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## A New Era, New Strategies: Education and Communication Strategies to Manage Greater Access to Genomic Information

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Genomic information is challenging to communicate. The results of genome sequencing can be numerous, uncertain, and ambiguous.<sup>1</sup> The impact of many gene variants on a person's health and future can be difficult to predict, especially when no symptoms have yet emerged, in part because these effects are moderated by numerous other factors, such as a person's behaviors and environmental exposures over time. As next-generation genomic sequencing, including whole-genome sequencing information becomes more common in research, clinical, and public health contexts, there is a need for comprehensive communication strategies and education approaches to prepare patients and clinicians to manage this information and make informed decisions about its use. Nowhere is that imperative more pronounced than when genomic sequencing is applied to newborns.

Communicating a newborn's sequencing information to their parents is fraught with challenges. For parents to process the information at an already busy time in their lives, it needs to be relevant, accessible, and actionable. Yet many genomic results may not meet these criteria, a fact that raises questions about the usefulness of such information for parents and calls for communication strategies that can deal with complexity and uncertainty. Most existing communication paradigms use an in-person genetic counseling approach. Interpersonal communication is an effective way to communicate complex information because it interpersonal communication changes in real-time to meet a parent's specific needs.<sup>2</sup> Unfortunately, because this approach takes time and requires special expertise, it is not scalable. There simply are not enough genetic counselors<sup>3, 4</sup> or genetic counseling training programs to rely solely on one-to-one counseling sessions.

Even if more counselors can be trained, in-person counseling is unlikely to be applicable or cost-effective when genomic risk information is obtained by parents directly via the internet.<sup>5</sup> As a general rule, communication strategies should match how people are accessing health information. In the past, people obtained health information in clinical settings, which at least in theory, created the opportunity for in-person counseling. Today, however, many people can obtain health information in a variety of settings, including over the Internet and through direct-to-consumer services, via websites, and through other digital channels or

settings. In response to these changes, new communication strategies need to be considered. Adopting a comprehensive communication model means understanding the multiple levels of influence experienced by parents and the clinicians who serve them.<sup>6</sup> In addition, applying communication science principles can help address some key challenges to effectively communicating genomic information to parents.<sup>7</sup>

At the level of the individual, it is important to recognize that while parents in the United States vary in their beliefs, values, and knowledge about genomics, health literacy and genomic literacy are generally quite low, which limits parents' ability to understand the benefits, risks, and overarching ramifications of their child's genomic information.<sup>8</sup> One way to begin addressing these literacy issues is to ensure that the information is presented in plain language that lay people understand. Research shows that when information is made more accessible, by using plain language and applying clear communication principles, it is more effective and actionable for recipients.<sup>9</sup> Communicators should seek to identify and prioritize parental needs, including putting the most important information first, using common words, explaining technical terms, using bullets or lists, summarizing information, and using short sentences. These all represent easy, evidence-based approaches that can be implemented in digital, written, or verbal communication. Additionally, using best practices for conveying risk information—such as risk arrays and using smaller denominators (e.g., 100 vs 10,000)—and using multiple and graphical formats can help make complex information more easily understandable and accessible.<sup>10</sup> Easily understandable information can be disseminated through a variety of channels online or via paper-based methods. Information design makes the information more accessible, but does not address low population genomic literacy, which can only be enhanced by significant, long-term investments in national and community educational campaigns. These educational campaigns could be launched by state and federal agencies or large health systems or insurers.

At the interpersonal level, an important challenge is the potential need for parents to work collaboratively with each other and with clinicians to make informed and shared decisions about whether and how to act on their newborn's sequencing results. One evidence-based method to facilitate collaborative decisions is the use of decision aids,<sup>11, 12</sup> particularly if they are delivered digitally. Typically, decision aids include values clarification exercises that help parents understand what is important to them when making a decision about sequencing information and can help prepare them for clinical encounters. Emerging research suggests that decision aids can be particularly effective for supporting informed decision making among parents with lower health literacy.<sup>13</sup> Decision aids could be implemented in visits women make to clinical providers during pregnancy to prepare them for newborn screening decisions.

At the organizational level, family physicians, pediatricians, and other clinicians working in health care systems that serve pregnant women and new mothers, are currently under-prepared to support the additional complexity that sequencing may bring to the newborn period. Focused clinician education in genomic medicine needs to start in premedical education and continue through ongoing professional and specialty training for practicing clinicians.<sup>14</sup> In addition, healthcare organizations need to find ways to support clinicians in

deciding how and when to communicate with parents about sequencing. Thus far, these initiatives have included clinical decision support tools that are typically embedded in the electronic health record and intended to guide providers as they discuss results and next steps.<sup>15</sup> Such systems require complex informatics and input from multiple stakeholders to be useful in clinical care.<sup>16</sup> How they can be implemented effectively in genomic studies, particularly in the pediatrics field, is an active area of research<sup>17</sup> and should include studies to determine their utility.

At a community level, the many settings parents visit in their pregnancy journey could be used to disseminate information and potentially gather input. While communication strategies that can be used in clinical contexts are important, parents are likely to learn about and discuss newborn sequencing in other settings as well. Communication science principles call for “meeting people where they are” by understanding where and how they seek information, what types of communication channels they use, and what sources parents view as trustworthy. Today, these channels invariably include the internet, social media, and other information accessible via mobile devices, none of which have yet been adequately integrated into efforts to communicate sequencing results. Doing so could support and prepare parents for encounters in clinical contexts and address any questions that may emerge afterward.

Finally, at the policy level, we note that the absence of protections for parents and their newborns has the potential to limit the type of information parents would want to receive or make the information less actionable once they do receive it. Parents have valid concerns about the possible acquisition and storage of genome sequencing data by state newborn screening programs and about the inclusion of sequencing information in their child’s medical record.<sup>18–20</sup> These concerns could deter parents from accepting offers of sequencing, thereby stymieing research and practice in this area. Policy changes are needed to protect families who need or desire to learn genomic information from privacy violations and discrimination.

Communication research focusing on how genomic information should be communicated to parents of newborns is in its infancy. More resources must be devoted to this research area to help fill the large translational gap in this area. Most research funds often go to basic science discovery versus to solving translational issues.<sup>21</sup> This has left the parents and the clinicians who serve them without access to the tools, knowledge, and resources to collaboratively make preference-sensitive, family-centric decisions about a newborn’s genomic information.<sup>22, 23</sup> To assist families, healthcare providers, companies and others tasked with communicating sequencing information we must apply evidence-based communication science practices and work to develop new tools that can effectively and efficiently communicate sometimes complex and uncertain genomic information. Application of these practices and development of new tools will help alleviate some of the ethical concerns related to providing sequencing information. We also recommend that education about genomics be incorporated in settings that already provide education to clinicians and parents. In addition, larger scale awareness and educational campaigns need to be developed, implemented and evaluated to address low genetic literacy levels in the community as a

whole. Advancing the field of genomics means advancing both education and communication strategies to fill the large translational gap in this area.

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