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Impact of the increased adoption of prenatal cfDNA screening on non-profit patient advocacy organizations in the United States

Stephanie Meredith, Christopher Kaposy, Victoria J. Miller, Megan Allyse, Subhashini Chandrasekharan, and Marsha Michie on behalf of the Prenatal Testing PAG Coalition*

Abstract

The “Stakeholder Perspectives on Noninvasive Prenatal Genetic Screening” Symposium was held in conjunction with the 2015 annual meeting of the International Society for Prenatal Diagnosis. During the day-long meeting, a panel of patient advocacy group (PAG) representatives discussed concerns and challenges raised by prenatal cfDNA screening, which has resulted in larger demands upon PAGs from concerned patients receiving prenatal cfDNA screening results. Prominent concerns included confusion about the accuracy of cfDNA screening and a lack of patient education resources about genetic conditions included in cfDNA screens. Some of the challenges faced by PAGs included funding limitations; lack of consistently implemented standards of care and oversight; diverse perspectives among PAGs and questions about neutrality; and lack of access to training and genetic counselors. PAG representatives also put forward suggestions for addressing these challenges, including improving educational and PAG funding and increasing collaboration between PAGs and the medical community.

Introduction

Cell-free DNA screening tests (cfDNA) to detect fetal chromosomal conditions became commercially available in 2011. Sometimes referred to as “non-invasive prenatal screening

Corresponding author: Marsha Michie, UCSF Institute for Health & Aging, 3333 California St., Laurel Heights 340, San Francisco, CA 94118, 415-502-6491, marsha.michie@ucsf.edu.

*Prenatal Testing Patient Advocacy Group (PAG) Coalition:

Megan A. Allyse, Assistant Professor, Mayo Clinic, Rochester, MN

Linzee Carroll, President, 11q Research and Resource Group, Woodstock, GA

Subhashini Chandrasekharan, Assistant Professor, Duke University, Durham, NC

Virginia Isaacs Cover, Volunteer, Association for X and Y Chromosome Variations, Pine, CO

Cori Feist, Genetic Counselor, Oregon Health and Science University, Portland, OR

Janalee Heinemann, Coordinator of Research and International Affairs, Prader-Willi Syndrome Association, Sarasota, FL

Christopher Kaposy, Associate Professor, Memorial University of Newfoundland, St. Johns, Newfoundland

Heidi Lindh, Executive Director, Genetic Support Foundation, Richardson, TX

Stephanie Meredith, Information Services Director, National Center for Prenatal and Postnatal Down Syndrome Resources, University of Kentucky, Louisville, KY

Marsha Michie, Assistant Professor, University of California, San Francisco, CA

Victoria J. Miller, Executive Director, Trisomy 18 Foundation, Dale City, VA

Mollie A. Minear, Postdoctoral Associate, Duke University, Durham, NC

Vardit Ravitsky, Associate Professor, University of Montreal, Montreal, Quebec

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tests,” “NIPS,” or “NIPT,” prenatal cfDNA screening may detect trisomies 13, 18, and 21; sex chromosome aneuploidies (such as Turner and Klinefelter syndromes); and fetal sex. Some cfDNA screens advertise the additional detection of sub-chromosomal genetic deletions that lead to conditions such as Prader-Willi/Angelman syndromes on chromosome 15q, 22q11.2 deletion syndrome, 4p- syndrome (Wolf-Hirschhorn), 5p- syndrome (Cri-du-Chat), and 11q deletion (Jacobsen syndrome).

In July 2015, members of the Prenatal Information Research Consortium organized the “Stakeholder Perspectives on Noninvasive Prenatal Genetic Screening” Symposium, held in conjunction with the annual meeting of the International Society for Prenatal Diagnosis. Attendees and presenters included representatives of patient advocacy organizations, researchers, bioethicists, physicians, and genetic counselors. Attendees also included industry representatives, although representatives from commercial cfDNA laboratories were not invited as presenters, primarily to avoid the potential for bias if only a subset of those laboratories agreed to participate. Among the goals of the Stakeholders Symposium was to discuss the impact of the introduction and expansion of cfDNA screening on disability and patient advocacy organizations (PAGs). Symposium participants addressed some of the concerns and challenges of PAGs in keeping pace with the rapid growth of the prenatal genetic screening industry in the US, including funding limitations; a lack of consistently implemented standards for pre- and post-test counseling and information; diverse perspectives and questions about neutrality; and limited access to training and genetic counseling services. Below we outline these concerns and offer suggestions for addressing them.

Filling the gaps

Because the US Food and Drug Administration (FDA) has historically declined to regulate laboratory developed tests, cfDNA screening has (like many other genetic tests) entered the US market without FDA review and approval.¹ Laboratory-funded studies have demonstrated high test sensitivity and specificity for the detection of Down syndrome in pregnancies at elevated risk for fetal aneuploidy.^{2–5} However, independent or clinic-based studies have placed some of these early findings into context, emphasizing positive predictive value, incidences of false positive results, and other test limitations not highlighted in earlier industry-funded studies.^{6–8} Moreover, the limited data regarding cfDNA screening for rarer aneuploidies and microdeletions indicate lower sensitivity and specificity than that for Down syndrome and other common trisomy conditions, posing challenges for clinicians assessing the positive predictive values of cfDNA screening and for patients who may receive screen-positive results for these rarer conditions.^{9, 10} In addition, aggressive marketing campaigns have contributed to misperceptions among some patients and clinical providers that cfDNA screens are diagnostic or “near-diagnostic.”¹¹ At the Stakeholders Symposium, PAG panelists reported that their organizations often fielded panicked calls or emails from prospective parents who had received cfDNA screen-positive results from their providers that they understood as definitive. Despite professional guidelines that strongly recommend confirmatory diagnostic testing, research also suggests that some women have elected to terminate pregnancies solely on the basis of cfDNA screening results, potentially aborting unaffected fetuses.^{11–13}

Providers and patients also face challenges in accessing genetic counseling to help them prepare for and interpret screening results. Various commentators have noted the lack of capacity of existing genetic counselors and genetic counseling training programs to meet the increasing demand for these professionals, particularly in pre-test counseling.^{14, 15} While some companies that market cfDNA screening offer genetic counseling services, concerns have been expressed regarding conflicts of interest for company-employed genetic counselors. In addition, obstetricians and family practitioners, who already have heavy demands on their clinical time, may lack both time and training to help patients understand cfDNA screening options and results. General practitioners may also have limited knowledge and experience of the social and clinical profile of individuals with rare genetic conditions. To fill these systemic gaps, many pregnant women turn to non-profit PAGs for assistance. For example, women who have received a positive cfDNA screening result for trisomy 18, many of whom mistakenly believed their result was diagnostic, have often sought out the Trisomy 18 Foundation (trisomy18.org) for help interpreting and figuring out what to do with these results. As cfDNA screening expands to more pregnancies and rarer conditions, even more women and families are likely to seek out these support organizations for answers.

The patient advocacy community has made efforts to address the increasing volume of patient questions prompted by cfDNA screening by fielding questions, connecting expectant parents with parent mentors, and creating or contributing to patient education materials that cover both medical and psychosocial outcomes. Studies have demonstrated that pregnant women and their families want comprehensive and balanced information about the full spectrum of life with a genetic condition, including clinical features, developmental abilities, reproductive options, range of potential outcomes, and available support services; and condition-specific advocacy groups have both a rich lived experience and, often, the most comprehensive collection of clinical and other information about those conditions.^{16, 17} However, the capacity of PAGs to provide support for expectant parents and widely disseminate resources is limited by the challenges and constraints outlined below.

Funding Limitations

Advocacy groups often devote considerable time and resources to helping patients understand prenatal screening results, advocating for patients and their families, creating and publishing educational materials, facilitating and funding research, and staying up-to-date on the constantly evolving prenatal testing landscape. However, PAG funding limitations pose significant challenges. During the Stakeholders Symposium, Linzee Carroll, president of the 11q Research & Resource Group (11qusa.org), shared the experience of her small group representing those with Jacobsen syndrome in fielding numerous emails from patients with questions about prenatal screening tests, distributing printed information about the condition, and providing emotional support for expectant parents, all while raising money through bake sales and other grassroots fundraising efforts. Virginia Cover, from the Association for X and Y Chromosome Variations (AXYS), pointed out that she appreciates that some cfDNA laboratories list her organization's website (genetic.org) as a patient resource. However, questions from patients about prenatal screening results consume 4–6 hours per week from her completely unpaid staff, not including time spent on later support or follow-up. In fact,

many of the PAGs that find themselves counseling parents about cfDNA screening, such as the 11q Research and Resource Group, rely primarily on volunteer staff.¹⁸

These resource strains are compounded by the fact that many leaders of patient advocacy organizations are also parents, some with affected children whose disabilities may require significant time and advocacy. Victoria Miller, Executive Director of the Trisomy 18 Foundation, echoed the feelings of several advocates at the Stakeholders Symposium when she expressed her frustration that commercial testing labs, which expend considerable resources on marketing highly profitable tests, subsequently rely on non-profit PAGs with already overcommitted time and over-extended resources to help customers assimilate and follow up on test results. Indeed, the financial disparities are stark: one testing lab reported an annual revenue of \$151 million in 2014 (<https://www.genomeweb.com/business-news/sequenom-reports-preliminary-q4-revenue-37m>), while the 11q Research and Resource Group reported \$58,756 in annual revenue (and no paid staff) for that same year (guidestar.org).

Some laboratories have made efforts to address these funding disparities. For example, Ariosa purchased awareness materials from the Trisomy 18 Foundation to be distributed at the 2013 National Society for Genetic Counselors conference; Natera initiated an awareness and fundraising campaign for the International 22q11.2 Foundation in 2014 (<http://www.reuters.com/article/ca-natera-inc-idUSnBw125535a+100+BSW20140512>); and Sequenom contributed to the printing and distribution of materials for the Global Down Syndrome Foundation/National Down Syndrome Congress (<http://www.downsyndrometest.org/distribution-announced-for-down-syndrome-prenatal-testing-pamphlet/>). Nonetheless, such financial support is currently *ad hoc* rather than standardized and sustainable. Moreover, commercial funding may create ethically problematic conflicts of interest for PAGs, who would prefer to be perceived as patient- and family-centered sources of information. Financial power imbalances and a desire to remain independent make it challenging for PAGs to negotiate with testing laboratories for financial support to respond to the growing demands on advocacy groups caused by cfDNA screening.

In 2008 the unanimously-passed Prenatally and Postnatally Diagnosed Conditions Awareness Act (Kennedy-Brownback Bill) authorized the Secretary of Health & Human Services to provide \$25 million over 5 years in grants for the development of patient education resources around prenatal and neonatal conditions (Pub. L. No. 110–374, 2008). However, since the passage of the law only one appropriation—of less than US\$1 million—has been made (<http://www.grants.gov/custom/viewOppDetails.jsp?oppId=47902>). Systemic strains from the lack of a cohesive patient education strategy, already in evidence when the law was passed 7 years ago, have only intensified with the availability of cfDNA screening.

Lack of Consistently Implemented Standards

PAGs who support expectant parents are further hindered by the lack of regulatory oversight (whether from governmental or professional bodies) regarding cfDNA test information; the lack of oversight regarding pre- and post-test counseling as recommended by professional societies; and the lack of oversight and consistency regarding the provision of information

about screened genetic conditions. Marketing materials and informed consent documentation from testing laboratories may include test information that has not been published or independently verified; and these resources usually focus on the high test sensitivity for the detection of trisomy 21 in high-risk pregnancies—rarely acknowledging lower sensitivity for other conditions or positive predictive values for any condition or population.^{19, 20} Professional clinical guidelines for providing information about the capabilities and limitations of prenatal cfDNA screening are relatively consistent across organizations.^{21–23} However, written materials specifically geared toward helping patients understand cfDNA screening have only recently been published by professional societies.^{24, 25} Moreover, these resources must be sought out by clinicians or patients, in contrast to the abundant distribution of laboratory-produced marketing materials.

In addition, professional guidelines regarding pre- and post-test counseling for cfDNA screening are often implemented inconsistently and without oversight.²⁶ Guidelines from professional societies (e.g., the American College of Medical Genetics and Genomics,²¹ the American Congress of Obstetricians and Gynecologists/Society for Maternal-Fetal Medicine,²² and the International Society for Prenatal Diagnosis²³) all recommend pre- and post-test counseling for prenatal cfDNA screening and the provision of patient education resources. However, symposium participants reported inconsistencies in the implementation of these recommendations by prenatal providers. And providers who want to provide such counseling and information are often hindered by a lack of available data and resources. Only recently have organizations released tools that enable clinicians to more reliably report positive predictive values of cfDNA screening for common aneuploidies; a useful example of such a tool is the online NIPT Predictive Value Calculator, available from the Perinatal Quality Foundation (<https://www.perinatalquality.org/Vendors/NSGC/NIPT/>).

This lack of oversight and regulation may impact even more strongly the availability and quality of information about specific conditions screened via cfDNA, and PAGs often step in to fill those gaps. Laboratories that market cfDNA screening are not required by any governmental or professional bodies to supply educational materials to health care providers; those that do frequently refer consumers and providers to PAGs for information about screened conditions. Professional societies have recently offered recommendations for medically-reviewed patient education materials about Down syndrome.^{21–23} However, most professional societies do not make specific recommendations about resources for rarer conditions, aside from a recommendation to contact relevant PAGs. Patients testifying in support of recent Down Syndrome Information Acts (now passed in 15 US states) have spoken about their dissatisfaction with the materials and support they received from clinicians.²⁷ And research by Nelson Goff *et al.* has found that 35% of those receiving a prenatal diagnosis of Down syndrome indicated a negative experience of post-test support from their provider, compared to only 11% reporting a positive experience.²⁶ Similarly, in a 2008 study by Walker *et al.* of 19 families receiving a prenatal diagnosis of trisomy 18, over half reported a lack of written resources and support group information; and those families who opted to continue the pregnancy often reported feeling unsupported by their provider.²⁸ A gap clearly exists between the support and information patients are receiving from medical providers following prenatal testing and what patients want to receive. It is crucial that PAGs and medical organizations collaborate regarding the development and distribution

of patient education materials, and establish clinical training curricula that address the gap between prenatal counseling best practices and the realities of current clinical experiences with the introduction of prenatal cfDNA screening.

Diverse perspectives and questions about neutrality

The diversity of perspectives among PAGs also poses challenges for providing reliable and up-to-date information to expectant parents. Individuals and families affected by a genetic condition may be represented by a number of PAGs with differing agendas, perceptions, and methodologies. When the PAGs for a condition convey different messages, particularly in regard to reproductive choices, it can be difficult for both testing laboratories and the medical community to establish guidelines.

When engaging in dialogues about prenatal testing and inevitable conversations about post-test reproductive options, PAGs often encounter conflicts between the perspectives of prospective parents during pregnancy and those of families who live with a genetic condition. Consider the case of the Down Syndrome Consensus Group, which convened in 2008 at the University of South Carolina and included representatives of ACOG, ACMG, NSGC, and national Down syndrome PAGs. This consensus group worked to establish common ground between the patient advocacy and medical communities in order to better meet the needs of patients receiving a diagnosis of Down syndrome, and all members of the consensus group agreed that “[i]nformation and counseling provided to parents regarding a prenatal or postnatal genetic diagnosis should be complete, consistent, non-judgmental, and non-coercive,” and should include all reproductive options, including elective termination.²⁹ Less than four years later, the executive director of the National Down Syndrome Congress, which had been part of the consensus group, stated, “[W]e don’t feel it’s appropriate to promote the value of those with Down syndrome while at the same time also discussing the possibility of abortion. ... Our self-advocates told us that it was not appropriate in a pamphlet coming from their advocacy organization to talk about abortion as co-equal to any other option.” (<http://www.firstthings.com/web-exclusives/2012/11/the-down-syndrome-communitys-abortion-rift>)

PAGs for many conditions face similar cultural tensions in their communities as they strive to respect the views of individuals living with genetic conditions and their families, while supporting prospective parents who may be exploring reproductive options. Moreover, medical professionals often struggle to balance clinical data on the health impacts of genetic conditions with contextual information about the positive experiences of families raising children with those conditions. Healthcare providers report that they are sometimes reluctant to distribute materials created by patient support groups because of concerns that these materials may provide an overly rosy perspective about life with the condition.³⁰ Therefore, it is important to establish neutral entities, such as non-profit foundations like the Genetic Support Foundation (geneticsupportfoundation.org) and academic centers like the National Center for Prenatal and Postnatal Resources at the University of Kentucky (lettercase.org), where all stakeholders can engage in meaningful conversations. These organizations are uniquely placed to generate balanced patient education materials about prenatal testing and

genetic conditions to reflect collective concerns without compromising the diverse needs of patients and their supporters as the primary target audience.

Lack of access to training and genetic counselors

Another challenge facing PAGs in this expanding era of prenatal testing is the lack of medically trained personnel to support expectant parents of children with a genetic condition. Given the technical nature of screening results, the wide variation in how genetic conditions may manifest, and regional inequities in public services and support, counseling expectant parents through the decision-making process is far from simple. Given the strained resources of most PAGs, obtaining the services of a trained genetic counselor, or seeking educational opportunities for other staff members or volunteers, is beyond the financial means of most advocacy organizations. When lacking such qualifications, members of PAGs must be cautious about crossing the line into providing medical advice.

Moving forward

Overall, members of the patient advocacy community have expressed appreciation that some cfDNA companies refer customers to PAGs, particularly after a screen-positive result; however, the success of these companies has driven more referrals and increasing workloads for PAGs without providing needed financial support or other resources to meet this demand.

A few cfDNA laboratories have allocated small amounts of funding, mostly for distribution agreements for educational materials. We maintain, however, that cooperative efforts should be undertaken to ensure that all patients who are offered cfDNA screening receive up-to-date and accurate information about screened genetic conditions, and that patients receive consistent amounts and types of information, regardless of which laboratory's test they use. Such a cooperative effort might take several forms: a suggestion emerged from the Stakeholders Symposium that a clearing-house organization might be developed to receive funds from multiple testing companies and distribute them to individual organizations and/or multi-organization collaborative efforts to support and inform expectant parents.

PAGs serve a vital role in providing support, community, and condition-specific information for families with a pregnancy affected by a genetic condition. As PAGs are on the front lines of actual patient experiences with cfDNA screening results and variably-implemented screening protocols, all stakeholders would benefit from the inclusion of these groups in discussions about the most effective and ethical ways to implement this new technology. In order to facilitate informed and values-appropriate decision-making, we urgently need to bring all stakeholders together to discuss how best to ensure appropriate use of cfDNA screening and provide the requisite support services for parents who receive screen-positive results. As we continue to strategize the best ways to support families and PAGs impacted by cfDNA screening, we welcome and invite collaborations and connections with others engaged in this work from all sectors, and encourage future efforts that will continue this dialogue between PAGs and other stakeholders.

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What's already known about this topic?

- Prenatal cfDNA screening has strongly impacted the provision of prenatal testing for fetal genetic conditions.
- Non-profit patient advocacy groups often provide support for potential and new parents and families who have received a genetic diagnosis.

What does this study add?

- Non-profit patient advocacy groups face new challenges as a result of the proliferation of prenatal cfDNA screening, particularly in providing education and support for potential and new parents and families who have received a genetic diagnosis.
- Representatives of patient groups have proposed several remedies for the challenges they face, including independent funding mechanisms and collaborations with other stakeholders in prenatal screening and testing.