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Using the diffusion of innovations model to guide participant engagement in the genomics era

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Abstract

Exome and genome sequencing (EGS) are increasingly the genetic testing modalities of choice amongst researchers owing to their ready availability, low cost, and large data output. Recruitment of larger, more diverse cohorts into long-term studies with extensive data collection is fundamental to the success of EGS research and to the widespread benefit of genomic medicine to various populations. Effective engagement will be critical to meeting this demand. The Diffusion of Innovation (DOI) model provides a framework for how new technologies are adopted in communities, including antecedents of an individual's decision to adopt the technology, how the technology's attributes influence its acceptability, the predictors of sustained use of that technology, and its diffusion through society. We apply the DOI model to frame participant engagement in EGS research and to guide the proposal of potential strategies that aim to overcome forecasted challenges. Finally, we suggest a variety of ways genetic counselors can apply their skills and training to the development and implementation of these strategies.

Keywords

exome sequencing; genome sequencing; engagement; diffusion of innovation; early adopters; diversity

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Katie L. Lewis, Flavia M. Facio, and Courtney D. Berrios each meet the International Committee of Medical Journal Editors criteria for authorship including:

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- Final approval of the version to be published; AND
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Conflict of Interest

Katie L. Lewis, Flavia M. Facio, and Courtney D. Berrios declare that they have no conflict of interest.

Introduction

Much has been written about the implications of increased use of exome and genome sequencing (EGS) for informed consent (Facio, Sapp, Linn, & Biesecker, 2012) and result disclosure policies (Berg, Khoury, & Evans, 2011; Green et al., 2013), but little attention has been given to EGS's repercussions for participant engagement. Engagement is a component of research that is of growing value due to its association with improved recruitment rates (Domecq et al., 2014), consistency with ethical principles (Ellis & Kass, 2017), potential to improve the utility of findings from research (Goddard et al., 2012), and participant demand for it (Terry, 2017). What constitutes participant engagement varies greatly in terms of the time commitment made by subjects and the amount of influence they exert on the study's preparation, execution and translation (Shippee et al., 2013). At the most basic level, engagement refers to recruiting participants to serve as subjects in a cross-sectional study. On the other end of the spectrum, participants may engage as co-investigators on a project, contributing significant time and effort to all stages of the research process.

We posit that the increasing use of EGS is altering engagement in two ways. First, this shift makes *broader* engagement even more critical. Realizing the promise of genomic medicine for all people will require the capacity to predict phenotypic consequences of genomic variants found in individuals from a variety of racial and ethnic groups. This is dependent on the availability of more diverse study cohorts and databases than those that exist (Corbin et al., 2018; Lindor, Thibodeau, & Burke, 2017; Mills & Haga, 2014). Thus, it is imperative that future engagement efforts target individuals from communities that have traditionally been under-represented in genomics research. Second, the optimization of EGS will require *long-term* engagement of participants. A longitudinal approach to data collection is critical to improving our understanding of penetrance in a genotype-first approach to medicine (Corbin et al., 2018). It also allows researchers to communicate with participants about their results over time, which is uniquely important considering the volume of data generated by EGS and the potential for periodic variant reanalysis as evidence changes (Richards et al., 2015).

Although genetics is becoming increasingly visible to the public and a majority in a recent poll said they are interested in at least one kind of DNA test (AP-NORC, 2018), EGS is still unfamiliar to the majority and interactions with research study staff may provide a first exposure. The novelty of EGS must be a central consideration when identifying a useful framework for planning engagement strategies. Therefore, we propose the application of the Diffusion of Innovations (DOI) model (Rogers, 2003) to forecast how engagement may change in the genomics era and discuss how genetic counselors can use this model to guide engagement with a broader pool of potential participants over long-term studies.

Diffusion of Innovation Model

The DOI model (Rogers, 2003) integrates several fields of research into a framework for how new technologies are adopted by individuals and communities. The model posits five phases of an individual's decision to adopt a technology (Figure 1): knowledge, persuasion, decision, implementation and confirmation. During the knowledge and persuasion phases, an

individual becomes aware of a new technology, learns about it and considers it in the context of their personal and community's values. These two phases are useful for identifying opportunities to expand the breadth of participants who are engaged in EGS research. Once a decision is made to adopt the innovation, an individual actively employs it and may learn additional practical information about its use during the implementation phase. Lastly, in the confirmation phase, the individual seeks information supporting or refuting their decision, which leads to continuing or discontinuing the innovation's adoption. The last two phases are useful for identifying potential mechanisms for promoting long-term engagement. In addition, the Model describes the diffusion of the technology through communities over time (Figure 2). This centers around five categories of individuals that subsequently adopt innovations: innovators, early adopters, early majority, late majority, and laggards. Each group has common characteristics that provide insight into their adoption decisions. These categories are useful for thinking about how engagement strategies may need to be refined as subsequent adopter groups are enrolled in EGS research.

Broadening EGS Research Engagement: Knowledge & Persuasion

In Rogers' model, the knowledge phase consists of both awareness that a technology exists and understanding of what it can offer. Although some individuals actively seek out EGS research, most only become aware of EGS research if it is offered to them through clinical care, research participation or marketing campaigns. Therefore, to recruit large and diverse populations, researchers must engage individuals who are not seeking EGS. At the most basic level, researchers can boost awareness by recruiting in more diverse communities. One study found that a common reason that African-Americans had not joined genetics research studies was that they had not been asked to do so (Millon Underwood, Buseh, Kelber, Stevens, & Townsend, 2013). Another strategy for overcoming barriers to awareness is using active recruitment techniques, such as sending personalized letters to individuals on a patient registry, having healthcare providers recruit their patients, or partnering with existing healthcare delivery systems. A recent survey of over 1,500 participants in the Henry Ford Health System Insights Community found that 72–76% of participants would be likely to participate in a genetics research study if asked by their doctor (Wagner, Peltz-Rauchman, Rahm, & Johnson, 2016). A review article also found that active strategies that reached out to potential participants from a known resource, such as a registry or clinic database, were more effective than impersonal strategies for recruiting racial and ethnic minorities to cancer genetics research (Hughes et al., 2004). One possible reason that personalized recruitment strategies work well is that participants are more likely to listen to initial recruitment messages that come from a trusted provider. Trust in an existing, clinical provider can extend to trust in healthcare research and may be a key factor in recruiting diverse populations into genomic research (Kraft et al., 2018; Persaud & Bonham, 2018). Thus, where and how researchers make participants aware of their studies is likely to have impacts on the breadth of participants they successfully engage.

Once participants are aware of EGS research, researchers must provide them with sufficient information so that they can choose whether or not to participate. Genomics knowledge levels vary amongst the general public, but are reportedly lower amongst individuals with less education (Haga et al., 2013; Ostergren et al., 2015) without previous exposure to

genetics (Schmidlen et al., 2016) or from racial or ethnic minority groups (Haga et al., 2013; Kaphingst et al., 2012). Effective engagement strategies must account for this and tailor education to reach a greater breadth of participants. One study found that a one-on-one informed consent conversation with a genetic counselor improved knowledge about sequencing benefits and limitations, and was particularly effective for increasing knowledge about the limitations of EGS amongst participants with less than a college education (Kaphingst et al., 2012). A subsequent study showed that a 10-minute, animated educational video also produced significant improvements in knowledge (Sanderson, Suckiel, et al., 2016). Similar, brief educational interventions could be incorporated into study recruitment approaches. In addition, researchers could prioritize interactive recruitment opportunities, such as staffing a table at a community health fair, that allow for provision of education.

The second step, persuasion, can be conceptualized as the formation of attitudes. Attitudes toward genetic research and testing are largely positive (Calsbeek, Morren, Bensing, & Rijken, 2007; Etchegary, 2014; Singer, Antonucci, & Van Hoewyk, 2004). These positive attitudes may be due to the potential to learn information that is useful for one's own health or the health of one's family members and to satisfy curiosity. However, some individuals express concerns about EGS testing and research, including the confidentiality of their genetic information and protection of their privacy (Murphy & Thompson, 2009), genetic discrimination (Catz et al., 2005), or adverse psychological reactions to testing results (Catz et al., 2005; Yu, Crouch, Jamal, Tabor, & Bamshad, 2013). Furthermore, attitudes are fluid, changing based on one's feelings and beliefs about the benefits and risks of a particular behavior (Condit, 2010). This may be particularly true in the context of an unfamiliar technology, such as EGS, toward which attitudes are most likely newly-formed (Wright et al., 2014) and the social norms that typically govern one's behavior may not be obvious. Kim et al. (Kim, Cappella, & Price, 2016) found that individuals who participated in online group discussions moderated by genetic counseling or communication students had fewer negative beliefs about participating in genetics research than those not offered group discussions. The authors suggest that the online discussions may have lessened negative beliefs by providing concrete information and mitigating fears of the unknown. Having fewer negative beliefs about participation was associated with more positive attitudes toward genetic research and greater intentions to volunteer. Researchers may consider facilitating similar discussions amongst potential participants, or perhaps could achieve similar effects by encouraging participants to discuss the study with their doctor. This may be followed by individual counseling in the context of informed consent, which would give researchers the chance to elicit participants' beliefs and attitudes and to clarify misperceptions in an effort to promote an informed choice.

The DOI model indicates that persuasion is also influenced by attributes of the technology. Two attributes of EGS with relevance to participants' attitudes are its advantages over other genetic testing options and the compatibility of what EGS offers with participants' values and needs. The main advantage of EGS is its breadth, and this may be apparent to participants from the inclusion of the word "whole" in some variations of its name. For individuals with a suspected genetic condition, participation in EGS research may be seen as a 'last frontier' that enables families to feel that everything possible to find a diagnosis has been done (Halverson, Clift, & McCormick, 2016). This characteristic of EGS may be

perceived as a relative advantage which can be useful in recruiting participants, although researchers should be careful to set realistic expectations and avoid misperceptions about the technology's limitations. For similar reasons, EGS research may be perceived by individuals with suspected genetic conditions as relevant to their needs and values. Researchers can leverage EGS's breadth to optimize the alignment of their goals with the priorities of unaffected or traditionally under-represented communities, as well. One model for achieving this is provided by community-based participatory research (CBPR) principles, such as building partnerships between researchers and the community from the time of study planning. For example, EGS researchers studying Pacific Islanders living in Arkansas utilized CBPR approaches to identify diabetes as a common and substantial problem amongst the population, and achieved a 95% recruitment rate for a study focused on its genetic etiology (McElfish et al., 2017). Participants in that project emphasized that they were more willing to participate, even in intensive projects, if the topic was relevant to them. In a survey of a population-based sample in the U.S., 55% of participants agreed that research participants should 'help choose what research questions to answer' and 35% were willing to be involved in helping make those decisions (Kaufman, Baker, Milner, Devaney, & Hudson, 2016), lending further support to the feasibility of engaging community members to make research relevant to them.

Lengthening EGS Research Engagement: Implementation & Confirmation

Rogers' Model provides guidance about promoting long-term engagement after a decision has been made about EGS research, as participants implement and confirm their decision. In the implementation phase, individuals focus on the practical aspects of using a new technology. There are two practical aspects of EGS research participation that we anticipate may influence participants' willingness to engage in the long-term. First is the ongoing collection of phenotypic, behavioral, and environmental data from participants. This process is essential to the success of many EGS projects, but requires participants' engagement, and thus must be carefully crafted. Open-forum discussions amongst participants in Geisinger's MyCode study suggest that techniques that allow for automatic collection of this data from wearable devices or environmental monitors given to participants, rather than requiring participants to manually input the data, are attractive. The participants also suggested that "fun" or interactive ways to promote retention should be prioritized. For example, participants might like to receive updates on how their data is being used through online platforms or participate in online forums for generating research ideas (Wagner et al., 2016).

The second practical aspect that may influence long-term engagement is the receipt of individual results. As participants receive individual results, they have the opportunity to learn how participation in EGS research impacts their lives. The breadth of EGS testing also allows for a wide range of result return policies. Ethical and legal analyses supported by research documenting participants' positive attitudes toward receiving results and minimal psychological harms following receipt (Lewis et al., 2016) has influenced the development of policies encouraging the offering of at least some results to participants (National Academies of Sciences, Engineering and Medicine, 2018). Although many people surveyed to date have expressed an interest in receiving sequencing results (Facio et al., 2013; Kaufman, Murphy, Scott, & Hudson, 2008), individual participant preferences are not

universally positive (Jamal et al., 2017; Yu et al., 2013) and ideal engagement policies may need to allow participant choice in the return of results and use informed consent strategies that promote informed and preference-based decision making. Furthermore, continued assessment of whether EGS research meets participants' needs will promote their continued investment. This may be accomplished through CBPR methods in which community advisors formally provide continued input to the research team or through invitations for feedback from participants through study newsletters and communications.

In the confirmation stage, an individual looks for reinforcement of the decision and if positive attitudes about the innovation are not sustained, one may reverse their decision, leading to less willingness to engage in further data collection for the study, or in the extreme, withdrawal from the study. This reminds us that optimal policies for long-term engagement in EGS research will likely require intermittent reinforcement of the original decision to participate. Whether or not expectations are met may be a key aspect of this stage. Expectations of EGS are reportedly high. The potential for benefit to themselves and family members has been continually identified as a motivator by participants in EGS research studies, including in both apparently healthy populations (Facio et al., 2011; Sanderson, Linderman, et al., 2016) and those with a suspected genetic condition (Berrios et al., 2018). The potential to receive multiple results from EGS and participants' perceptions of it as the newest and most comprehensive test to identify genetic variants can lead to inflated expectations. Managing these expectations has been identified by researchers and providers as an ongoing challenge of EGS research (Wynn et al., 2018). Participants in a trial using EGS who were surveyed at the time of enrollment and six months after receipt of results reported deriving much less utility from their genetic results than they expected at the outset, particularly with regard to their ability to use the results to affect medical decision making and medication choice (Roberts et al., 2018). Unmet expectations may also have emotional impacts that could impact the confirmation stage. For example, Cacioppo et al. (Cacioppo, Chandler, Towne, Beggs, & Holm, 2016) identified that emotional responses to the return of genetic of research results were related to whether or not participants received the utility they expected from the results and those who held negative emotions felt a great deal of uncertainty about their child's future despite receiving a diagnostic EGS result. Since EGS often includes uncertainty, one key facet of building realistic expectations is anticipating and managing uncertainty. Evidence suggests that participant responses to genomic data may reflect their epistemological beliefs about the nature of genome knowledge. Researchers who frame uncertainty as expected and a source of opportunity rather than a threat (Biesecker et al., 2014) may promote more realistic expectations, and thus, sustained engagement.

Diffusion Through Communities

The DOI model also describes five groups based on how quickly they adopt a technology (Figure 2) and posits that sociodemographic characteristics and personality traits are correlated with each adopter category. Many who have enrolled in EGS research thus far, particularly apparently healthy individuals, are likely innovators and early adopters. According to DOI (Rogers, 2003), people in these groups are likely to be tolerant of uncertainty, willing to accept risk and more knowledgeable about new technology than their

peers. Some research has explored the personality traits of participants in EGS research and confirmed that these traits were common in one EGS research cohort (Lewis et al., 2015). Recognizing that the next several million participants who enroll in EGS research will likely be innovators or early adopters allows researchers to effectively appeal to these groups through targeted engagement strategies and materials. For example, framing uncertainty as a potential risk of participation may be less effective with risk-tolerant early adopters. Rather, this supports the idea that researchers may build more rapport with participants by discussing uncertainty in the context of hope and potential for benefit.

Early reports of healthy EGS cohorts suggest that the majority of participants have positive emotional reactions to receiving personal results (Sanderson et al., 2017). This could be explained by the DOI model's predictions about the traits of early adopters, such as uncertainty tolerance or resilience, which would also likely facilitate positive adaptation to genetic testing results, especially when the results are not expected. Furthermore, early adopters are less likely to have negative emotional reactions to their results, and so it may be prudent to focus less on this as a risk and better to emphasize readiness to use results for health behavior change or to explain undiagnosed symptoms. However, researchers and clinicians should also be aware that subsequent groups may have different outcomes than initial participants and continue to revise the engagement strategies and study policies that are shaped by these initial participants.

One group that may inform engagement strategies for later adopters is unaffected family members recruited to EGS cohorts due to a family member's diagnosis. Although some family members have self-selected for participation, many others may have been persuaded to enroll in spite of reservations. These individuals may fit into the DOI's early majority adopter group, which is comprised of people who are deliberate in their decisions about innovations. For instance, although parents recruited to EGS due to their child's symptoms are motivated by potential benefits to their child or family, they are primarily focused on the disease by which their child is affected (Tabor et al., 2011). Parents also report feeling a sense of duty to participate in EGS research in search of a diagnosis for their child (Rosell et al., 2016). A focus on answers for their child and a sense of duty in these groups may mean that individuals who are more skeptical and have less favorable attitudes toward science, more common characteristics in late adopters, may be enrolled in these cohorts. These parents also report more tempered expectations of EGS based on their previous experiences with genetic testing (Rosell et al., 2016), which may mirror the skepticism that is more characteristic of later adopters. While their experience with disease shapes many attitudes in disease cohorts (Hitch et al., 2014), their experiences with and responses to EGS research may provide lessons for engaging later adopter groups due to their shared characteristics within the DOI model.

The Role of Genetic Counselors

In response to the 2018 National Society of Genetic Counselors Professional Status Survey, 21% of genetic counselors reported being involved in research (The National Society of Genetic Counselors, 2018). Those self-identified research counselors and others straddling the lines of clinic and research (for example, in large academic and hospital centers) may

have job responsibilities that include participant recruitment and engagement. Genetic counselors can apply their training and skills to implement several of the suggestions based on the DOI model themselves or may take an active role in developing protocols and procedures that others implement (as, for example, in the case of large scale studies). For example, genetic counselors are trained to tailor education to meet the needs and characteristics of a wide variety of clients (Accreditation Council for Genetic Counseling, 2015) and could apply this not only to interactions with individual participants but also the development of study materials, such as recruitment brochures or informed consent documents. A similar skill is key in the DOI's knowledge phase to meet the informational needs of the diverse groups required for EGS and in adapting educational approaches to different adopter categories over time. CBPR, which may also be used to engage broader populations, requires researchers to have skills in listening, communication, conflict resolution, and self-reflection (Israel, Schulz, Parker, & Becker, 2001), which also overlap considerably with competencies of genetic counselors (Accreditation Council for Genetic Counseling, 2015). In addition, facilitating informed decision making has been a cornerstone of genetic counseling practice for decades (National Society of Genetic Counselors' Definition Task et al., 2006), and is another central aspect of participant engagement to which genetic counselors' skills can easily be applied one-on-one with participants or by developing and researching decision aids. Furthermore, genetic counselors can apply their basic and advanced counseling skills to help EGS research participants adapt to their personal results, which may facilitate long-term engagement. Their familiarity with various methods of service delivery makes them vital members of research teams that seek to develop and support innovative strategies for long-term engagement, particularly in cohorts with broad geographic distributions. These attributes are well aligned not only with genetic counselors' training and competencies, but also with their duties from the National Society of Genetic Counselor's Code of Ethics, to promote the welfare of society through responsible research while treating individuals with care and respect for autonomy, individuality, welfare, and freedom (The National Society of Genetic Counselors, 2018).

Conclusions

The potential and expanding applications of EGS in research and clinical care make it imperative that participants in research are diverse and representative of many races and ethnicities. Moreover, the shift in paradigm to a genotype-driven research approach can only be optimized with the active engagement of research participants longitudinally, over the course of years and decades.

We propose the DOI model as a framework for recruiting and engaging participants long-term, utilizing the phases an individual goes through for adopting and employing a new technology, and the diffusion of technology through communities (Rogers, 2003). Table 1 summarizes multiple techniques supported by the research literature that may be useful for engaging participants at each stage of the DOI Model. Genetic counselors are well-positioned to implement strategies based on the DOI model in their research and academic practices for a number of reasons. First, the training of genetic counselors includes competencies in listening and communicating, as well as facilitating informed decisions. In this process, genetic counselors are well adapted to tailoring their counseling and/or study

materials to the psychosocial and educational needs of their individual clients or research participants, including research participant communities in larger-scale studies. This approach is central to the recruitment and long-term support of research participants that are engaged in the research process. Second, genetic counselors in research positions are often involved with the design and implementation of recruitment strategies. As such, they may personally interact with the participants over the course of the study, supervise staff who conduct such activities, or have a role in creating study protocols. Through this wide variety of mechanisms, genetic counselors are poised to lead the wider spread of EGS research by advocating for research protocols that engage participants in a partnership and take into account individual differences in adopting new technologies. Moving forward, it would be useful to document the effectiveness of DOI-informed techniques in various populations and across distinct EGS studies.

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Adopted from Rogers 2003, p.170, Figure 5-1

Figure 1:
Innovation Decision Process

Adopted from Rogers 2003, p.281, Figure 7-1

Figure 2:
Adopter Categories over Time

Table 1.

Summary of DOI-Informed Engagement Techniques

DOI Model Phase	Central Challenges to Engagement	DOI-Informed Engagement Techniques
Knowledge	Making individuals aware of and informed about EGS research	<ul style="list-style-type: none"> ■ Use active or personalized recruitment techniques, such as recruitment through doctors or in-person at health fairs ■ Provide opportunities for education during recruitment, such as videos or one-on-one interaction ■ Use CBPR techniques to ensure that your study messages address the community's needs
Persuasion	Promoting positive attitudes toward EGS research	<ul style="list-style-type: none"> ■ Address areas of common concern, such as privacy or genetic discrimination, through policies or education ■ Allow community input on study policies that may make it more consistent with their values and needs
Implementation	Ensuring that the practical aspects of EGS research are acceptable	<ul style="list-style-type: none"> ■ Make ongoing phenotyping easy, convenient, fun and interactive ■ Offer participants choice about result return or influence of result return policies
Confirmation	Meeting participants expectations about EGS research	<ul style="list-style-type: none"> ■ Assess expectations and promote realistic ones ■ Frame uncertainty as hopeful rather than threatening