



Baby Blueprints

Acupuncture and genetic testing
before
you get pregnant
plus modern options with IVF

by Karen Reynolds,
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CHAPTER

6

GENETIC
COUNSELING—WHY?

GENETIC COUNSELING—WHY?

A genetics counselor is highly trained to not only educate you on what tests to consider but also to educate you about the reliability of any tests you pursue. It is good to remember that there are limitations for all tests—few of them are regarded as absolutely infallible. There can be false positives as well as false negatives for many tests, and an experienced counselor can review all of those specifics with you. If you know the limitations of the tests, you'll be better prepared to review and consider the results as well as think about further testing if necessary. You will also learn about the panels recommended to be most useful to you based upon your unique family history. It may also surprise you to know that genetics counseling is frequently covered under many insurance policies.

Here is one example of testing reliability, which has to do with cystic fibrosis (CF) and the CTFR gene, which causes the disease. More than 1,000 different types of mutations have been discovered on CTFR gene, all of which produce CF. However, some of the less common types do not scar the pancreas as much as others do. Because the testing would be cost prohibitive to check for 1,000+ variants of the CTFR gene, carrier tests are typically checking for the 23 most common CTFR genetic mutations.

This brings us back to an example of test reliability. Usual CF testing will pick up about 90% of carriers. This means that 10% of the time it might be presumed that parents are not carriers but in fact are. In this situation, there is a possibility of a child with CF being born to a couple despite negative test results.

In addition to expertise in determining which types of genetics tests are needed, genetic counselors also will investigate your family history with you. At first glance, this may seem like an easy or straight forward task. However, there commonly are risk factors or indicators that the average person may not note when reviewing their family health history. You can actually begin gathering information on your family history now and below are some on-line tools.

Charles Eng, Director of the Cleveland Clinic's Genomic Medicine Institute is quoted here: "Family history is a major tool for gauging genetic risk of disease. It's a wonderful, inexpensive quick snapshot of genes and environment. Family history is the sum total of what you inherit and what you are exposed to. Once you identify someone at high risk, you can proceed to the next stage of evaluation by genetics professionals, and testing can begin if a gene is known."²¹

Tips on how to begin your family history

- List diseases your blood relatives have had (including parents, siblings, half-siblings, aunts, uncles and cousins)
- How old were these individuals at the time a disease started?
- For deceased family members, what was the cause of death?
- How old were individuals at the time of death?
- Did the deceased relatives have risk factors such as smoking, obesity, etc.?

Do you want to get started on a family history right now? If so, you can immediately begin to collect and document information for your own family history using these two online resources:

- The United States surgeon general offers a free online tool called My Family Health Portrait. Please see <http://www.hhs.gov/familyhistory/portrait/> This tool allows you create a document that is organized for easily presenting information to your MD. It also allows for updating information over time, sharing it with family members or printing it out. It is available in both Spanish and English.
- The March of Dimes family history tool is available at: <http://www.marchofdimes.org/materials/family-health-history-form.pdf>

Within my acupuncture practice I often treat stress and anxiety reduction for clients who are considering genetic tests while looking at risk statistics. Sometimes numbers give comfort, other times not so much. It is key to remember that when you weigh facts, look at the big picture and when doing so, look at it with people who are educated in the field of genetic testing. This will give you a more informed space versus a focus on any single percentage. While part of my role is to calm the spirit, genetics counselors are trained to conduct a thorough family history, recommend tests, be supports for the seriousness of decision-making, and ultimately help you as the patient navigate complex topics.

Genetic counselor Amy Vance was kind enough to meet with me and share information about her specialty as well as her

substantial clinical background and experience. Please also see her website at: <http://www.bayareagc.com/BAGC/home.html>.

Karen Reynolds, LAc, MS, RN

Please tell me about your background.

Amy Vance, MS, LCGC

I have a BA in zoology and completed the pre-med curriculum from Miami University. I have a master's in genetic counseling and graduated in 1991. I am also board certified by the American Board of Genetic Counseling and licensed by the State of California. Licensure for genetic counselors is relatively new and is not available in all states yet.

Karen Reynolds, LAc, MS, RN

Can you elaborate on the benefits of consulting with a genetic counselor?

Amy Vance, MS, LCGC

The major benefits are to gather information about family history and assess any possible risk to pregnancy. These are discussed, and testing can be offered *before* pregnancy because it opens a lot of options. If both individuals carry, for example, a recessive gene for the same condition, they can consider embryo screening if the couple is doing IVF. It offers people an option to avoid finding out stressful information during pregnancy, when there are fewer options.

Karen Reynolds, LAc, MS, RN

What are some pros or cons about screening prior to pregnancy?

Amy Vance, MS, LCGC

Even basic carrier screening during pregnancy is hard, because waiting for any type of test result is very stressful. If it

is an option, it is better to do the testing when not pregnant because it is less waiting, wondering and worrying. It gives more of a footing for decision- making.

Karen Reynolds, LAc, MS, RN

For women who are trying to conceive, what does genetic counseling entail and what are the most typical genetic tests offered?

Amy Vance, MS, LCGC

First, a Family Health Risk Assessment is made for three generations of family history. It is then discussed and analyzed. For most people, this screening tool in and of itself clarifies individual medical issues.

In addition to the risk assessment, genetic screening for Society recommended diseases can be offered. The American Congress of Obstetricians and Gynecologists (ACOG), the American Society for Reproductive Medicine (ASRM), and the American College of Medical Genetics (ACMG) have certain guidelines for carrier screening.

I don't do larger (non-society based) genetic testing panels. The large extended screening panels typically screen for over 50-100 variants, which increases the number of people who test positive significantly. Additionally, typically these panels don't screen for all mutations for each condition. So if you are "negative," you are only "negative" for certain mutations on the panel. I advocate offering a test in the context of genetic counseling and appropriate follow up, providing knowledge of testing limitations, and giving support if a test is positive.

People can get incorrect information or misinformation in the absence of proper support and that can do potential harm.

Karen Reynolds, LAc, MS, RN

What are the inadequacies you see in current pregnancy related genetic testing?

Amy Vance, MS, LCGC

In the prenatal setting, anyone over 35 at the time of delivery in the state of California is referred to genetic counselor. However, most OBs do not refer women under the age of 35 years for preconception genetic counseling.

It is also standard that most OB's do not see patient until week 8 of pregnancy, which is because there is a high miscarriage risk in the first 8 weeks.

Another point many consumers do not understand is that most of the time, genetic counseling is covered by insurance, and you can meet with a genetic counseling before becoming pregnant.

Genetic counseling can be thought of as a way to receive information about options during pregnancy, including family history screening, genetic testing and screening including blood testing, prenatal diagnosis, and ultrasound. Genetic testing is optional and the genetic counselor can help you understand what information is available and how it can help inform you during your pregnancy.

Karen Reynolds, LAc, MS, RN

It sounds as though there are needs in the realm of the egg donor population for genetic counseling as well then?

Amy Vance, MS, LCGC

Donors who are asked to consent to genetic testing deserve informed consent about what the testing is and what the test

results mean. They deserve to be provided genetic counseling for positive test results. Many times this does not occur. There are no specific regulations or requirements regarding provision of genetic counseling in this situation.

One idea I had was charging an extra \$50 for genetic counseling for each donor. The fee is nominal and would be pooled and used for counseling donors and educating intended parents. Yes, it would cost everyone a bit more, however, at least there would not be a penalty for the donor with a positive test result.

An organization called SEEDS is currently working with donor agencies, genetic counselors, attorneys to try to achieve guidelines for certain processes related to egg donation, including genetic counseling and testing. ASRM currently does not address these specific issues.

Karen Reynolds, LAc, MS, RN

Thank you, Amy for sharing your time and expertise. For further information, please see Amy's Bay Area Genetic Counseling website at: www.bayareagc.com