To Want to Know is Human
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The doctor rubbed a gel over the abdomen of her young patient. Pointing at the grainy sonogram, she animatedly indicated the various structures of the growing fetus.

"Those are the ten little piggy toes, those are the feet, and-" the physician paused. After peering more closely at the monitor, she spoke to the couple. "Your baby has a thickened nuchal fold-- the back of the neck area. This is a marker that geneticists screen for in ultrasound exams of women in their second trimester. It is a potential indicator for Down syndrome (Kataguiri, Júnior, Bussamra, Nardozza, & Moron, 2014)."

The parents stared at the physician in fear. She kindly smiled at them and said, "I will discuss with you the benefits and risks of having an amniocentesis to diagnose for Down syndrome, and then you may decide whether to schedule one-- after which, I would discuss the results with you."

The wife responded, "Oh no! I have read that that's invasive and could possibly harm the baby. My husband and I underwent in-vitro fertilization to conceive it. What are my other options?"

The physician said, "Well, there's a recently developed, noninvasive alternative called the Non-Invasive Prenatal Screening test-- NIPS for short. It has a very high detection rate, but it is not diagnostic and has a chance for false-positive test results (Strom et al., 2017). After reading the results, we could discuss your next possible options."

Down syndrome is the most common chromosomal disorder (Division of Birth Defects and Developmental Disabilities, 2018). Because bringing a child with a disability into a family heavily impacts family dynamics, and the opportunity for various reproductive decisions exists, knowledge of an unborn child's risk for Down syndrome is important (Kazemi, Salehi, & Kheirollahi, 2016). Consequently, the option of prenatal testing for Down syndrome has become routinely offered to expecting couples during pregnancy. However, without the expertise of certified genetic counselors to guide them through this step-wise process and values-based decision making, they could not make *informed* decisions on knowing the disease risk of their child and determining the course of a pregnancy involving a living fetus. A unique back-and-forth about the invasiveness, risk, and predictive value of tests informs the consent of the patient to genetic testing and medical intervention, as well as the provider of the patient's needs.

Contrary to this physician-patient dialogue, there has been a marked increase in the availability of direct-to-consumer genetic testing (DTCGT), via which consumers can independently gain insight into their genome. Companies currently offer DTCGT that can discover many more genetic variants and disorders than what is recommended by major health organizations like the American College of Medical Genetics and Genomics (Phillips, 2016; American College of Medical Genetics and Genomics, 2015). While the benefits of this is obvious-- more information about one's ancestry and oneself, earlier diagnoses, earlier prevention and treatment-- lack of consent to testing that is definitively informed by a

genetic counselor actually lowers the autonomy of the patient that is perceived to increase (Hogarth & Sakko, 2017). Undergoing testing with an insufficient understanding of the implications of potential test results can lead to unwanted knowledge and decision-making (Middleton, Mendes, Benjamin, & Howard, 2017). Further, without a genetic counselor contextualizing obtained results within patients' lifestyle and environmental influences on disease risk nor validating the accuracy of results, unnecessary or inappropriate medical intervention can result. Thus, in instances in which the person undergoing the test will *manifest* the disorder, both pre- and post-testing counseling should ideally be available.

In contrast, pre-test counseling is not necessary in instances in which the person undergoing the test or a living person impacted by the results will *not* manifest the disorder(s) being tested, as the aforementioned emotional consequences will not result. For example, in a type of DCTGT called carrier screening, a couple planning to become pregnant is screened for its carrier status for many genetically inherited diseases (U.S. National Library of Medicine, 2018). If the couple together tests positive as carriers for a recessive disorder that could affect a potential fetus, the couple should consider post-test genetic counseling.

There is a shortage in genetic counselors, so practically speaking, it is difficult to offer genetic counseling pre- and post-test for all individuals undergoing genetic testing (Johnson, Farrell, & Parens, 2017). To maximize patient autonomy and promote the most appropriate and healthy medical intervention-- for expecting couples, for individuals, for planning couples-- genetic counselors should be required at least post- test.

References

American College of Medical Genetics and Genomics. (2015). *Five things patients and providers should question* [Fact sheet]. Retrieved March 6, 2018, from Choosing Wisely website: http://www.choosingwisely.org/wp-content/uploads/2015/07/ACMG-Choosing-Wisely-List.pdf

Division of Birth Defects and Developmental Disabilities. (2018, February 15). *Down syndrome* [Fact sheet]. Retrieved March 6, 2018, from Centers for Disease Control and Prevention website: https://www.cdc.gov/ncbddd/birthdefects/downsyndrome.html

Hogarth, S., & Sakko, P. (2017). A market in the making: The past, present and future of direct-to-consumer genomics. *New Genetics and Society*, *36*, 197-208. https://doi.org/10.1080/14636778.2017.1354692

Johnson, J., Farrell, R. M., & Parens, E. (2017). Supporting women's autonomy in prenatal testing. *The New England Journal of Medicine*, *377*(6), 505-507. https://doi.org/10.1056/NEJMp1703425.

Kataguiri, M. R., Júnior, E. A., Bussamra, L. C. S., Nardozza, L. M. M., & Moron, A. F. (2014). Influence of second-trimester ultrasound markers for Down syndrome in pregnant women of advanced maternal age. *Journal of Pregnancy*. https://doi.org/10.1155/2014/785730

Kazemi, M., Salehi, M., & Kheirollahi, M. (2016). Down syndrome: Current status, challenges and future perspectives. *International Journal of Molecular Medicine*, *5*(3), 125-133. Retrieved from PubMed database.

Middleton, A., Mendes, A., Benjamin, C. M., & Howard, H. C. (2017). Direct-to-consumer genetic testing: Where and how does genetic counseling fit? *Personalized Medicine*, *14*(3), 249-257. https://doi.org/10.2217/pme-2017-0001

Phillips, A. M. (2016). 'Only a click away — DTC genetics for ancestry, health, love…and more: A view of the business and regulatory landscape. *Applied & Translational Genomics*, *8*, 16-22. https://doi.org/10.1016/j.atg.2016.01.001

Strom, C. M., Anderson, B., Tsao, D., Zhang, K., Liu, Y., Livingston, K., . . . Rowland, C. (2017). Improving the positive predictive value of non-invasive prenatal screening (NIPS). *PLOS One*, *12*(3). https://doi.org/10.1371/journal.pone.0167130

U.S. National Library of Medicine. (2018). *Genetic testing* [Pamphlet]. Retrieved from Genetics Home Reference database.