherlands: Springer; 2010:686:223-250.
4. Williams PH. Policy framework for rare disease health disparities. *Policy Polit Nurs Pract*. 2011;12(2):114-118. doi: 10.1177/1527154411404243
5. Bouwman MG, Teunissen QG, Wijburg FA, Linthorst GE. 'Doctor Google' ending the diagnostic odyssey in lysosomal storage disorders: parents using internet search engines as an efficient diagnostic strategy in rare diseases. *Arch Dis Child*. 2010;95(8):642-644.
6. Henderson SL, Packman W, Packman S. Psychosocial aspects of patients with Niemann-Pick disease, type B. *Am J Med Genet A*. 2009;149A(11):2430-2436.
7. Swanson ME. Need for the life course model for spina bifida. *Pediatr Clin North Am*. 2010;57(4):893-901.
8. Joachim G, Acorn S. Life with a rare chronic disease: the scleroderma experience. *J Adv Nurs*. 2003;42(6):598-606.
9. Dures E, Morris M, Gleeson K, Rumsey N. The psychosocial impact of epidermolysis bullosa. *Qual Health Res*. 2011;21(6):771-782.
10. Griffith G, Hastings R, Nash S. You have to sit and explain it all, and explain yourself; Mothers experiences of support services for their offspring with a rare genetic intellectual disability syndrome. *Journal of Genetic Counseling*. 2011;20(2):165-177.
11. Stoller JK, Fromer L, Brantly M, Stocks J, Strange C. Primary care diagnosis of alpha-1 antitrypsin deficiency: issues and opportunities. *Cleveland Clin J Med*. 2007;74(12):869-874.
12. Strange C, Stoller JK, Sandhaus RA, Dickson R, Turino G. Results of a survey of patients with alpha-1 antitrypsin deficiency. *Respiration*. 2006;73(2):185-190.
13. Walsh JW, Snider GL, Stoller JK. A review of the alpha-1 foundation: its formation, impact, and critical success factors. *Respir Care*. 2006;51(5):526-531.
14. Plumridge G, Metcalfe A, Coad J, Gill P. The role of support groups in facilitating families in coping with a genetic condition and in discussion of genetic risk information. *Health Expectations*. 2011;15(3): 255-66.
15. Nordgren A. Genetics and identity. *Community Genet*. 2008;11(5):252-266.
16. Ashmore RD, Deaux K, McLaughlin-Volpe T. An organizing framework for collective identity: articulation and significance of multidimensionality. *Psychol Bull*. 2004;130(1):80-114.
17. Andrews JO, Newman SD, Meadows O, Cox MJ, Bunting S. Partnership readiness for community-based participatory research. *Health Educ Res*. 2010;27(4):555-71. doi:10.1093/her/cyq050.
18. Israel B, Eng E, Schulz A, Parker E. *Methods in Community-Based Participatory Research for Health*. San Francisco, CA: Jossey-Bass; 2005.
19. Eng E, Moore K, Rhodes S, et al. Insiders and outsiders assess who is "the community". In: Israel B, Eng E, Schulz A, Parker E, eds. *Methods in Community Based Participatory Research for Health*. San Francisco, CA: Jossey-Bass; 2005:73-101.
20. Hull PC, Canedo JR, Reece MC, et al. Using a participatory research process to address disproportionate Hispanic cancer burden. *J Health Care Poor Underserved*. 2010;21(1 Suppl):95-113.
21. Yin R. Collecting case study evidence: the principles you should follow in working with six sources of evidence. In: Yin R. *Case Study Research Design and Methods*. 4th ed., Thousand Oaks, CA: Sage; 2009:99-124.
22. Alpha-1 Foundation. Alpha-1 Foundation Alpha-1 Research Program. 2012; <http://www.alpha1registry.org/>. Accessed April 9, 2012.
23. Finn S. *Astute, Assertive, and Alpha-1: Quantifying Empowerment in a Rare Genetic Community* [Doctoral Dissertation], University of Florida; 2008.
24. Alpha-1 Association. Alpha-1 Association website. 2011. <http://www.alpha1.org/>. Accessed April 2011.
25. Alpha-1 Association. Alpha-1 News A Publication of the Alpha-1 Association. 2008, 2009. 2010.

<http://www.alpha1.org/>. Accessed April 2011.

26. Leppakoski T, Paavilainen E. Triangulation as a method to create a preliminary model to identify and intervene in intimate partner violence. 2012; *Appl Nurs Res*. 25(3):171-180.

27. Macias W, Lewis LS, Smith TL. Health-related message boards/chat rooms on the web: discussion content and implications for pharmaceutical sponsorships. *J Health Commun: Int Perspectives*. 2005;10(3):209 - 223.

28. Nolan M, Hodgins M, Olsen S, et al. Spiritual Issues of family members in a pancreatic cancer chat room. *Oncol Nurs Forum*. 2006;33(2):239-244.

29. Chmiel A, Sienkiewicz J, Thelwall M, et al. Collective emotions online and their influence on community life. *PLoS One*. 2011;6(7):e22207.

30. Fox S. Peer-to-peer healthcare. California HealthCare Foundation. 2011.

<http://pewinternet.org/Reports/2011/P2PHealthcare.aspx>. Accessed April 9, 2012.

31. Zwart H. Genomics and identity: the bioinformatisation of human life. *Med, Health Care, Philos*. 2009;12(2):125-136.

32. Brodwin P. Genetic Knowledge and Collective Identity. *Culture, Medicine and Psychiatry*. 2005;29(2):139-143.

33. Rabeharisoa V. From representation to mediation: the shaping of collective mobilization on muscular dystrophy in France. *Soc Sci Med*. Feb 2006;62(3):564-576.

34. Nicolaidis C, Raymaker D, McDonald K, et al. Collaboration strategies in nontraditional community-based participatory research partnerships: lessons from an academic-community partnership with autistic self-advocates. *Prog Community Health Partnersh*. 2011;5(2):143-150.

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