Frequently Asked Questions about Genetic Testing

A discussion with genetic counselors from Yeshiva University's **Program for Jewish Genetic Health**

Dr. Nicole Schreiber-Agus, scientific director and program liaison for Yeshiva University's *Program for Jewish Genetic Health* (PJGH), sits down with the PJGH's two genetic counselors, Estie Rose and Chani Wiesman Berliant, following spring 2012 genetic screening and educational programs at both undergraduate campuses of YU.[1]

Nicole Schreiber-Agus (NSA): *Tell me a little bit about your dual roles in genetic counseling and with the PJGH.*

Estie Rose (ER): Well to start, Chani and I both are board certified genetic counselors. At our offices, we meet with people who are identified as being at risk for genetic diseases or for having children with these diseases. We take detailed family medical histories for each of our patients and make recommendations for genetic testing based on these histories. Our involvement with the PJGH allows us to apply our expertise specifically to the Jewish community and engage in how issues of genetics interplay with Jewish law, tradition, and values.

Chani Wiesman Berliant (CWB): Back in March, the PJGH, in coordination with the student-led YU Medical Ethics Society of Yeshiva University's Center for the Jewish Future, ran two events, one on each undergraduate campus, to educate students about the medical and halachic importance of genetic carrier screening for diseases that are common in the Ashkenazi Jewish

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population, like Tay-Sachs disease. After these, we ran subsidized genetic screening events in April on each undergraduate campus. We screened about 120 students, and 30 percent of our screenees were identified to be carriers. This is just about what we'd expect, as one in three Ashkenazi Jews are found to be carriers for at least one of the Ashkenazi Jewish genetic diseases that we screen for.

NSA: *You mentioned the term "Ashkenazi Jewish genetic diseases." Can you explain what they are and why carrier screening for them is important?*

ER: Ashkenazi Jewish (AJ) genetic diseases, for the most part, are severe diseases that can be passed down from unsuspecting healthy parents to their children. These conditions can technically affect anybody, but we called them AJ diseases because they are more commonly found in the AJ population, and individuals of AJ descent carry specific and well-defined *founder mutations*. Carriers will never develop the disease in question, however being a carrier is very significant for a couple in which both members are carriers of the same genetic condition. In that case, there is a one in four chance in each pregnancy for them to have an affected child. This type of inheritance is called "recessive inheritance." Statistically, it has been found that one in 100 couples will be carriers of the same AJ genetic disease, or "carrier couples." Genetic screening for Sephardic Jews is not as straightforward because unlike the more uniform Ashkenazi Jewish gene pool, the Sephardic community is more diverse and there is no well-defined list of founder mutations. We recommend that Sephardic Jews have a genetics consultation, since the Sephardic diseases that need to be addressed depend on one's country of origin.

CWB: The AJ disease screening process itself is fairly simple. The genetic testing is done on a blood sample. Currently we are testing for 18 AJ genetic diseases, and we are looking for the specific AJ founder mutations. We also offer testing for spinal muscular atrophy (SMA), an inherited disease that does not have a specific AJ mutation, but has a mutation that is common in all populations. If the mutations are not present, then that individual is most likely not a carrier. However, there is always that slight possibility that one could still be a carrier of a less common mutation that we did not look for.

NSA: *If someone doesn't have a family history of these conditions, do they still need to be screened?*

CWB: Absolutely! Because carriers of these genetic conditions will never develop any signs or symptoms of the condition in question, the only ways to know if you are a carrier is either to find out through this genetic blood test, or to be identified as a carrier after having an affected child. Of course, we want to avoid having individuals finding out the hard way, and so we encourage people to get screened *before* they start a family.

ER: If you were to meet a family with a child affected with any of these diseases, you would understand how important it is to take these precautionary measures. Carrier screening is crucial because it provides couples with the knowledge and options to help build healthy families. In fact, many rabbis have included genetic carrier screening as part of the same obligation to protect one's health implied from the words in Deuteronomy 4:9 "רק השמר לך ושמר נפשך מאד, *However, be careful and guard yourselves very well.*" and Deuteronomy 4:15 " ונשמתרם מאד לנפשתיכם, *You shall be very careful of yourselves.*"

NSA: *What do you tell people who are found to be carriers?*

CWB: When our patients are identified to be carriers, we remind them not to panic. Remember that being a carrier does not change anything you've always known or thought about yourself. In fact, we are all carriers of somewhere between five and 10 recessive genetic conditions. We encourage our patients to tell their partners so that they could be tested for that condition as well. We also remind our patients that if they are a carrier, it means at least one of their parents must be a carrier, and so too, each of their siblings has a 50 percent chance to be carriers. Aunts, uncles and cousins may be carriers as well, and it is important to share this information with other family members so that they can be tested and potentially learn valuable information which may have important implications for family planning in the future.

NSA: *If one in 100 couples are "carrier couples," wouldn't we expect to see more families having children with these diseases?*

CWB: That's a good question. We don't see so many families having children with these diseases because there are ways for "carrier couples" to avoid having an affected child. Some carrier couples will decide to break up a relationship and, in some communities, being carriers for the same genetic disease may disqualify the couple even before they meet. For couples who choose to stay together, there are other reproductive options. Remember that there is still a 75 percent chance in each pregnancy for those couples to have a healthy child. That being said, 25 percent is quite a high risk to have an affected child, and we work with carriers to help them decide how they want to go about building a healthy family. Some couples may decide not to have children, others may consider adoption, and others may utilize gamete donors.

ER: It's a common misconception that couples in which both members are carriers of the same genetic disease can't have healthy children and shouldn't pursue a relationship together. Some couples may decide to achieve a pregnancy naturally and "take a chance." Those couples can opt to test the fetus for the disease through procedures such as a chorionic villus sampling (CVS) or amniocentesis. Both the CVS and amniocentesis do have a small risk of miscarriage, however the end result of this testing would give a couple the information of whether or not their child is affected. Some couples may choose to continue a pregnancy knowing that their child is affected, while other couples may seek rabbinic guidance to help them decide whether or not to terminate the pregnancy. Our job is to present the options so that the families we work with know what is out there.

CWB: Another option available to carrier couples to achieve a healthy pregnancy is through in vitro fertilization (IVF). Prior to implanting the fertilized embryos in the woman's uterus, the embryos are tested for the specific genetic disease through a process called preimplantation genetic diagnosis (PGD). At the end of this process, only the embryos that are unaffected are implanted back into the woman's uterus. While this is indeed a great option for carrier couples, it is important to recognize that IVF with PGD is a very expensive and challenging process. It may not be covered by insurance, could add quite a bit of stress onto a couple's relationship, and may require multiple cycles before achieving a successful pregnancy and the birth of a healthy child.

ER: As you can see, there are a lot of options out there, but each couple is going to have their own feelings about these. Couples who are trying to determine what the best route is for them might find it helpful to talk with rabbis, therapists, genetic counselors, friends, family members and even couples who have already had an affected child.

NSA: *How do you counsel singles who have been identified as carriers? Must they disclose this information when they go on dates?*

CWB: There is a point in each relationship where couples should discuss genetic testing, and that point will be different for each couple. Some use it as a pre-screening process, while others will discuss their carrier status later in the dating process. I tell my patients that it is up to them to decide when to discuss their genetic carrier status, but it is definitely something that needs to be discussed. Follow-up testing for the other partner, once determined, will help us clarify the risk for a couple to have an affected child, and the earlier that couples have this information, the better. One's carrier status for these types of diseases does not reflect who somebody is as a person or how suitable he or she is as a spouse. This is information which couples must be able to discuss in an open and honest way.

NSA: *If someone meets with you and gets tested and the results are negative, can we safely say that they are "in the clear"?*

CWB: Unfortunately not! When we do this AJ genetic testing, we are testing for a panel of genetic diseases that are more common in Ashkenazi Jews. However there are many other genetic diseases out there that we are not testing for. Some are not necessarily due to AJ founder mutations, but could still affect AJ individuals, while others could occur because of spontaneous changes in the fetus. People don't like hearing this, but every couple has about a 3 percent risk of having a baby with a birth defect or mental retardation. Unfortunately, we cannot test for every disease or condition out there, and it is impossible for us to ever "guarantee" a healthy child.

ER: When it comes to the AJ screening in particular, I want to emphasize that if you are tested now, it does not mean that you are "in the clear" forever. New diseases are constantly being added to the panel, and so it is important to update your AJ screening before each pregnancy. Our hope is that through screening, we can identify couples who are at risk to have an affected child and give them the best chance to have a healthy child. The mutations, however, will never disappear; they will always be passed down, and that's why testing needs to be done in every generation.

CWB: Our hope is that the more knowledge we can provide, the better equipped our communities will be to take ownership over their own health and take a more proactive role in building healthy families.

*The **Program for Jewish Genetic Health**'s philosophy revolves around servicing the Jewish community and listening to its needs, with the ultimate goal of protecting its health and the health of its future generations. www.yu.edu/geneticheath*

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1 IN 3 ARE CARRIERS.

The Program for Jewish Genetic Health of Yeshiva University offers *accessible* and *affordable* carrier screening for genetic diseases that are common in Ashkenazi Jews.

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