Stephanie Meredith

Directed Reading Course: Disability and Public Health

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July 28, 2021

Annotated Bibliography

1. **Physicians’ Perceptions Of People With Disability And Their Health Care**

*Iezzoni, L. I., Rao, S. R., Ressalam, J., Bolcic-Jankovic, D., Agaronnik, N. D., Donelan, K., ... & Campbell, E. G. (2021). 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 Affairs, 40(2), 297-306.*An article summarizing the conscious and unconscious bias faced by people with disabilities when trying to access health care. N=714 physicians
2. **Delivering a new diagnosis of Down syndrome: Parent experience**

*Lehman, A., Leach, M., & Santoro, S. L. (2021). Delivering a new diagnosis of Down syndrome: Parent experience. American Journal of Medical Genetics Part A.*An article about the impact of Down Syndrome Information Acts on the diagnosis experiences of patients in Ohio. No impact was detected. N=79 prenatal parents (76 White; 3 Other)
3. **Women’s Knowledge, Attitudes, and Beliefs About Down Syndrome: A Qualitative Research Study**

*Levis, D. M., Harris, S., Whitehead, N., Moultrie, R., Duwe, K., & Rasmussen, S. A. (2012). Women's knowledge, attitudes, and beliefs about Down syndrome: A qualitative research study. American Journal of Medical Genetics Part A, 158(6), 1355-1362.*

Article about the results when focus groups of women who were pregnant or about to become pregnant explored their knowledge, attitudes, and beliefs about Down syndrome; resources used to obtain information about health and DS; and information needs on the topic of DS. N=109 (54 White; 55 Other)

1. **ACMG statement on noninvasive prenatal screening for fetal aneuploidy**

*Gregg, A. R., Best, R. G., Monaghan, K. G., Bajaj, K., & Skotko, B. G. (2013). ACMG statement on noninvasive prenatal screening for fetal aneuploidy. congenital anomalies, 16, 19.*

2016 position statement from the American College of Medical Genetics and Genomics about the implementation of cell-free DNA/non-invasive prenatal screening, including the limitations of NIPS as a technology and recommendations for pre-test and post-test counseling.

The document is helpful in actually stating that condition-specific education should be part of post-test counseling and naming examples of resources: "Accurate, up-to-date, and balanced information about Down syndrome (or other tested conditions) should be provided. There are a number of resources available (see Resources)."

1. **Genetic Testing and the Rush to Perfection Part of the Bioethics and Disability Series**

*National Council on Disabilities (2019). Genetic Testing and the Rush to Perfection: Part of the Bioethics and Disability Series.* [*https://ncd.gov/sites/default/files/NCD\_Genetic\_Testing\_Report\_508.pdf*](https://ncd.gov/sites/default/files/NCD_Genetic_Testing_Report_508.pdf)

Genetic Testing and the Rush to Perfection examines the impact of genetic testing on people with disabilities and on disability communities, examines the range of scientific, commercial, medical/ professional, and social factors that converge around prenatal genetic testing as it affects people with disabilities, and also provides an update on the interaction between genetic testing and employment discrimination. It concludes with recommendations aimed at greater federal and state oversight and quality control of genetic tests, and improving genetic counselor education on disability.

1. **Beyond the Genetic Diagnosis: Providing Parents What They Want to Know**

*Saul, R. A., & Meredith, S. H. (2016). Beyond the Genetic Diagnosis: Providing Parents What They Want to Know. Pediatrics in review, 37(7), 269-278.*

Clinicians need to provide accurate, up-to-date, and balanced information to parents following a prenatal or postnatal diagnosis of Down syndrome and other genetic conditions. Families want information about the genomic outcomes and medical issues, but they also want information about life outcomes and social supports. Because the anticipated outcomes of a condition can change significantly based on available social support, health care, and services, it is important for clinicians to stay up-to-date about new developments and credible, medically reviewed information about Down syndrome and other genetic conditions to access resources for clinical care.

1. **Impact of the increased adoption of prenatal cfDNA screening on non-profit patient advocacy organizations in the United States***Meredith, Stephanie, et al. "Impact of the increased adoption of prenatal cfDNA screening on non‐profit patient advocacy organizations in the United States." Prenatal diagnosis 36.8 (2016): 714-719.*
2. **NSGC Practice Guidelines for Communicating a Diagnosis of Down syndrome**

*Sheets, K. B., Crissman, B. G., Feist, C. D., Sell, S. L., Johnson, L. R., Donahue, K. C., ... & Brasington, C. K. (2011). Practice guidelines for communicating a prenatal or postnatal diagnosis of Down syndrome: recommendations of the national society of genetic counselors. Journal of genetic counseling, 20(5), 432-441.*

NSGC Practice Guidelines for Communicating a Diagnosis of Down syndrome. This guideline serves to 1) summarize the various etiologies of Down syndrome and the associated recurrence risks; 2) review the key components of disclosing a diagnosis of Down syndrome in both the prenatal and the postnatal settings; and 3) provide references for both professional and patient resources.

1. **AAIDD: The Down Syndrome Information Act: Balancing the Advances of Prenatal Testing Through Public Policy**
*Leach, M. W. (2016). The Down Syndrome Information Act: Balancing the advances of prenatal testing through public policy. Intellectual and developmental disabilities, 54(2), 84-93.*
Summary of Down Syndrome Information Acts published in a range of states and the implementation in state departments of public health.
2. **Receiving the Initial Down Syndrome Diagnosis: A Comparison of Prenatal and Postnatal Parent Group Experiences**

*Nelson Goff, B. S., Springer, N., Foote, L. C., Frantz, C., Peak, M., Tracy, C., ... & Cross, K. A. (2013). Receiving the initial Down syndrome diagnosis: a comparison of prenatal and postnatal parent group experiences. Mental Retardation, 51(6), 446-457.*
This study explored the preliminary experiences of parents upon learning of their child’s diagnosis of Down syndrome. Qualitative data from a web-based, national survey were analyzed based on two groups: prenatal (n 5 46) or postnatal (n 5 115) diagnosis. Three primary categories emerged from the data analysis: prenatal screening/testing decisions by parents, the adjustment process for parents, and postdiagnosis resources and support for parents. Participants’ rationale behind pursuing testing ranged from wanting to be better prepared to not pursuing testing because it was not a factor in continuing the pregnancy. Participant reactions to the diagnosis involved a range of intense preliminary emotions; participants described their extreme grief and loss experience at the initial news of the diagnosis, which also was ambiguous in nature and required differing timelines of adjustment. Finally, participants described experiences with medical professionals, information/ education, and faith/religion as resources and areas of support, although not all were described as positive in nature. Participants in both groups identified having negative experiences with medical professionals during the diagnosis process. The results indicated the importance of these early experiences for parents of children with Down syndrome and emphasize providing effective education, resources, and practical information from reliable sources. N=46 prenatal (40 White,; 6 Other) and 115 postnatal
3. **Adherence of cell-free DNA noninvasive prenatal screens to ACMG recommendations**
*Skotko, B. G., Allyse, M. A., Bajaj, K., Best, R. G., Klugman, S., Leach, M., ... & Gregg, A. R. (2019). Adherence of cell-free DNA noninvasive prenatal screens to ACMG recommendations. Genetics in Medicine, 21(10), 2285-2292. Chicago*

"Noninvasive prenatal screening (NIPS) for fetal aneuploidy via cell-free DNA has been commercially available in the United States since 2011. In 2016, the American College of Medical Genetics and Genomics (ACMG) issued a position statement with specific recommendations for testing laboratories. We sought to evaluate adherence to these recommendations."

1. **Having a Son or Daughter With Down Syndrome: Perspectives From Mothers and Fathers**
*Skotko, B. G., Levine, S. P., & Goldstein, R. (2011). Having a son or daughter with Down syndrome: Perspectives from mothers and fathers. American Journal of Medical Genetics Part A, 155(10), 2335-2347.*
Survey of parents of children with Down syndrome to determine their attitudes about their quality of life. The study found that: “The overwhelming majority of parents who have children with DS report that they love their son or daughter and are proud of them; The overwhelming majority of parents who have children with DS report that their outlook on life is more positive because of their son or daughter with DS; Parents who have children with DS mention that while there are struggles and challenges, their children with DS bring them much joy and many rewards. They cite life lessons in acceptance, patience, and purpose; The overwhelming majority of parents who have children with DS say that their other children have good relationships with their brothers and sisters with DS; The majority of parents who have children with DS report that their other children are more caring and sensitive, as a result; A very small percentage of parents who have children with DS say that they are embarrassed by their son or daughter or even regret having them altogether. The majority of these parents had children with significant medical and learning challenges; Slightly more than half of parents who have children with DS say that they have found non-profit DS organizations helpful.” N=2,044
2. **Self-Perceptions From People With Down Syndrome**

*Skotko, B. G., Levine, S. P., & Goldstein, R. (2011). Self‐perceptions from people with Down syndrome. American Journal of Medical Genetics Part A, 155(10), 2360-2369.*
A summary of the results of a survey of individuals with Down syndrome about their quality of life. The study determined that: “The overwhelming majority of people with DS are happy with their lives; The overwhelming majority of people with DS like who they are and how they look; The overwhelming majority of people with DS love their families, including their brothers and sisters; The majority of people with DS feel they can easily make friends; The majority of people with DS feel that they help other people; Only a small percentage of people with DS feel sad about their lives, which appears to be associated, in part, to transition points of adolescence." N=284
3. **Balanced information about Down syndrome: what is essential?**

*Sheets, K. B., Best, R. G., Brasington, C. K., & Will, M. C. (2011). Balanced information about Down syndrome: what is essential?. American Journal of Medical Genetics Part A, 155(6), 1246-1257.*
This is an article about survey that compares what information prenatal and postnatal genetic counselors deem as more important as compared to new and expectant parents.
4. **Disability training in the genetic counseling curricula: Bridging the gap between genetic counselors and the disability community**

*Sanborn, E., & Patterson, A. R. (2014). Disability training in the genetic counseling curricula: bridging the gap between genetic counselors and the disability community. American Journal of Medical Genetics Part A, 164(8), 1909-1915.*

Peer-reviewed article in the American Journal of Medical Genetics about the disability training in genetic counseling curricula. This article pulls old data about genetic counseling student attitudes and experiences, and more recent research suggests that genetic counselors have become more attuned to providing information about psychosocial outcomes and supports and services at the moments of diagnosis. Moreover, the article does not address the gaps in training for obstetricians, maternal-fetal medicine specialists, general practitioners, etc.
5. **When Bad News Isn’t Necessarily Bad: Recognizing Provider Bias When Sharing Unexpected News**

*Carroll, C., Carroll, C., Goloff, N., & Pitt, M. B. (2018). When bad news isn’t necessarily bad: recognizing provider bias when sharing unexpected news. Pediatrics, 142(1).*

Article in Pediatrics that reviews patient experiences when receiving. diagnosis of Down syndrome, some of the implicit and unconscious biases of medical professionals, and strategies for addressing bias.
6. May, C. P., Dein, A., & Ford, J. (2020). New insights into the formation and duration of flashbulb memories: Evidence from medical diagnosis memories. *Applied Cognitive Psychology*, *34*(5), 1154-1165.