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Commentary

Recommendations to improve the patient experience and avoid bias when prenatal screening/testing

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ABSTRACT

While prenatal screening and testing have expanded substantially over the past decade and provide access to more genetic information, expectant parents are more likely to describe the diagnosis experience as negative than positive. In addition, the conversations that take place during these experiences sometimes reflect unconscious bias against people with disabilities. Consequently, an interdisciplinary committee of experts, including people with disabilities, family members, disability organization leaders, healthcare and genetics professionals, and bioethicists, reviewed selected published and gray literature comparing the current state of the administration of prenatal testing to the ideal state. Subsequently, the interdisciplinary team created recommendations for clinicians, public health agencies, medical organizations, federal agencies, and other stakeholders involved with administering prenatal screening and testing to create better patient experiences; conduct training for healthcare professionals; create, enforce, and fund policies and guidelines; and engage in more robust data collection and research efforts.

Prenatal screening and testing have expanded substantially over the past decade, giving access to more accurate results about potential disabilities earlier during a pregnancy. However, research has shown that prenatal testing can lead to poor clinical experiences when expectant parents do not receive adequate support and resources about conditions upon receiving screening results.[1,2] In addition, the delivery of the results can inadvertently project bias against people with disabilities when health professionals do not

Abbreviations: SB, Spina Bifida; DS, Down syndrome; CDHPD, Center for Dignity in Healthcare for People with Disabilities.

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adequately navigate the delicate intersection between reproductive and disability rights.[3] Therefore, clinicians and expectant parents can benefit from recommendations "to provide effective support and communication throughout the decision-making processes and follow-up care" when discussing disabilities.[4]

Research indicates that expectant parents learning about a range of prenatally diagnosed conditions often do not receive adequate resources about prenatal screening or disabilities. Studies show that the reading levels of pamphlets informing patients about prenatal screening are typically higher than recommended for public health education and do not address multicultural needs.[5] Additionally, parents of children with Trisomy 18,[6] Spina bifida (SB) and hydrocephalus,[7] Down syndrome (DS),[1] and congenital heart defects[8] have indicated that they often do not receive sufficient information about those conditions following screening results. Approximately 20 states have passed DS or Genetic Conditions Information Acts to attempt to address these critical patient educational needs through public policy measures.[9]

Most expectant parents also conveyed that they want to learn more about life outcomes, recreation and employment opportunities, and supports and services, reflecting the **social model of disability**—which defines disabilities in terms of how society eliminates barriers and offers adaptations for people with disabilities.[10] Parents describe their diagnosis experience as positive in studies when they perceived the delivery as balanced, sensitive, and compassionate; were given a range of non-directive reproductive options; and received accurate, balanced, and up-to-date resources and information promptly about the condition and available supports and services.[1,6–8] However, healthcare professionals tend to focus on medical and genetic issues when providing information about a prenatal diagnosis, conveying a **medical model of disability** where disabilities are defined as problems to be cured.[10] Expectant parents expressed that elements of both the social and medical models of disability are essential when learning about a prenatal diagnosis.

When these patient needs are not met, studies have indicated long-term negative emotional outcomes and trauma for parents of children with DS and SB caused by poor diagnosis experiences.[2,7] Nelson-Goff et al. found that expectant parents receiving a prenatal diagnosis of children with Down syndrome (DS) described their experience with medical professionals as negative three times more often than positive.[1] Over a range of studies, expectant parents described their experience as negative when they perceived that medical professionals shared negative stereotypes about disabilities, did not offer accurate and balanced information about conditions, pressured pregnancy decisions, or did not exhibit compassion.[1,2,4,6,7]

Unfortunately, prenatal screening can inadvertently reflect biases about disabilities from clinicians. For example, a recent study found that 82.4% of clinicians believe people with significant disabilities have a poorer quality of life than nondisabled people.[3] However, this perception is not broadly supported by research regarding family and self-perceptions about living with disability. Research shows that parents rated the health-related quality of life of their children with severe disabilities significantly higher than their physicians[11] and that 99% of people with DS surveyed said they were happy with their lives.[12] Thus, perceptions of disabilities by healthcare providers likely reflect biases that do not accurately represent the experiences of people living with conditions and their families.

Since its inception, prenatal testing and diagnosis have warranted concern and questions from the disability community, but technological developments and clinical adoption have outpaced ethical discussions and decisions, especially those that center experiences of people with disabilities.[13] Additionally, experiences of parents with heritable disabilities, who bring their own nuanced views of disability to diagnostic exchanges with providers, are underrepresented and sometimes excluded from research about diagnosis experiences, which has largely centered on nondisabled parents as the recipients of unexpected prenatal diagnoses. However, essays and interviews by disabled scholars have posed ethical disagreements with non-disabled providers who present biased prenatal information about disabilities to disabled parents who have lived experience.[14] Adrienne Asch notably raised ethical concerns about the impact of prenatal testing on people with disabilities as a historically marginalized population and advocated for both the inclusion of people with disabilities in the development of guidelines and the inclusion of the social model of disability in genetic counseling discussions about parenting.[13]

Consequently, clinicians and expectant parents can benefit from recommendations, created by a broad-based stakeholder team, "to provide effective support and communication throughout the decision-making processes and follow-up care" when discussing disabilities.[4] The Dobbs v. Jackson Women's Health Organization Supreme Court decision in June 2022 has made the development of such recommendations even more critical as states limiting reproductive rights are more likely to have families in crisis who will be continuing a pregnancy and will need immediate information about available supports and service for people with disabilities.[15] Conversely, clinicians in states with fewer reproductive restrictions will need further guidance on how to avoid bias in discussions about disabilities while assuring patients of their reproductive rights.[16]

Within the context of this rapidly changing national landscape, the interdisciplinary prenatal subcommittee of the Center for Dignity in Healthcare for People with Disabilities (CDHPD)[§] and CDHPD staff reviewed selected published and gray literature[17] comparing the current state of the administration of prenatal testing to the ideal state, and created recommendations for clinicians, public health agencies, medical organizations, federal agencies, and other stakeholders involved with administering prenatal screening and testing to: 1. create better patient experiences; 2. conduct training for healthcare professionals; 3. create, enforce, and fund policies and guidelines; and 4. engage in more robust data collection and research efforts.[17]

Process

With the expansion of prenatal screening, healthcare professionals are increasingly called upon to meet complex patient informational needs, preserve patient autonomy in decision-making, and avoid bias against people with disabilities as a historically marginalized population.[13,14] Therefore, these recommendations aim to improve the patient experience while also promoting equity toward people with disabilities in the administration of prenatal screening/testing. Although existing prenatal screening and testing guidelines have been developed by national medical and genetics organizations, most did not include input from stakeholders in the disability community.

Toward that end, the following recommendations were uniquely developed for healthcare providers by an interdisciplinary

[§] CDHPD partners include: The University of Cincinnati Center for Excellence in Developmental Disabilities (UCCEDD); The Maryland Center for Developmental Disabilities, the Boggs Center on Developmental Disabilities; the University of Kentucky Human Development Institute; the Vanderbilt Kennedy Center for Excellence in Developmental Disabilities; Family Voices; The Autistic Self-Advocacy Network; and The American Academy of Developmental Medicine and Dentistry. Recommendations reflect the opinions of experts participating in the prenatal subcommittee and do not infer official positions of institutions.

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committee of experts including people with disabilities, family members, disability organization leaders, healthcare and genetics professionals, and bioethicists comprising the members of the prenatal subcommittee of the CDHPD. The 21 members of this committee were invited and selected by the CDPHD staff and partners. In addition, these recommendations are informed by a review of 76 selected documents recommended by the prenatal subcommittee "such as organizational statements, policy guidance documents, media coverage, governmental reports, existing curricula, and research literature to identify gaps and investigate the impact of discrimination and disparities in the healthcare of people with ID/DD."[17]

Recommendations

Create better patient experiences

Recommendation 1: ***Discuss with expectant parents the potential medical issues associated with prenatally-diagnosed conditions and also the broader spectrum of life outcomes for people with prenatally diagnosed conditions*** *(including physical, developmental, educational and psychosocial outcomes, life expectancy, clinical course and intellectual and functional development, and treatment options).*[18]

This approach underscores both the social and medical models of disability as described above. To avoid inadvertently conveying outdated stereotypes about people with disabilities, verify that the information discussed is accurate and up-to-date.

Directed to: Healthcare and genetics professionals; other patient education health professionals

Recommendation 2: Share information about available supports and services, including early intervention services, local and national diagnosis-specific advocacy groups, and/or local family connections with lived experience.[4,6,10]

Provide information about national parent support organizations such as the National Parents First Call Center, Parent to Parent USA, Family Voices, the Disabled Parenting Project, and condition-specific advocacy groups which can offer a family-to-family support network of trained parent mentors.

Directed to: Healthcare and genetics professionals; other patient education health professionals

Recommendation 3: ***Provide expectant parents with up-to-date, evidence-based, and unbiased informational resources about disabilities***, which are developed collaboratively between medical experts/representatives of national medical and genetics organizations and disability experts/representatives of patient advocacy groups. Specifically, research shows that expectant parents prefer a range of information about medical issues, life outcomes, supports and services, and photography of people living with the conditions when learning about disabilities.[10,19] Resources should be provided at the same time screening and testing results are delivered. Generally, patient-centered information resources should be evaluated for readability, comprehensibility, and communicative effectiveness, and resources should be culturally and linguistically appropriate and available in multiple languages.[5]

Guidelines from the American College of Genetics and Genomics and the National Society of Genetic Counselors specifically recommend various patient education resources, such as the American Academy of Pediatrics (AAP) "Health Supervision for Children with Down Syndrome" and resources from the Lettercase National Center for Prenatal and Postnatal Resources (Lettercase), created with input from the national medical, genetics, and advocacy organizations.[20,21] To successfully implement the provision of patient education resources about conditions among providers, national medical and genetics organizations need to provide clear guidance to clinicians about strategies and resources to meet the informational needs of expectant parents and assist with the dissemination of patient education resources.

Directed to: Healthcare and genetics professionals; healthcare and genetics organizations; testing laboratories; patient advocacy organizations

Recommendation 4: ***When delivering a prenatal diagnosis, offer support and compassion through reflective listening, but avoid saying "I'm sorry" or assuming the patient will perceive the diagnosis as bad news—unless the life of a child is imminently at risk for death or severe health complications***.

Apologies convey an unconscious bias about disabilities when expectant parents may have a range of views based on their own value systems and life experiences. Instead, use value neutral language such as "I have news" or "We have received your results" when discussing prenatal screening results.[22]

Directed to: Healthcare and genetics professionals

Recommendation 5: ***Provide pre-test and post-test counseling as an essential part of any screening program in understandable language, and offer a thorough explanation of results by doing the following:***

a. Discuss pregnancy preferences and values when presenting prenatal screening options to help expectant parents make informed choices about whether to proceed with prenatal screening or not.
b. Provide expectant parents with information about tests in multiple mediums based on their needs, health literacy, language, and cultural preferences to ensure informed consent.[23]
c. Explain the limitations of prenatal screening, as well as the interpretation of results. For example, sensitivity can tell patients how often a screen can detect a condition in the whole population, but positive predictive value is the probability the screen is accurate for an individual patient.[23]
d. If patients choose to undergo screening and receive results suggesting a higher chance for genetic conditions, discuss pregnancy preferences and values to guide conversations about possible diagnostic testing and pregnancy management.[22]
e. When expectant parents receive patient education resources about a condition and clearly communicate a decision about whether or not to continue a pregnancy, avoid repeated questions about pregnancy decisions, second guessing reproductive decisions, or assuming that a pregnancy decision is not well-informed. Empower parents to direct the conversations and set the parameters based on their preferences.[22]

Directed to: Healthcare and genetics professionals; test manufacturers and laboratories

Recommendation 6: ***Provide follow-up appointment referrals and coordination of care among specialists*** and ensure clear communication about family preferences regarding birth plans to the team so that all providers communicate effectively.[22]

Directed to: Healthcare professionals

Healthcare, professional training, and accountability

Recommendation 7: ***Consult and/or partner with leaders from relevant disability advocacy organizations when developing any training materials that address disabilities*** to ensure materials reflect the social and civil rights context for understanding disability. Training materials should also include up-to-date life outcome information.

Recommended partner organizations ideally include those that represent the community addressed in the training materials, including individuals with disabilities who have lived experience

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and family members. Be mindful to develop authentic relationships with these advocacy leaders as partners to receive meaningful feedback and avoid tokenism.

Directed to: Healthcare professionals; health educators; medical educational accreditation entities; disability advocacy organizations; professional medical associations

Recommendation 8: ***Include comprehensive training on how to deliver a sensitive and non-directive prenatal diagnosis*** in clinical training for healthcare providers and continuing professional education for any health professional involved in prenatal care,[24] and consider requiring this skill training as a component of licensure. Online simulation has already been developed through Lettercase/Brighter Tomorrows to practice these skills.[24,25] While medical students often receive instruction on how to deliver "bad news," discussions about disabilities are more nuanced and complex when avoiding bias against a stigmatized population. For example, research shows a clinician should avoid saying "I'm sorry" when delivering a diagnosis that a person can consider as part of their identity, such as Down syndrome or achondroplasia; however, empathy and condolences might be appropriate if a child has a life-threatening condition or is undergoing major surgery.

Directed to: Healthcare professionals; health educators; medical education and advanced practice nursing program accreditation entities and licensing and certification organizations

Recommendation 9: ***Provide clinicians with a comprehensive education about the science of prenatal testing, including the limitations, strengths and weaknesses of prenatal screening and testing, as well the interpretation of results.***[26] ***Subsequently, provide clinicians with training on communication strategies to share information about prenatal testing with expectant parents***, including discussing testing and genetics in plain language; using culturally sensitive language and terms regarding the disabilities being discussed; making appropriate referrals to genetics professionals and other specialists for further testing and consults; and sharing information about local and national patient advocacy organizations.

Directed to: Healthcare professionals; medical education and advanced practice nursing program accreditation entities and licensing and certification organizations; health educators; genetic counselors; social workers.

Recommendation 10: ***Develop mandatory requirements for disability education and cultural awareness in the educational training and continuing education of healthcare and genetics professionals*** who provide patient education related to prenatal testing and diagnosis.[27] Specifically, these entities should consider the following:

a. Promote disability awareness that includes current information about conditions.
b. Build genuine relationships with the disability community.
c. Recruit people with disabilities and family members to be incorporated into the education and training of healthcare professionals.
d. Incorporate disability studies, with concepts such as the models of disability, the history of disability civil rights, and ableism, into medical, genetics, and nursing training curricula.
e. Develop measurable outcomes to evaluate disability-related curricula objectives.
f. Examine conscious and unconscious bias about disabilities.
g. Encourage the use of "Family-Team" training models in which individuals with disabilities and their family members share their first-hand life experiences and recommendations and dialogue with current and future healthcare providers.

Directed to: Medical and advanced practice nursing program accreditation boards and licensing and certification organizations; National Institute of Health

Recommendation 11: ***Incentivize medical organizations and national disability advocacy organizations (such as Special Olympics, The Arc, and other disability organizations) to collaborate and partner toward identifying, accessing, supporting, and disseminating quality public awareness campaigns in multimedia formats that showcase the lived experiences of people with disabilities.***

Healthcare professionals who have an understanding about real-life experiences of people with disabilities and their families are in a better position to support expectant parents undergoing prenatal testing.

Directed to: Advocacy organizations; medical organizations; media

Create, enforce, and fund policies and guidelines

Recommendation 12: ***Support policies to ensure that expectant parents get the information they need by implementing the following federal and state policies.*** Research indicates that these policies are most effective when they are specifically pro-information/neutral initiatives that are not prescriptive about reproductive decision-making; when they include funding; when they include accountability measures beyond voluntary and independent efforts; and when they include reporting mechanisms.[27]

a. ***Advocate for funding the Kennedy-Brownback Prenatally and Postnatally Diagnosed Conditions Awareness Act.*** This act was passed unanimously and was supposed to provide funding to "provide up-to-date, comprehensive information about life expectancy, development potential, and quality of life for a child born with" a prenatally or postnatally diagnosed condition; strengthen networks of support; improve data collection; and ensure the provision of accurate information about testing. However, the $25 million for this Act was never appropriated.[18]
b. ***Implement and collaborate on the dissemination of information about state DS/Genetic Conditions Information Acts to ensure expectant parents receive the recommended information about genetic conditions and available supports and services.***

For example, the Washington and Massachusetts state departments of public health directly disseminate resources from Lettercase and work collaboratively with state medical and advocacy organizations on dissemination; and Ohio, Nebraska, and Illinois disseminate the DS Fact Sheet template developed collaboratively between the National Society of Genetic Counselors DS Information Act Working Group and Lettercase.

Clinicians should also be aware of any state requirements regarding the provision of information as outlined in any pertinent DS or Genetic Conditions Information Acts.[9]

Directed to: Public health authorities; policy leaders; national healthcare organizations

Recommendation 13: ***Develop enforceable Sunshine and Conflict-of-Interest laws that will bring transparency to any financial relationships among genetic counselors, healthcare providers, and commercial laboratories.***

Directed to: Public health authorities; policy leaders; national healthcare organizations

Recommendation 14: ***When creating guidelines, recommendations, and practice bulletins about prenatal testing, national medical, nursing, and genetics organizations should explicitly include recommendations for clinicians to do the following for expectant parents with a likely prenatal diagnosis***:

a. Offer information about local and national patient advocacy organizations.

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b. Provide medically accurate, family-centered, evidence-based, and up-to-date information about prenatally-diagnosed conditions, including supports and services; medical issues; and life outcome information about psychosocial outcomes, life expectancy, and social/educational/vocational outcomes.[9] Recommended materials should be developed with input from representatives of medical, genetics, and disability advocacy organizations.[28] For example, the NSGC and ACMG position statements specifically identify patient education resources: Down syndrome pregnancy, Genetics Home Reference, Genetic Support Foundation, AAP Guidelines, and the Lettercase/The National Center for Prenatal and Postnatal Resources.[20,21]

Directed to: Professional healthcare associations; healthcare and genetics professionals

Recommendation 15: ***Develop measured and ethically balanced approaches to prenatal and carrier screening by including disability stakeholders*** when determining which conditions to incorporate in panels. Likewise, ***the development of prenatal guidelines, recommendations, and practice bulletins should include representation from leaders in the disability community*** as stakeholders to achieve health equity. Leadership in the disability community available to consult include: CDHPD, National Organization for Rare Disorders (NORD), Association of University Centers on Disability, American Academy of Developmental Medicine & Dentistry (AADMD), Genetic Alliance, NIH condition groups, Self Advocates Becoming Empowered (SABE), or condition-specific advocacy groups if guidelines are specific.

Directed to: Professional healthcare associations; disability advocacy organizations; federal agencies; public health authorities

Recommendation 16: ***Actively regulate and oversee the marketing claims and practices of prenatal genetic testing companies to ensure that the Positive Predictive Value (PPV), negative predictive value (NPV) and utility of prenatal tests are accurately reported with transparency.***

Many patients and some clinicians misunderstand marketing claims about sensitivity to mean PPV, so patients need understandable information about PPV to understand the likelihood of a disability for each pregnancy.

Directed to: Federal agencies and regulatory authorities; professional healthcare and genetics associations

Recommendation 17: ***Advocate for policies that allow providers to bill insurance for genetic counseling as a reimbursable service that is fully covered by healthcare insurance programs***, including the Affordable Care Act and Medicaid for women as a standard of care provided during pregnancy. Genetic counseling should be provided by an ABGC-board certified genetic counselor when possible. It is critical that health insurance companies understand their role as partners and stakeholders and join in advocacy and education efforts by "broadening reimbursement policies for patient education provided in physician offices" including the cost of educational infrastructure in the cost of care."[29] The COVID-19 pandemic has demonstrated that through methods like telehealth, genetic counseling services can be more easily accessible. Establishing this protocol can help reduce health disparities and inequities.

Directed to: Health insurance companies; policymakers; NIH; professional healthcare and genetics organizations

More robust data collection and research efforts

Recommendation 18: ***Support multi-disciplinary research*** to understand the following (see Table 1):

Directed to: Healthcare professionals; funding *sources for social science research; testing laboratories.*

Table 1
Research priorities.

Research priority	Why
Examine experiences, choices, and needs of families receiving results.	Determine differences and similarities in needs depending on conditions, race and culture, economic needs, and other social determinants of health.
Survey physician perspectives on delivering news to families.	Contrast most and least effective practices and identify potential biases.
Informed-decision making strategies to help parents find support after making whatever decision they may.	Determine what spaces, resources, tools and organizations are most helpful for parents making a range of reproductive decisions.
Strategies for supporting a pregnancy following a prenatal diagnosis for each condition.	Research has historically focused on Down syndrome and needs to expand to other conditions.

Recommendation 19: ***Research the relationship between prenatal testing, patient outcomes, and various factors in the field of genetic testing and obstetric care***, such as genetic counseling, cultural conditions, social expectations, and social determinants of health for particular disability communities. We want to determine how these factors impact decisions about prenatal testing, patient experiences upon receiving screening results, reproductive decisions, and access to services and healthcare across the lifespan. ***Research how various models of prenatal genetic counseling (including who delivers these services and who pays for them) impacts prenatal testing choices to illuminate any possible conflicts of interest in counseling services provided by testing companies.***

Directed to: Policy makers; professional healthcare associations

Recommendation 20: ***Conduct research on the attitudes of healthcare providers regarding prenatally-diagnosed conditions and strategies to improve healthcare workforce training to counter implicit biases.***[3]

Directed to: Funding *sources for social science research; healthcare professionals; medical and advanced practice nursing program accreditation boards and licensing and certification organizations; disability advocacy organizations; professional medical, genetics, and nursing associations*

Conclusion

Moving into a future where prenatal testing continues to expand, reproductive rights are in flux, and people with disabilities are leading social justice movements, these concepts are running into a collision course unless the medical and advocacy communities thoughtfully engage on how to best support expectant parents learning about a prenatal diagnosis. These recommendations—created with input from people with disabilities, family members, disability organization leaders, healthcare and genetics professionals, and bioethicists and grounded in representation and equity—provide a framework for health professionals to balance the needs of various stakeholders when engaging in prenatal testing, including, including patients, people with disabilities, health professionals, and the broader public. It is also vitally important that these recommendations be shared with families and self-advocates; to promote equity, the CDHPD also provides accessible Public and Easy Read versions of these prenatal recommendations at www.centerfordignity.org.

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These recommendations were collaboratively developed with equal input by the interdisciplinary Prenatal Subcommittee of the Center for Dignity in Healthcare for People with Disabilities.

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Conflicts of interest

Ms. Meredith serves on the board of the Genetic Support Foundation in a volunteer capacity and is the parent of an adult with Down syndrome. She has also received remuneration from Down syndrome non-profit organizations and non-profit medical/genetics organizations/programs (American College of Osteopathic Obstetricians and Gynecologists and American Society of Human Genetics) for speaking engagements and associated travel expenses.

Ms. Brackett is the mother of a child with Down syndrome.

Dr. Diaz is the father of a child with Down syndrome.

Dr. Freeman is the mother of an adult with autism.

Ms. Khan is the mother of a non-verbal son with global and developmental delays.

Mr. Leach is the father of a daughter with Down syndrome.

Mr. Levitz is an advocate with Down syndrome.

Dr. Skotko occasionally consults on the topic of Down syndrome through Gerson Lehrman Group. He receives remuneration from Down syndrome non-profit organizations for speaking engagements and associated travel expenses. This past year, Dr. Skotko received annual royalties from Woodbine House, Inc., for the publication of his book, *Fasten Your Seatbelt: A Crash Course on Down Syndrome for Brothers and Sisters*. Within the past two years, he has received research funding from F. Hoffmann-La Roche, Inc., AC Immune, and LuMind Research Down Syndrome Foundation to conduct clinical trials for people with Down syndrome. Dr. Skotko is occasionally asked to serve as an expert witness for legal cases where Down syndrome is discussed. Dr. Skotko serves in a non-paid capacity on the Honorary Board of Directors for the Massachusetts Down Syndrome Congress and the Professional Advisory Committee for the National Center for Prenatal and Postnatal Down Syndrome Resources. Dr. Skotko has a sister with Down syndrome.

Ms. Smith is the CDHPD Project Coordinator and has achondroplasia.

Ms. Onufer is the mother of a daughter with Spina Bifida.

Dr. White is the mother of a child with Down syndrome.

Dr. Ayers is the CDHPD Project Director, has Osteogenesis imperfecta, and is the mother of a child with achondroplasia.

The authors have no other conflicts of interest to disclose.

References

1. Nelson Goff BS, Springer N, Foote LC, et al. Receiving the initial Down syndrome diagnosis: a comparison of prenatal and postnatal parent group experiences. *Intellect Dev Disabil*. 2013;51(6):446–457. https://doi.org/10.1352/1934-9556-51.6.446.
2. May CP, Dein A, Ford J. New insights into the formation and duration of flashbulb memories: evidence from medical diagnosis memories. *Appl Cogn Psychol*. 2020;34(5):1154–1165. https://doi.org/10.1002/acp.3704.
3. Iezzoni LI, Rao SR, Ressalam J, et al. Physicians' perceptions of people with disability and their health care: study reports the results of a survey of physicians' perceptions of people with disability. *Health Aff (Millwood)*. 2021;40(2):297–306. https://doi.org/10.1377/hlthaff.2020.01452.
4. Hodgson J, McClaren BJ. Parental experiences after prenatal diagnosis of fetal abnormality. *Semin Fetal Neonatal Med*. 2018;23(2):150–154. https://doi.org/10.1016/j.siny.2017.11.009.
5. Kloza EM, Haddow PK, Halliday JV, O'Brien BM, Lambert-Messerlian GM, Palomaki GE. Evaluation of patient education materials: the example of circulating cell free DNA testing for aneuploidy. *J Genet Couns*. 2015;24(2):259–266. https://doi.org/10.1007/s10897-014-9758-8.
6. Walker LV, Miller VJ, Dalton VK. The health-care experiences of families given the prenatal diagnosis of trisomy 18. *J Perinatol*. 2008;28(1):12–19. https://doi.org/10.1038/sj.jp.7211860.
7. Chaplin J, Schweitzer R, Perkoulidis S. Experiences of prenatal diagnosis of Spina bifida or hydrocephalus in parents who decide to continue with their pregnancy. *J Genet Couns*. 2005;14(2):151–162. https://doi.org/10.1007/s10897-005-0488-9.
8. Carlsson T, Bergman G, Wadensten B, Mattsson E. Experiences of informational needs and received information following a prenatal diagnosis of congenital heart defect: experiences following a prenatal diagnosis of congenital heart defect. *Prenat Diagn*. 2016;36(6):515–522. https://doi.org/10.1002/pd.4815.
9. Center for Dignity in Healthcare for People with Disabilities. PRENATAL LAWS: Down Syndrome Information Acts. https://centerfordignity.com/wp-content/uploads/2021/07/Prenatal-Laws-Down-Syndrome-Information-Acts.pdf. Accessed August 25, 2022.
10. Sheets KB, Best RG, Brasington CK, Will MC. Balanced information about Down syndrome: what is essential? *Am J Med Genet A*. 2011;155(6):1246–1257. https://doi.org/10.1002/ajmg.a.34018.
11. Nolan R, Luther B, Young P, Murphy NA. Differing perceptions regarding quality of life and inpatient treatment goals for children with severe disabilities. *Acad Pediatr*. 2014;14(6):574–580. https://doi.org/10.1016/j.acap.2014.02.012.
12. Skotko BG, Levine SP, Goldstein R. Self-perceptions from people with Down syndrome. *Am J Med Genet A*. 2011;155(10):2360–2369. https://doi.org/10.1002/ajmg.a.34235.
13. Parens E, Asch A. The disability rights critique of prenatal genetic testing. Reflections and Recommendations. *Hastings Cent Rep*. 1999;29(5):S1–S22.
14. Stramondo JA. Disability and the damaging master narrative of an open future. *Hastings Cent Rep*. 2020;50:S30–S36. https://doi.org/10.1002/hast.1153.
15. Kirkegaard D. The Disproportionate Harm of Abortion Bans: Spotlight on Dobbs v. Jackson Women's Health. Center for Reproductive Rights. Published November 29, 2021 https://reproductiverights.org/supreme-court-case-mississippi-abortion-ban-disproportionate-harm/. Accessed August 25, 2022.
16. Reardon S. Genetic Screening Results Just Got Harder to Handle Under New Abortion Rules. Kaiser Health News. Published June 27, 2022 https://khn.org/news/article/genetic-screening-results-just-got-harder-to-handle-under-new-abortion-rules/. Accessed August 25, 2022.
17. Center for Dignity in Healthcare for People with Disabilities. Healthcare Discrimination and Inequities Facing People with Disabilities: A Gap Analysis. Published online October 14, 2020 https://centerfordignity.com/wp-content/uploads/2021/02/CDHPD-Gap-Analysis-Report.pdf. Accessed August 25, 2022.
18. Text of S. 1810 (110th): Prenatally and Postnatally Diagnosed Conditions Awareness Act (Passed Congress version). GovTrack.us. Accessed August 25, 2022. https://www.govtrack.us/congress/bills/110/s1810/text.
19. Levis DM, Harris S, Whitehead N, Moultrie R, Duwe K, Rasmussen SA. Women's knowledge, attitudes, and beliefs about Down syndrome: a qualitative research study. *Am J Med Genet A*. 2012;158A(6):1355–1362. https://doi.org/10.1002/ajmg.a.35340.
20. Sheets KB, Crissman BG, Feist CD, et al. Practice guidelines for communicating a prenatal or postnatal diagnosis of Down syndrome: recommendations of the national society of genetic counselors. *J Genet Couns*. 2011;20(5):432–441. https://doi.org/10.1007/s10897-011-9375-8.
21. Gregg AR, Skotko BG, Benkendorf JL, et al. Noninvasive prenatal screening for fetal aneuploidy, 2016 update: a position statement of the American College of Medical Genetics and Genomics. *Genet Med*. 2016;18(10):1056–1065. https://doi.org/10.1038/gim.2016.97.
22. Skotko BG, Kishnani PS, Capone GT, for the Down Syndrome Diagnosis Study Group. Prenatal diagnosis of Down syndrome: how best to deliver the news. *Am J Med Genet A*. 2009;149A(11):2361–2367. https://doi.org/10.1002/ajmg.a.33082.
23. Kuppermann M, Pena S, Bishop JT, et al. Effect of enhanced information, values clarification, and removal of financial barriers on use of prenatal genetic testing: a randomized clinical trial. *JAMA*. 2014;312(12):1210. https://doi.org/10.1001/jama.2014.11479.
24. Lunney CA, Kleinert HL, Ferguson JE, Campbell L. Effectively training pediatric residents to deliver diagnoses of Down syndrome. *Am J Med Genet A*. 2012;158A(2):384–390. https://doi.org/10.1002/ajmg.a.34422.
25. Jackson L, Cichon M, Kleinert H, Trepanier A. Teaching medical students how to deliver diagnoses of Down syndrome: utility of an educational tool. *Patient Educ Couns*. 2020;103(3):617–625. https://doi.org/10.1016/j.pec.2019.10.011.
26. Oxenford K, Daley R, Lewis C, Hill M, Chitty LS. Development and evaluation of training resources to prepare health professionals for counselling pregnant women about non-invasive prenatal testing for Down syndrome: a mixed methods study. *BMC Pregnancy Childbirth*. 2017;17(1):132. https://doi.org/10.1186/s12884-017-1315-7.
27. Lehman A, Leach M, Santoro SL. Delivering a new diagnosis of Down syndrome: parent experience. *Am J Med Genet A*. 2021;185(12):3615–3622. https://doi.org/10.1002/ajmg.a.62408.
28. National Council on Disability. Genetic Testing and the Rush to Perfection: Part of the Bioethics and Disability Series. 100.
29. Leach MW. The Down syndrome information act: balancing the advances of prenatal testing through public policy. *Intellect Dev Disabil*. 2016;54(2):84–93. https://doi.org/10.1352/1934-9556-54.2.84.

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