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COMMENTARY

Commentary: "My Identical Twin Sequenced Our Genome"

Sabrina A. Suckiel[1,2] · **Randi E. Zinberg**[1,3]

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In this issue of the Journal of Genetic Counseling, Sam and Arielle, monozygotic twin sisters, describe their personal experience with genomic testing. Sam's interest in genetics and her concern about a family history of cancer compelled her to pursue predisposition whole genome sequencing (WGS) testing. This particular WGS test is designed for individuals who are not seeking a specific genetic diagnosis in the context of a medical evaluation, but who are interested in predispositional genetic health information and other genetic information such as pharmacogenetics and carrier status. Arielle was not interested in having testing and was initially not supportive of Sam's pursuit. In sharing their story, the authors call attention to the ethical dilemma raised by genetic/genomic test results that may impact both the patient and family members. The authors also highlight other potential concerns associated with this type of WGS testing, such as the accessibility of genetic counselors and genetic data security/privacy.

In the article, Sam states that her physician ordered the WGS test, but did not provide access to genetic counseling. Her physician had never ordered this type of testing before, though Sam states that she was provided a "rigorous informed consent discussion". It is unclear whether the physician asked Sam about her family history, addressed the potential implications of the results on her family members, or discussed any issues beyond what was included on the testing company's informed consent document. As a result, Sam had access to an abundance of her genetic information but no concrete guidance on what to do with it, how to interpret it, or how to share the information with her twin sister, all of which perhaps lead to unnecessary anxiety and stress. Had Sam sought genetic counseling her choice to proceed with testing may not have been impacted, nor is that the point of genetic counseling. However, her experience may have been very different. A genetic counselor likely would have discussed the type and scope of genetic/genomic results, including the potential to receive very little health altering information, and the impact findings could have on Sam. Providing this anticipatory guidance may have helped Sam manage her expectations with regard to the results. Additionally, since genetic/genomic findings always have implications for family members, a genetic counselor would have explored the potential impact of Sam's test results on her family, including importantly Arielle. Sam and Arielle describe the tensions that Sam's decision to pursue testing raised between the sisters. Assistance, including the offer of a meeting between the genetic counselor and both Sam and Arielle, might have helped facilitate the process. Regardless of the impact on family members, including Arielle, Sam has decisional capacity and can therefore make her own autonomous medical decisions. Ultimately, in this case, the sisters chose to work out a compromise privately, which was in fact an appropriate solution.

The article highlights a number of concerns that Arielle had about predisposition WGS testing, including concerns about finding a pathogenic variant, the potential psychological impact of the results, and genetic data security/privacy. The concerns emphasized here are similar to those identified in research of individuals considering undergoing the same type of predisposition WGS testing (Robinson et al. 2016; Sanderson et al. 2016). It is important that

✉ Randi E. Zinberg
randi.zinberg@mssm.edu

[1] Department of Genetics and Genomic Sciences, Icahn School of Medicine at Mount Sinai, One Gustave L. Levy Place, New York, NY 10029, USA

[2] Icahn Institute of Genomics and Multiscale Biology, Icahn School of Medicine at Mount Sinai, New York, NY, USA

[3] Department of Obstetrics, Gynecology and Reproductive Science, Icahn School of Medicine at Mount Sinai, New York, NY, USA

genetic, or in this case genomic, counseling explore such concerns in order to help patients make informed decisions about testing as well as to help patients emotionally prepare for the range of potential results.

As we move further into the era of genomic medicine, access to genomic testing is increasing exponentially. There are currently a number of companies offering predisposition genome sequencing testing for healthy individuals (Baylor Miraca Genetics Laboratory, 2016; www.bmgl.edu, www.illumina.com, Veritas Genetics, 2016; www.veritasgenetics.com) and more will likely emerge in the near future, especially given the steadily decreasing costs of such tests. Research studies have confirmed public interest in this type of testing (Biesecker et al. 2009; Hood and Price 2014; Sanderson et al. 2016; Vassy et al. 2014). The Centers for Disease Control and Prevention has noted that many genetic/genomic tests are being marketed prematurely to the public through mass media, and this article demonstrates that individuals can and will ask their physicians to order this type of test. It is safe to say that predisposition genome sequencing testing is not going away. The breadth and depth of potential information generated from this type testing is immense and brings with it specific counseling challenges, such as conveying this information in a clear and concise manner and assisting patients in making decisions about a wide range of potential results as well as coping with any uncertainty the results may bring. Genetic counselors should be providing genomic counseling services to patients undergoing predisposition genome sequencing ideally prior to such testing. However, when a referral to a local genetic counselor is not an option, physicians should make use of the growing genetic counseling services provided by the testing companies. In either case, it is becoming increasingly clear that patients, as well as physicians, are in need of more guidance with regards to predisposition genome sequencing of healthy adults.

Compliance with Ethical Standards

Conflict of Interest Sabrina Suckiel and Randi Zinberg declare that they have no conflict of interest.

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