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ORIGINAL ARTICLE

Understanding genetic learning needs of people affected by rare disease

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Abstract

Nearly 350 million people worldwide are affected by a rare disease (RD) and ~80% of RDs have a genetic type, underscoring the need for access to reliable genomics education. Patient assistance in resource development can help ensure content is appropriate. The aim of this study was to define the needs and practical usage of the RD community to inform the scope and content of an online genetic course targeted toward the entire RD ecosystem. A high-level online survey (OS) was disseminated to 586 RD patients and family members/caregivers. A total of 251 individuals responded to the OS. Eight respondents were invited to participate in a follow-up focus group (FG). Nearly 87% of OS respondents have made efforts previously to learn more about genetics and 95.6% indicated a current interest in genetic education. Navigating healthcare systems, information sharing, and advocacy support were driving factors for this desire. Respondents indicated difficulty finding information on gene function, genetic testing, disease pathogenesis, and scientific advances. FG outcomes dove deeper into psychological needs including reducing emotional burden, alleviating fear of the unknown and seeking hope. Research identified high levels of interest in genetic education across all stages of the RD journey. Key themes identified in this study may help guide genetic counselors as they create their own patient and family-facing content.

KEYWORDS

complex disease, education, health promotion, patient advocacy, psychosocial

1 | INTRODUCTION

There are over 7,000 rare diseases (RDs) affecting nearly 350 million people worldwide (Bick, Jones, Taylor, Taft, & Belmont, 2019; Eurordis 2018; Ferreira, 2019; Global Genes, 2015; Nguengang Wakap et al., 2019). Knowledge and resources surrounding RD are limited, especially for newly diagnosed individuals (Lewis, Snyder, & Hyatt-Knorr, 2017; Molster et al., 2016; Nutt & Limb 2011; Wittink & Oosterhaven 2018). Given the high number of RD coupled with a shortage of medical experts for each condition, there is a deficiency in reliable RD information (Budych, Helms, & Schultz, 2012; Kole & Faurisson 2009; Lewis et al., 2017; Molster et al., 2016). This deficiency extends to broader genomics education as well.

The broad need for genomics education is especially important for patients, caregivers, and healthcare providers (Topol 2019). This gap continues to grow along with rapid advances in genomic testing technologies that can offer accurate, molecular diagnoses to those previously undiagnosed (Kole & Faurisson 2009; Talwar, Tseng, Foster, Xu, & Chen, 2017). Reliable resources regarding basic genetic concepts, including testing and diagnosis,

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are available but are generally not targeted to the RD community and are often unreliable or unclear (Aaronson, Joshua, & Boss, 2018; Badiu et al., 2017; Kakazu, Schumaier, Minoughan, & Grawe, 2018; Schoof & Wallace 2014). Healthcare providers may not be equipped to answer genetics-related questions that arise throughout a patient's RD journey (Litzkendorf et al., 2016). In addition, a healthcare provider's unfamiliarity with RD may lead to a lack of knowledge about available services and an inability to identify when a referral is indicated (Kole & Faurisson 2009). A vast majority of patients and caregivers with RD are self-directed learners (Budych et al., 2012). Parents of children with RD feel the burden of responsibility due to the complexity of their child's condition and the lack of knowledge and understanding from healthcare providers (Currie & Szabo 2019).

Patient organizations are often the main or only resource for RD patients and caregivers and are therefore vital (Limb & Nutt 2010; Pauer et al., 2017; Pinto, Martin, & Chenhall, 2016). Larger patient organizations often take on the responsibility to serve as global providers of support and education. For example, Genetic Alliance, the National Organization for Rare Disorders (NORD), Global Genes, and EURORDIS each provide educational content on their websites (Eurordis: Rare Diseases Europe 2015, Genetic Alliance, 2018, Global Genes, 2015, and National Organization for Rare Disorders, 2019). RD advocacy websites include disease-specific information for hundreds to thousands of genetic conditions, as well as general RD facts, and guidance for advocacy and orphan drug development. The NIH Genetics Home Reference website includes a primer section entitled, 'Help Me Understand Genetics' that can serve as an introductory review of basic genetic concepts. NORD has brief videos targeted toward key topics, including gene therapy and insurance coverage.

A recent, independent report from the United Kingdom addressed the importance of including patients as partners in their health care and recommends the development of genomics education programs with the support of patient and caregiver organizations (Topol 2019) . Global Genes is a non-profit organization focused on building networks among stakeholders in RD. They developed an online learning initiative, Rare University, which provides free education on topics salient for people affected by RD. Assai Health Solutions was approached to develop a genetic course specifically tailored to the needs of people affected by RD. The aim of this study was to assess the needs and practical usage of such a course from the perspective of the RD community, which is defined in this paper as the RD patient, family members, caregivers, and other members of their support team. These findings would then go on to inform the scope, content, and features of an online course.

2 | METHODS

This study was submitted to Sterling IRB for review and determined to be of minimal risk to human subjects and thus exempt from review.

2.1 | Survey

2.1.1 | Survey development

An online survey (OS) was prepared by the lead researcher from ASSAI Health Solutions and two members of the Global Genes staff. The survey was reviewed and amended by additional members of the Global Genes staff with experience conducting qualitative research including surveys in the RD community. A pilot survey was not conducted. The survey was developed on the surveying platform SurveyPlanet™ and included 10 quantitative and 2 qualitative questions for a total of 12 questions (Appendix S1). The objective of the OS was to collect *self-reported*: duration of time indirectly or directly affected by a RD, interest in genetic education, level of knowledge of genetics and information-seeking behaviors including types of digital platforms used and preferred for learning, intended practical application of information, and content of interest. Demographic information was not collected in the survey.

2.1.2 | Participants

Survey participants were identified through the Global Genes Foundation Alliance (FA) which at present comprises over 600 independent RD organizations. Eligible organizations consisted of support groups, foundations in the United States with 501(c) (3) status and international foundations that provide access to RD information. (A complete list of all FA members is available at https://globalgenes.org/wp-content/uploads/2019/09/Global-Genes-FA-Sept-19.pdf) At the time this survey was distributed, there were 586 FA members. Survey inclusion criteria were limited to anyone having been affected directly or indirectly by a RD. The first survey question was used to determine eligibility as it asked how long the individual was affected directly or indirectly by RD. Exclusion criteria were limited to any individual who did not consider themselves to be directly or indirectly affected by RD and therefore did not complete the survey or provided no answer to the first question.

2.1.3 | Survey distribution

The OS was distributed to all 586 FA members by Global Genes through email to the lead contact for each FA member organization giving them permission to forward the survey on to their RD organization memberships and social media networks. Information is not available about the total number of survey invitations sent out beyond the initial 586. The survey launched in January 2018 and was available for 14 days. Participants were informed that their answers would help in the development of an online genetic course for people affected by RD. Participation in the survey implied consent.

2.1.4 | Data analysis

Analysis of survey results, excluding statistical analyses, was performed using the SurveyPlanet™ platform functionality and Microsoft Excel 2016 for Windows.

Statistical analysis was performed using IBM SPSS Statistics for Windows 24 to determine whether there was a significant difference in scores in responses to online survey question 2, '*How would you rate your understanding of genetics at the start of your rare disease journey (1 = little to no knowledge, 10 = expert level knowledge)*' and question 3, '*How would you rate your current understanding of the genetic factors involved in the Rare Disease that affects you or your loved one? (1 = little to no knowledge, 10 = expert level knowledge)'*.

Respondents were categorized into three groups based on length of their RD journey as reported: <5 years, 5–10 years, and >10 years. Wilcoxon tests were used to assess differences in self-reported genetic knowledge at the start of the journey versus present day *within each group* of respondents. Kruskal–Wallis one-way ANOVA and post hoc analyses were used to assess differences in self-reported genetic knowledge *between groups* both at the start of the journey and at present.

2.2 | Focus groups

Two focus groups (FGs) were conducted over five days in February 2018, to explore in depth, what course structure and tailoring of content the RD community preferred. FG participants comprised a convenience sample of eight survey respondents who were invited to participate and accepted to do so. Participation was voluntary, and no compensation was provided.

2.2.1 | Participants

Focus groups participants were invited to represent a variety of stakeholder profiles including individuals directly affected by a RD, parents/caregivers, those with <3 years of RD experience, and those with >10 years of RD experience. Specific efforts to include individuals who were recently diagnosed (<3 years) were made as this population was not well represented in the OS. Collection of demographic information was limited to gender, length of time in RD journey, and country of residence at the time of participation.

2.2.2 | Materials

The FGs were conducted via video conference using the Zoom™ platform. A semi-structured interview guide was prepared to explore needs related to course structure and tailoring of content to the RD community (Appendix S2).

A presentation in PowerPoint 2016 was prepared for display during the FG video conference. The presentation included an overview of objectives, an explanation of the development process for the genetic course, a presentation of selected results from the OS, and the questions comprising the semi-structured interview guide. Facilitation was conducted by a single moderator with experience conducting FGs with individuals in the RD community. The decision to conduct two FGs was driven by scheduling availability of participants and both FGs followed the same format and content as described above.

2.2.3 | Methods

Each FG began with verbal consent for the recording of the FG. Consent was given by all participants. The prepared presentation was reviewed, and semi-structured interview questions asked in the sequence in which they appear in the interview guide (Appendix S2).

2.2.4 | Data analysis

Discussions were recorded, transcribed into Microsoft Excel 2016 spreadsheets, and analyzed by the lead researcher from Assai Health Solutions. Analysis began with grouping answers to each question by theme and summarizing the answers provided.

3 | RESULTS

3.1 | Online survey results

Overall, 251 individuals responded to the OS within 14 days of initial distribution. All respondents provided a response to the first question, how long have you been affected by a RD, to determine eligibility (251/251; 100%). No respondents were excluded. Details regarding the OS participants are presented in Table 1. The SurveyPlanet™ platform collected geographic location data for all 251 respondents; therefore, it was inferred that 224 (89%) reside in the United States, while the remaining reside in Canada (12;4.8%), Europe (10;4.0%) Australia/New Zealand (3;1.2%), or other countries (2;0.4%). Approximately half (127/251; 50.5%) of respondents had been directly or indirectly affected by a RD for over 10 years. Few respondents had been affected for less than one year (9; 3.6%). Nearly 96% of respondents stated they are interested in learning about genetics either now or in the future.

Respondents were asked to rate their understanding of genetics on a scale of 1 to 10 at the start of their RD journey (question 2) and at present (question 3) to assess improvements over time (Table 1).

Self-reported level of genetic knowledge at the start of the journey was not significantly different between the three groups (<5 years (*median* = 3), 5–10 years (*median* = 2), >10 years (*median* = 3), $X^2(2, N = 251) = 1.225$, $p = .542$). Genetic knowledge improved significantly since the start of the journey for all groups (<5 years, $Z = -5.737$, $p < .001$, 5–10 years, $Z = -6.060$, $p = <0.001$, and >10 years, $Z = -8.364$, $p < .001$). Present knowledge was significantly higher

TABLE 1 Profile of online survey respondents (n = 251)

Country of residence[a]			
United States			224 (89%)
Canada			12 (4.8%)
Europe			10 (4.0%)
Australia/New Zealand			3 (1.2%)
Other (Venezuela; India)			2 (0.4%)
Length of time directly or indirectly affected by a RD			***N* (%)**
Less than 1 year			9 (3.6%)
1–3 years			23 (9.2%)
3–5 years			30 (12%)
5–10 years			62 (24.7%)
More than 10 years			127 (50.5%)
Understanding of genetics at start of RD journey vs. today[b]	**Start (*Median*)**	**Today (*Median*)**	***p* value**
Respondents <5 years' experience (n = 62)	3.0	6.0	<.001
Respondents 5–10 years' experience (n = 62)	2.0	6.0	<.001
Respondents >10 years' experience (n = 127)	3.0	7.0	<.001
p value	.524	.032[c] .047[d] .870[e]	
Efforts made to learn about genetics since start of RD journey			***N* (%)**
Yes			218 (86.9%)
No			33 (13.1%)
Interested in learning about genetics now or in future			***N* (%)**
Yes			240 (95.6%)
No			11 (4.4%)

Note: Summary of online survey respondents, including years of experience with the rare disease (RD) community, current country of residence, assessment of genetic knowledge, and interest in learning more about genetics and RD. Data presented in this table are based on the survey responses (N = 251).

Abbreviation: RD, rare disease.

[a]Information ascertained through SurveyPlanet™ data, not directly from respondent.

[b]Scale 1–10; 1 = little to no knowledge; 10 = expert.

[c]Comparison in median knowledge between <5 years and >10 years' RD experience.

[d]Comparison in median knowledge between 5–10 years and >10 years' RD experience.

[e]Comparison in median knowledge between <5 years' and 5–10 years' RD experience.

for those with >10 years (*median* = 7) compared to both those with <5 years (*median* = 6), $X^2(1, N = 189) = 4.624$, $p = .032$ and those with 5–10 years (*median* = 6), $X^2(1, N = 189) = 3.947$, $p = .047$. However, present genetic knowledge did not differ significantly between those with <5 years (*median* = 6) and 5–10 years (*median* = 6), $X^2(1, N = 124) = 0.027$, $p = .870$.

A majority (218; 86.9%) of respondents have made personal efforts to expand their genetic knowledge over time and most (240; 95.6%) indicated a continued interest in gaining more knowledge in the future for both personal and medical reasons (Table 1). Reasons for previously seeking out genetic information included (multiple answers allowed) understand the impact of RD (n = 185), understand research into possible treatments (n = 166), help explain RD to other individuals (n = 164), and understand risk to family members (n = 139). Participants were asked to list up to five topics they have had difficulty locating information on previously and up to five topics they would like included in a beginner-level genetic course (Figure 1). Information regarding basic genetic concepts and disease pathogenesis were the most commonly reported topics for both questions.

The most commonly used resources to learn about genetics were Internet searches, including medical journals (n = 154), physicians (n = 129), other RD patients (n = 111), and patient groups (n = 108) (Figure 2). Factors that made resources especially helpful to respondents included the ability to reference over time (n = 132), understandable language (n = 114), ability to ask specific questions (n = 111), developed for the RD community (n = 108), self-paced learning (n = 103), trustworthy (n = 101), and interactive (n = 69).

Respondents were asked on what type of device they would prefer to access a genetic course. Home computer (n = 197) was the most frequent response followed by mobile phones (n = 127) and tablets (n = 122).

Potential uses of a genetic course included sharing with other RD families (n = 205), sharing with friends or family (n = 189), sharing with members of own patient advocacy organization (n = 163), and sharing with their child with RD when they reach adolescence (n = 144). A fourth response option, 'other', enabled respondents to write in additional uses. Sharing information with healthcare providers was mentioned by nine respondents.

3.2 | Focus group results

Participants provided detailed feedback on genetics learning needs and ideal course format, content, and accessibility. An overall summary of preferences regarding course scope, content, and features is presented in Table 2, and quotes from the FGs are presented in Table 3.

FG participants summarized ideal course objectives as enabling learners to become more aware of and able to apply genetic concepts and communicate this information to relevant individuals (e.g., healthcare providers, family members, members of the RD community). Participants also reported the need to feel a sense of confidence in navigating interactions with others based on a better understanding of basic genetics.

There was an emphasis on the consideration of a RD learner's emotional well-being, including sensitivity to diversity, which in this setting is operationally defined as the spectrum of how every

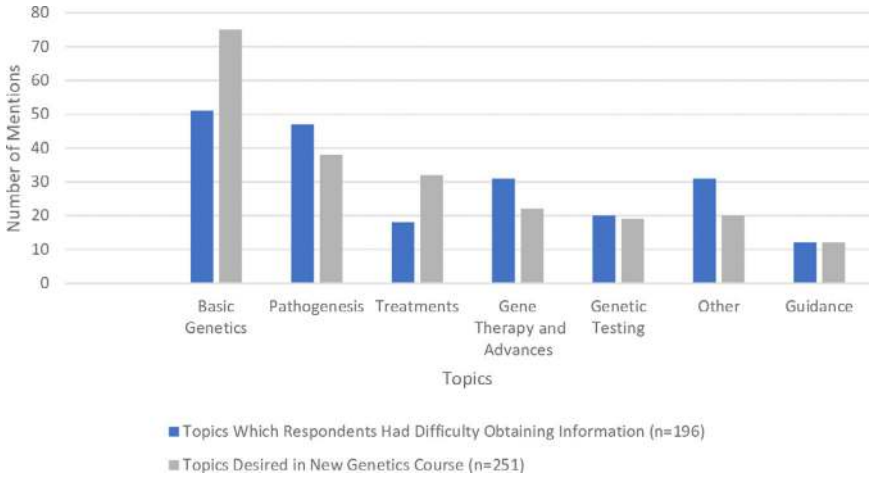

FIGURE 1 Preferred topics in a new genetics learning course. Topics respondents have had difficulty locating information on previously (*n* = 196) and topics respondents would like included in a new genetic education course (*N* = 251). Responses were grouped into themes. Respondents could provide upto five topics per question. Data presented are based on survey responses only

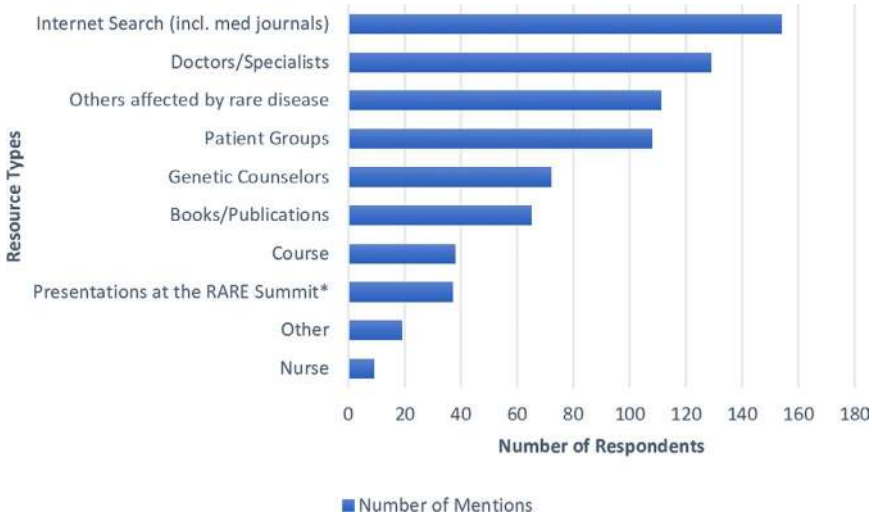

FIGURE 2 Resources used by survey responders to learn about genetics and rare disease. *RARE summit is an annual patient conference sponsored by Global Genes. Online survey respondents were asked to list resources they have utilized to learn about genetics. Multiple responses were allowed. Data presented based on survey responses (*N* = 196)

individual with a RD is uniquely affected and where they may be in their RD journey. Emotional impacts resulting from this diversity include apprehension and evolving emotional dynamics that can occur when one individual is seen as doing worse or better than another with the same condition. Difficult topics, such as heredity, fertility, comorbidity, and mortality, could be presented with consideration of a learner's frame of mind.

The facilitator asked how best to encourage course learners to reach out as needed. A common response was to offer a forum to answer questions, perhaps anonymously, that also allows learners to see questions previously asked by other learners. Emphasis was placed on providing resources to existing RD groups. The development of specific courses that could bring individuals together with similar diseases was also suggested.

4 | DISCUSSION

Knowledge can lead to confidence and empowerment which in turn can positively impact quality of life (Ayme, Kole, & Groft, 2008; Cohen & Biesecker 2010). This study demonstrates that there is a need and desire for genetic information tailored to the RD community. Participants from both the OS and FGs indicated that their motivations for seeking out information about genetics included a realization that healthcare providers lack knowledge about their condition, a desire to do right by their child, a need to accept the impact of RD and to alleviate fear of unknown. Significant interest in genetic education exists for all respondents, regardless of the length of time in their RD journey. One possible reason for this may be that the pace of advancement in genomics has generated a deeper interest in staying up to date.

Based on responses from the FGs, RD patients and caregivers appear to understand genetics through a holistic lens, considering their physical, psychological, and emotional well-being throughout the learning process. There is a desire for skill development and practical application support to accompany education which may lead to increased confidence and reduced anxiety. These results show some alignment with results of previous research into specific information needs within the RD community, including genetic causes, inheritance patterns and reproductive implications, access to therapies, research participation opportunities, legal and insurance considerations, and access to practical support resources including patient groups and social services (Litzkendorf et al., 2016).

TABLE 2 Summary of course preferences in the rare disease community

Scope	Gene/genome function Pathogenesis Management Gene therapy and scientific advances Genetic testing
Duration	6–12 weeks
Pacing	Self-paced, average of 1–2 hr of learning per week No pre-determined learner journey (choose own adventure)
Format	Easily referenced as needed over time Mix of video, audio, text catering to multiple learning styles Easy-to-navigate platform
Content	Suitable for individuals with varying levels of knowledge Explanation of genetic concepts relevant to rare disease beyond 'basic' genetics concepts Ready-to-use content to support navigating important interactions Medically accurate and peer-reviewed
Tone/style	Simple language, especially to explain complex genetic concepts Empathetic tone Considerate of cultural diversity Provides hope or does not take away hope
Interactivity	Opportunity to ask questions Availability of a mentor/administrator Enables sharing of information
Special considerations	Negative outcomes, heredity, and comorbidity discussed with special consideration for learner frame of mind Does not describe features/experiences/outcomes as universally representative Parental sign-off for learners under age 18
Duty of care	Non-prescriptive content Information and tools to support decision making Efforts made to provide robust sign-posting rather than relying on 'talk to your doctor'

Note: Summary of preferences in a genetic course including content, format, style, interactivity, special considerations, and duty of care. Includes details provided during the survey and focus group discussions.

4.1 | Genetic education and the patient perspective

Feedback from the FGs highlighted the importance of factoring in the diverse circumstances that exist among this community and how those circumstances may impact the learning process. Rare Disease UK published results from a survey of nearly 600 patients and their families who reported on various aspects of their RD experience (Limb & Nutt 2010). In addition to the lack of available information, individuals with RDs have many unique educational, medical, psychosocial, and therapeutic learning needs. Learning styles may be impacted by age, language, and physiological diversities; therefore,

TABLE 3 Selection of quotes from focus group participants

Focus group topic	Participant quote
Learning objectives	• Feel empowered and better able to discuss treatment with the doctors… • Be able to find resources or know where to look and conduct research, via web or social media • …educate the school system; educate the school admin, nurse, teachers on my (child's) rare disease
Timing of learning about genetics	• It was out of necessity for me needing to figure this out. Now that I'm an advocate it's also to help others in my rare disease community and others with any rare disease • As soon as I heard they couldn't tell me what version of my rare disease I have, I wanted to be tested and I needed to understand how that would work and how to access testing • We started trying to understand right from the very first day what was happening
Need to revisit genetics information later in time	• We had to look back at the beginning of the year when my son started school so that we could explain it to the school • Before doctor's visits • On a first read you may see something and not realize how it's applicable to you, then say a couple of months later you may find out it is and you might then need to go back and look at it again (in the context of) 'now it applies to me'
Socio-emotional considerations	• Some people may not have a great support system and this course may represent an important support system for them, so listening empathetically (is important) • You may want to have a section with guidance of how to approach discussions with family, that's a lot of people's questions around genetics
Motivators for learning	• My faith is what keeps me going, because sometimes there aren't answers to some questions and my faith is where I turn to • I try to stay focused on what's ahead and not think about the difficult things
Connections and support	• You want to make sure you point to existing groups so you're not taking away resources or remit from patient groups… • Provide info on the family groups that exist, connecting people to networks because many disorders already have organizations that exist • Beyond a general genetics course, it may be interesting to have a course specific to a group of disorders that share genetic similarities—to take it one step up for tailoring

Note: A selection of comments from focus group participants ($N = 8$) to highlight key points regarding learning objectives, course timing, format, socio-emotional considerations, motivation, and resource needs.

information must be provided in a range of formats and at various levels of medical and scientific detail to ensure full understanding and informed decision making (Beagley, 2011; Nutt & Limb 2011). Barriers may also exist for those individuals without a confirmed diagnosis and those still accessing or implementing a care plan with their provider.

The learner's frame of mind can impact interpretation and retainment of new information. Several participants indicated feelings of fear, loss of control, and panic as part of the emotional backdrop to early attempts to learn about genetics. This is an important consideration as optimism and hopefulness have been associated with a higher quality of life (Pauer et al., 2017).

The idea that a course of this type should avoid impacting feelings of hope by the user is somewhat unique to this population. Participants offered recommendations on how educational content could be presented in a positive way that does not interfere with feelings of hope while also alleviating the fear of the unknown. Medically accurate, peer-reviewed information that is sensitive to the spectrum of features and concerns associated with many RDs should be presented without marginalizing those individuals who do not fit the classical spectrum. Additional suggestions include redirecting users to disease-specific organizations and enabling learners to define their own path through self-guided content. A program that is self-paced and flexible, enabling personalized navigation based on where the learner is in their journey, is ideal for the RD community. In addition, for learners under the age of 18, parental sign-off was suggested as a method for which parents could assess the readiness of their children, both with and without RD, to learn about complicated topics.

Participants were asked how a course could effectively address personal concerns, for example, when considering options for family planning. There was a consensus that this type of course should not provide personal advice to learners; however, it should also not default to a simple 'talk to your provider' statement.

4.2 | Genetic education and online learning

Although interactive learning with a healthcare professional is generally preferred, there is significant interest in online learning resources. This is consistent with other studies that have reported Internet resources as a preferred method to obtain information for management of rare or chronic disease (Fox, ; Hamilton et al., 2015; Molster et al., 2016). Caution needs to be taken with online patient resources as they are not all rooted in science and written without bias. Foundation-sponsored sites are most reputable when their purposes are clear, and their board of directors includes individuals with medical degrees or other relevant and specialized degrees (Bergeron, 2004). One German study evaluated the quality and reliability of RD information on the Internet (Pauer et al., 2017). The top 20 hits for 8,000 RD searches were screened yielding a total of 693 RD information suppliers. Support groups/patient organizations (38.3%) and medical institutions (26.8%) were the two most common RD information providers. Overall quality was found to be comparable between these two types of providers; however, quality was significantly higher for sites produced by 'other associations and supporting bodies'. Only 6% of all sites had a certification or quality seal and 30% lacked appropriate privacy and updating criteria.

Younger individuals with RD are likely to turn to social media or video-sharing sites, such as YouTube, for information about genomics and genetic testing. A systematic review of health information and YouTube reported it contains misleading health information that contradicts scientific resources, except for sites sponsored by government organizations and professional societies (Madathil, Rivera-Rodriguez, Greenstein, & Gramopadhye, 2015). Anecdotal information is more prevalent on social media and video-sharing sites. Madathil and Greenstein evaluated the use and effect of anecdotal information on choice of healthcare facility and found an inverse relationship between age and the likelihood of seeking out anecdotal information, suggesting a need to educate younger consumers about the quality of healthcare information available on the Internet (Madathil & Greenstein 2018). This study further reported that misleading, anecdotal information was more influential when presented earlier in the learning journey compared to later in the journey. This type of narrative cannot be eliminated from Internet sources; however, ensuring accurate and reliable information, including anecdotal information, is available on reputable websites and provided early in the RD journey may help mitigate this challenge.

Both OS and FG participants emphasized the importance of having a foundation in genetics to prepare for appointments and for the purposes of sharing relevant information with various members of their ecosystem. Probability of information sharing was stated to be based on course functionality, and there was an interest in including guidance on best practices for sharing information. Maintenance of resources can be burdensome because of volume and content type. One FG participant keeps abbreviated information packets available for different healthcare providers for new physician appointments or emergency room visits. The development of this and future educational tools should consider the needs of the learner to not only understand the information but have the ability to accurately apply and communicate their knowledge to family members, healthcare providers, and other RD stakeholders in a variety of contexts.

OS and FG results were utilized to develop an online genetic course for the RD community. Course developers sought guidance from genetic counselors during the development and review processes. Genetic counselors play a vital role in developing reliable resources for patients and families. Key themes identified in this research may help guide genetic counselors as they create their own patient and family-facing educational content. These themes may also have relevance to other medical communication and education providers as they go beyond traditional concepts around health literacy and illustrate the importance of taking a holistic and functional approach to health education. Although specific to the RD community, many of the learning needs discussed in this study, especially

those relating to the socio-emotional state of the learner, may be applicable across various patient populations.

4.3 | Limitations

There were limitations to both the OS and FGs. Demographic information was not directly collected for the OS respondents; however, SurveyPlanet™ automatically collects location data associated with the Internet browser respondents used. These data indicated that 90% of OS respondents were in the United States at the time of their response submission. The FG participants were a convenience sample. Demographic information on FG participants was limited to country of residence and gender. Given the limited demographic information for both OS and FG participants, the results of this study may not be representative of the general RD population. Most respondents reported significant RD experience; therefore, the needs of individuals that are new to the RD journey were not well represented and may vary from more experienced counterparts. All eight FG participants were affiliated with the Global Genes Foundation Alliance member organizations. Coding and analysis for both the OS and FG output were conducted by the same researcher, and FG analysis was not validated with participants due to capacity constraints.

5 | CONCLUSION

The findings from this study have helped guide the development of a new genetic course targeted to the RD community. Research identified high levels of interest in genetic education for individuals across all stages of the RD journey. While a single course cannot deliver a clear picture of every RD, it can provide the foundation for future learnings and fill existing gaps in genetic education. Desired learning outcomes and course features reported here may be relevant to genetic education across other patient populations and, therefore, may help content developers appreciate the full scope of learning needs that may exist.

AUTHOR CONTRIBUTIONS

Laura Quinn—Ms. Quinn was the primary researcher for this study. She contributed substantially to the design, execution, and analysis. Ms. Quinn contributed substantially to the manuscript preparation, review, and final manuscript approval. She agrees to be accountable for the integrity of this study. Ms. Quinn has full access to the study data and takes responsibility for ensuring accuracy of data analysis and maintenance of data records. Kendall Davis, M.P.H.—Ms. Davis contributed substantially to the design, execution, and analysis of the study. She contributed to the manuscript preparation through review and final manuscript approval. Ms. Davis agrees to be accountable for the integrity of this study. Ashley Yee, M.S.W.—Ms. Yee contributed substantially to the design, execution, and analysis of the study. She contributed to the manuscript preparation through review and final manuscript approval. Ms. Yee agrees to be accountable for the integrity of this study. Ms. Yee has full access to the study data and takes responsibility for ensuring accuracy of data analysis and maintenance of data records. Holly Snyder, M.S., L.C.G.C.—Ms. Snyder contributed substantially to the analysis of the study data. She also contributed substantially to the manuscript preparation, review process, and final manuscript approval. Ms. Snyder agrees to be accountable for the integrity and accuracy of the data presented.

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COMPLIANCE WITH ETHICAL STANDARDS

Conflicts of interest

Laura Quinn is an employee of PRA Health Sciences. Ms. Quinn has no conflicts of interest to disclose. Kendall Davis is an employee of PRA Health Sciences. Ms. Davis has no conflicts of interest to disclose. Ashley Yee is an employee of Global Genes. Ms. Yee has no conflicts of interest to disclose. Holly Snyder is an employee of and holds equity in Illumina, Inc.

Human studies and informed consent

The above-referenced study, as submitted to Sterling IRB, does not appear to be in violation of any human subject protections (copy of this document available upon request).

Animal studies

No non-human animal studies were carried out by the authors for this article.

REFERENCES

Aaronson, N. L., Joshua, C. L., & Boss, E. F. (2018). Health literacy in pediatric otolaryngology: A scoping review. *International Journal of Pediatric Otorhinolaryngology*, *113*, 252–259.

Ayme, S., Kole, A., & Groft, S. (2008). Empowerment of patients: Lessons from the rare disease community. *Lancet*, *371*, 2048–2051.

Badiu, C., Bonomi, M., Borshchevsky, I., Cools, M., Craen, M., Ghervan, C., … Dwyer, A. A. (2017). Developing and evaluating rare disease educational materials co-created by expert clinicians and patients: The paradigm of congenital hypogonadotropic hypogonadism. *Orphanet Journal of Rare Diseases*, *12*(1), 1–9. https://doi.org/10.1186/s13023-017-0608-2

Beagley, L. (2011). Educating Patients: Understanding Barriers, Learning Styles, and Teaching Techniques. *Journal of Perianesthesia Nursing*, *26*(5), 331–337. https://doi.org/10.1016/j.jopan.2011.06.002

Bergeron, B. (2004). Online patient-education options. *MedGenMed: Medscape General Medicine*, *6*(2), 54.

Bick, D., Jones, M., Taylor, S. L., Taft, R. J., & Belmont, J. (2019). Case for genome sequencing in infants and children with rare, undiagnosed or genetic diseases. *Journal of Medical Genetics*, *56*(12), 783–791. https://doi.org/10.1136/jmedgenet-2019-106111

Budych, K., Helms, T. M., & Schultz, C. (2012). How do patients with rare diseases experience the medical encounter? Exploring role behavior and its impact on patient-physician interaction. *Health Policy*, *105*(2–3), 154–164. https://doi.org/10.1016/j.healthpol.2012.02.018

Cohen, J. S., & Biesecker, B. B. (2010). Quality of life in rare genetic conditions: A systematic review of the literature. *American Journal of Medical Genetics, Part A*, *152*(5), 1136–1156. https://doi.org/10.1002/ajmg.a.33380

Currie, G., & Szabo, J. (2019). It is like a jungle gym, and everything is under construction. The parent's perspective of caring for a child with a rare disease. *Child: Care, Health and Development*, *45*, 96–103.

EURORDIS (2018) What is rare disease. Retrieved from https://www.eurordis.org/content/what-rare-disease.

Eurodis: Rare Diseases Europe. (2015). *About rare diseases*. Retrieved from https://www.eurordis.org/about-rare-diseases.

Ferreira, C. R. (2019). The burden of rare diseases. *American Journal of Medical Genetics Part A*, *179*(6), 885–892. https://doi.org/10.1002/ajmg.a.61124

Fox, S.. Peer-to-peer healthcare: many people – especially those living with chronic or rare diseases – use online connections to supplement professional medical advice. Pew Internet and American Life Project: February 2011. Retrieved from https://www.pewinternet.org/wp-content/uploads/sites/9/media/Files/Reports/2011/Pew_P2PHealthcare_2011.pdf

Genetic Alliance (2018). Resources. Retrieved from http://www.geneticalliance.org/content/resources

Global Genes (2015). Rare Facts. Retrieved from https://globalgenes.org/rare-diseases-facts-statistics/

Hamilton, J. G., Hutson, S. P., Frohnmayer, A. E., Hanh, P. K., Peter, J. A., Carr, A. G., & Alter, B. P. (2015). Genetic information-seeking behaviors and knowledge among family members and patients with inherited bone marrow failure syndromes. *Journal of Genetic Counseling*, *24*(5), 760–770. https://doi.org/10.1007/s10897-014-9807-3

Kakazu, R., Schumaier, A., Minoughan, C., & Grawe, B. (2018). Poor Readability of AOSSM Patient Education Resources and Opportunities for Improvement. *Orthopaedic Journal of Sports Medicine*, *6*(11), 1–6. https://doi.org/10.1177/2325967118805386

Kole, A., & Faurisson, F. (2009). The Voice of 12,000 Patients. Health (San Francisco), 324. Retrieved from http://books.google.com/books?hl=en&lr=&xml:id=13g4hrVIBnoC&oi=fnd&pg=PT322&dq=The+Voice+of+12,000+Patients.+Experiences+and+Expectations+of+Rare+Disease+Patients+on+Diagnosis+and+-Care+in+Europe&ots=sik6-vhGmm&sig=qaHclWwfj9lcKHbWf.

Lewis, J., Snyder, M., & Hyatt-Knorr, H. (2017). Marking 15 years of the Genetic and Rare Diseases Information Center. *Translational Science of Rare Diseases*, *2*(1–2), 77–88. https://doi.org/10.3233/trd-170011

Limb, B. L., & Nutt, S. (2010). Experiences of Rare Diseases: An Insight from Patients and Families About Rare Disease UK. *Rare Diseases UK*, (December), 37.

Litzkendorf, S., Babac, A., Rosenfeldt, D., Schauer, F., Hartz, T., Lührs, V., & Frank, M. (2016). Information Needs of People with Rare Diseases - What Information Do Patients and their Relatives Require? *Journal of Rare Disorders: Diagnosis & Therapy*, *2*(2), 1–11.

Madathil, K. C., & Greenstein, J. S. (2018). An investigation of the effect of anecdotal information on the choice of a healthcare facility. *Applied Ergonomics*, *70*, 269–278. https://doi.org/10.1016/j.apergo.2018.03.010

Madathil, K. C., Rivera-Rodriguez, A. J., Greenstein, J. S., & Gramopadhye, A. K. (2015). Healthcare information on YouTube: A systematic review. *Health Informatics Journal*, *21*(3), 173–194. https://doi.org/10.1177/1460458213512220

Molster, C., Urwin, D., Di Pietro, L., Fookes, M., Petrie, D., Van Der Laan, S., & Dawkins, H. (2016). Survey of healthcare experiences of Australian adults living with rare diseases. *Orphanet Journal of Rare Diseases*, *11*(1), 1–12. https://doi.org/10.1186/s13023-016-0409-z

National Institute of Health Genetics Home Reference (2019). Help Me Understand Genetics. Retrieved from http://ghr.nlm.nih.gov/primer

National Organization for Rare Disorders (2019). Patient and caregiver resource center. Retrieved from https://rarediseases.org/for-patients-and-families/information-resources/patient-and-caregiver-resource-center/

Nguengang Wakap, S., Lambert, D. M., Olry, A., Rodwell, C., Gueydan, C., Lanneau, V., … Rath, A. (2019). Estimating cumulative point prevalence of rare disease: Analysis of the Orphanet database. *European Journal of Human Genetics*, *28*(2), 165–173. https://doi.org/10.1038/s41431-019-0508-0

Nutt, S., & Limb, L. (2011). Survey of patients' and families' experiences of rare diseases reinforces calls for a rare disease strategy. *Social Care and Neurodisability*, *2*, 195–199.

Pauer, F., Litzkendorf, S., Göbel, J., Storf, H., Zeidler, J., & Von Der Schulenburg, J. M. G. (2017). Rare diseases on the internet: An assessment of the quality of online information. *Journal of Medical Internet Research*, *19*(1), 1–11. https://doi.org/10.2196/jmir.7056

Pinto, D., Martin, D., & Chenhall, R. (2016). The involvement of patient organisations in rare disease research: A mixed methods study in Australia. *Orphanet Journal of Rare Diseases*, *11*(1), 1–15. https://doi.org/10.1186/s13023-016-0382-6

Schoof, M. L., & Wallace, L. S. (2014). Readability of American Academy of Family Physicians patient education materials. *Family Medicine*, *46*, 291–293.

Talwar, D., Tseng, T. S., Foster, M., Xu, L., & Chen, L. S. (2017). Genetics/genomics education for nongenetic health professionals: A systematic literature review. *Genetics in Medicine*, *19*(7), 725–732. https://doi.org/10.1038/gim.2016.156

Topol, E. (2019). Preparing the healthcare workforce to deliver the digital future: An independent report on behalf of the Secretary of State for Health and Social Care. Leeds, UK: Health Education England. Retrieved from: http://topol.hee.nhs.uk/

Wittink, H., & Oosterhaven, J. (2018). Patient education and health literacy. *Musculoskeletal Science & Practice*, *38*, 120–127. https://doi.org/10.1016/j.msksp.2018.06.004

SUPPORTING INFORMATION

Additional supporting information may be found online in the Supporting Information section.

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