Science AMA Series: We are a pair of genetic counselors here to talk about patient support and helping people understand their genetic test results. One of us underwent genetic testing herself. AUA!

Breast$_C ancer_A MA$[1]$and r/Science AMAs$[1]

[1]Affiliation not available

April 17, 2023

Abstract

Hi, redditors! We're genetic counseling experts with the National Society of Genetic Counselors. Genetic counseling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. October is Breast Cancer Awareness Month, but many of us do what we can to raise awareness all year long. As a part of our job, we help people understand: hereditary risk for breast cancer; how those with a higher risk of cancer can understand their testing and treatment options; the role of genetic counselors in healthcare; and how to educate family members about their potential risk of disease. Here's a bit about those of us answering your questions today, one of whom has a personal experience with genetic testing: Caroline Lieber, MS, CGC: I served as Director of the Joan H Marks Graduate Program in Human Genetics at Sarah Lawrence College for 15 years. After 35 years as both an academic clinician and an educator, I wanted to see what genomics was all about from the consumer perspective. The BRCA gene screening test seemed like a no-brainer. I'm not from a high-risk population, and there's no strong history of cancer in my family. I offered up my saliva without giving it much thought. When I got an email a week later asking me to make a genetic counseling appointment, I thought it was a normal part of the process. I made an appointment with a genetic counselor I know personally. After she called me back – a call that must have been terribly difficult for her – nothing seemed normal for a long time. I learned I had an altered BRCA1 gene. Read more about my experience and the emotions is brought up here. As a genetic counselor I always felt I understood the emotional impact genetic test results have on patients. Now that I've experienced it firsthand, I see even more clearly the importance of talking to a genetic counselor about these issues, especially before testing. Ask me anything about my genetic testing experience, cancer genetics and the ever evolving nature of genomic findings. For example, since I received my genetic test results, there has been updated research on my particular alteration. Joy Larsen Haidle, MS, CGC: I am the immediate past president of the National Society of Genetic Counselors and one of NSGC's Cancer Experts. I can discuss hereditary cancer syndromes such as Lynch syndrome and hereditary breast and/or ovarian cancer, as well as my experience as a public policy advocate for genetic testing. In my job, I help patients who have a higher chance of genetic diseases understand genetic testing results, navigate through treatment options, and educate family members about their potential risk of disease. Sometimes this means giving them the hard news that they have an increased risk for cancer. When this happens, as a genetic counselor, I am there to help them understand how their genetic test results might influence their surveillance and treatment options, as well how the result impacts their family members. Sometimes, it also means just listening to their fears and addressing their concerns. Ask me anything about my job as a genetic counselor. We'll be back at 12:30 p.m. EST to answer your questions, ask us anything! Want to know more about how genetics affects your risk for diseases, or how to capture your family health history? Sign up for our free webinar on Nov. 2

the WINNOWER

REDDIT

Science AMA Series: We are a pair of genetic counselors here to talk about patient support and helping people understand their genetic test results. One of us underwent genetic testing herself. AUA!

BREAST_CANCER_AMA R/SCIENCE

Hi, redditors! We're genetic counseling experts with the National Society of Genetic Counselors. Genetic counseling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. October is Breast Cancer Awareness Month, but many of us do what we can to raise awareness all year long. As a part of our job, we help people understand:
hereditary risk for breast cancer;
how those with a higher risk of cancer can understand their testing and treatment options;
the role of genetic counselors in healthcare;
and how to educate family members about their potential risk of disease.
Here's a bit about those of us answering your questions today, one of whom has a personal experience with genetic testing:
Caroline Lieber, MS, CGC: I served as Director of the Joan H Marks Graduate Program in Human Genetics at Sarah Lawrence College for 15 years. After 35 years as both an academic clinician and an educator, I wanted to see what genomics was all about from the consumer perspective. The BRCA gene screening test seemed like a no-brainer. I'm not from a high-risk population, and there's no strong history of cancer in my family. I offered up my saliva without giving it much thought.
When I got an email a week later asking me to make a genetic counseling appointment, I thought it was a normal part of the process. I made an appointment with a genetic counselor I know personally. After she called me back – a call that must have been terribly difficult for her – nothing seemed normal for a long time. I learned I had an altered BRCA1 gene. Read more about my experience and the emotions is brought up here.
As a genetic counselor I always felt I understood the emotional impact genetic test results have on patients. Now that I've experienced it firsthand, I see even more clearly the importance of talking to a genetic counselor about these issues, especially before testing. Ask me anything about my genetic testing experience, cancer genetics and the ever evolving nature of genomic findings. For example, since I received my genetic test results, there has been updated research on my particular alteration.
Joy Larsen Haidle, MS, CGC: I am the immediate past president of the National Society of Genetic Counselors and one of NSGC's Cancer Experts. I can discuss hereditary cancer syndromes such as Lynch syndrome and hereditary breast and/or ovarian cancer, as well as my experience as a public policy advocate for genetic testing.
In my job, I help patients who have a higher chance of genetic diseases understand genetic testing results, navigate through treatment options, and educate family members about their potential risk of disease. Sometimes this means giving them the hard news that they have an increased risk for cancer. When this happens, as a genetic counselor, I am there to help them understand how their genetic test results might influence their surveillance and treatment options, as well how the result impacts their family members. Sometimes, it also means just listening to their fears and addressing their concerns. Ask me anything about my job as a genetic counselor.
We'll be back at 12:30 p.m. EST to answer your questions, ask us anything!
Want to know more about how genetics affects your risk for diseases, or how to capture your family health history? Sign up for our free webinar on Nov. 2

READ REVIEWS

WRITE A REVIEW

CORRESPONDENCE:

DATE RECEIVED:
October 29, 2016

DOI:
10.15200/winn.147765.59052

ARCHIVED:
October 28, 2016

CITATION:
Breast_Cancer_AMA , r/Science , Science AMA Series: We are a pair of genetic counselors here to talk about patient support and helping people understand their genetic test results. One of us underwent genetic testing herself. AUA!, *The Winnower* 3:e147765.59052 , 2016 , DOI: 10.15200/winn.147765.59052

A lot of people see genetics as deterministic, eg "I have this variant, I am going to die of cardiovascular disease," when often these risk alleles have extremely low effect sizes and can be totally overwhelmed by modifying genetic and environmental factors. Admittedly, most clinical testing is done for larger effect variants, but I'm thinking more along the lines of people trying to interpret their direct-to-consumer genetic assessments (ie 23andMe, etc). Can you tell us a little bit about how you handle the uncertaintly when you discuss risk and medical interventions with your patients?

p1percub

Most parts of medicine have an element of uncertainty. Understanding what a test results mean and the nuances tailored from one person to the next in the same family is a key reason why meeting with a genetic counselor can be of value when pursuing genetic testing. Many genetic tests predict the chance of developing a condition over a lifetime such as with cancer genetic testing. A result often suggests that over my lifetime, my chance of developing breast cancer may be higher than a woman in the general population, but it does not give me the diagnosis. It says that I have a risk factor and may benefit from extra surveillance, perhaps starting at a younger age or from considering risk reduction. I may never develop the condition, but what do I wish to do in the meantime to help address concerns? These conversations are very important and should be tailored to the personal and family history. As a genetic counselor, my goal is to help patients understand what their chance is of developing cancer over their lifetime based on family history and/or genetic testing results, discuss options that could be considered to address that risk and help them feel empowered to play an active role in the discussion with their doctor to set the care plan that feels right for them. Not everyone views risk in the same way. Some people see it as 0% or 100% whereas others see risk in increments where it is tolerable to a given point and once it is higher than their personal comfort level, it is time to do something to address it. The personal comfort level can be quite different from one person to the next based on their personal experiences and that of their family members.

Joy

I'm going to be getting a masters in genetic counseling and have two questions.

1) Why is it such a female dominated profession?

2) What is a typical day for an entry level genetic counselor?

Dr_Snarky

Great to hear you are interested in the profession!

The number of men in the profession has grown over the years as has the number of genetic counselors (we now have more than 4000 GCs in the US!). The roles in which genetic counselors can contribute to the profession have also expanded which has helped bring a diverse group of people to the profession.

The typical day can depend on the subspecialty you have chosen and the work setting. Many genetic counselors work in a hospital or clinic setting seeing patients to discuss concerns about their personal or family history and determine if genetic testing may be of benefit and how they might use the information in their medical care. Some genetic counselors provide consultations by telephone; others by tele genetics (combination of telephone and computer) or in a group setting. We have done a lot of work to help make it easier to talk with a genetic counselor regardless of where in the country someone may live.

Some genetic counselors work in research or in a laboratory setting. Others work in public policy or

with insurance companies. The skill set is quite transferable and the kinds of jobs genetic counselors work in now has expanded really quickly. So, the day to day job for a genetic counselor can be very different depending on the work setting.

While you are in a graduate training program, it is useful to have exposure to a wide variety of subspecialties (cancer, pediatrics, prenatal, cardiac, laboratory, etc) to get an idea of where your interests lie and the kind of job that may interest you at graduation.

It is an exciting time for genetic counselors. After being in the field for 21 years, I still find the rapid pace, new advances, and patient interactions a rewarding experience and love my profession.

Good luck to you! Joy

Would you recommend everyone have their genes analyzed for potential risks? If so do you think it should be part of routine medical practice?

Super_Whack

Hi, Caroline Lieber, MS, CGC here. I am a believer in genetic screening, but I think it is important to understand what the screening means before doing it. I learned that lesson myself. As someone who has been in the genetics field for over 35 years, I thought I knew what to expect from the experience, and when my results came back with an unusual finding, I was unprepared for the emotional response. Genetic counselors are the experts, and they walk you through the science as well as what to expect from the results. You can find a genetic counselor in your area at http://www.nsgc.org/page/find-a-gc-search.

I'm a doc student studying precision medicine. Since precision medicine looks to add additional details about patient lifestyle, their environment, and their genetics, I'm concerned about lack of subject matter experts out there (mainly, genetic counselors and clinical geneticists). I know there are slightly over 3,000 genetic counselors as of 2015, and as of 2014, there were only 1,194 clinical geneticist, which accounts for around .18% of the physicians in the US [1]. Given that many physicians are unprepared to explain genetic tests [2] or don't feel comfortable doing so [3, 4], I worry that a spike in demand will over-tax genetic specialists. I know that we are still in the initial planning stages for precision medicine, and Obama's Precision Medicine Initiative aims to construct its cornerstones, but it is a daunting task. I worry that precision medicine will encounter similar adoption problems as EHRs have seen. What are some suggestions you might have to help avoid these pitfalls for precision medicine? Thanks!

1. ABMGG. Training options. 2014. http://www.abmgg.org/pages/training_options.shtml.
2. Guttmacher AE, Porteous ME, McInerney JD. Educating health-care professionals about genetics and genomics. Nat Rev Genet. 2007;8(2):151–7.
3. Nippert I, Harris HJ, Julian-Reynier C, Kristoffersson U, Leo P, Anionwu E, et al. Confidence of primary care physicians in their ability to carry out basic medical genetic tasks—a European survey in five countries—part 1. J Community Genet. 2011;2(1):1–11.
4. Powell KP, Cogswell WA, Christianson CA, Dave G, Verma A, Eubanks S, et al. Primary care physicians' awareness, experience and opinions of direct-to-consumer genetic testing. J Genet Couns. 2012;21(1):113–26.

DiscursiveMind

Wow! Tough, but thoughtful question!

There are currently more than 4100 GC in the US. We have grown by 88% in the last 10 years and are placing concerted efforts to help grow the profession in the next 10 years. You are correct that comfort level and knowledge in genetics can be quite variable from one physician to the next and it takes a

concerted effort to stay current in the field due to the rapid changes and complexity in testing. Increasing the comfort level in genetics across all subspecialties in medicine is important and the Precision Medicine Initiative may help to anchor the utility and emphasis on genetics and genomics in the future of medicine. So, education across the board will help identify people who would benefit from genetic counseling and/or testing and help more patients access care.

We have worked to develop multiple service delivery models to reach more people in ways that suit their needs. Interactive technology may also help to identify, triage, and refer the appropriate people, but may also help consumers learn more too so they can be an active part of their healthcare team. Technology may offer a potential solution to meet the demands too.

Many consumers are looking for information to help them determine what they can do to help protect their health and they would like tools to help them learn more.

Collaboration across medical specialties, consumers, and genetics professionals will help us all to collectively do a better job. It is going to take time to accomplish all of these tasks, but if we are all working towards a common goal, communicating and open to new ideas, we can move forward.

Good luck to you! Joy

I am a social worker and at times we work along side Genetic Counselors to help families process genetic testing results. Giving a client results can be challenging and difficult.

What skills and tools do you think are needed to share difficult news with a client? What interventions do you utilize to support them through the process?

Thanks!

bamajamuh

Hi, Caroline Lieber, MS, CGC here. That is a great question. It sounds like you are familiar with the complexities of genetic testing.
I think there are many tools that are helpful. First and foremost is empathy for their situation. That is, an attitude that allows the clinician to sit with the patient and share his/her emotions without passing any kind of judgment. The patient needs to know that you are there for them, regardless of how they use the information they learn. Once an atmosphere of trust is established, patients can really "walk around" in the information and better understand what it means to them and their families. I am sure you know other techniques that work, such as reflecting, rephrasing, etc. Staying in contact with the patient/family periodically is also useful. Genetic information has different meaning at different stages in life. Marriages, births and deaths all change the family dynamics. Having many healthcare providers available, including a genetic counselor, throughout these stages is very helpful.

I would like to say thank you for what you both do.

My wife and I just went through genetic testing prior to having our child. She went first, and discovered she is a carrier of CPTII. They suggested I get tested as well, as it is incredibly rare, but can be dangerous depending on which type. What dumb luck, I am a carrier too. We both are a carrier of the myopathic form.

That said, my first instinct was to go online and start reading. Terrible idea. Thousands of different things were racing through our minds. Should we try to eliminate this entirely, and go through IVF? 25% seems to be an incredibly high% when deciding on something that may potentially have life altering consequences for our child we were planning on having.

We went to see the genetic counselors at Lurie's children hospital in Chicago, and they were amazing. They put everything in normal terms for us, and allowed us to make a decision on our own knowing the risks both ways. Knowledge is obviously key, but something patients like my wife and I have none of. There is a lot of emotion involved, and people like yourselves do an amazing job. Thank you!

trev086

Caroline Lieber here. Thanks for sharing your story, it is so nice to hear that GCs made a difference for your family. The best to you and your family in the future.

I'm interested in learning more about my own genetics. What services are available to the average person with good health insurance? And are there better commercial ways or services beyond 23andme etc.?

bigandfullballs

Hi, Caroline Lieber, MS, CGC.
This question has no clear easy answer. Insurance companies differ, as do the genetic tests available. Are you interested because of something in your family history, or just generally interested? Family history can be useful in targeting the testing. Your question about health insurance is difficult to answer, because every insurance plan is different. Family history can play a role in whether testing is recommended/covered. I suggest you contact your provider to find out about your specific coverage. A genetic counselor can be helpful in addressing this issue. There are a number of ways in which to obtain genetic screening, from both medical centers and through commercial companies. Generally, the genetics service at a hospital near you can discuss family history concerns, and arrange for appropriate testing. Commercial companies often have genetic counselors available as part of their services, and they can be useful with regard to the specifics of what the particular company offers. There are large companies such as Quest and Labcorp, that offer screening, as well as more specialized companies, such as Counsyl, Ambry, Invitae, to name a few. It is helpful to have your primary physician involved in order to make ordering the tests easier. Ancestry.com has a service to investigate your ancestral background. There are many possibilities.

How strictly is the individual identity and privacy of test patients protected? This genetic data is stored somewhere, so what kinds of precautions are taken for limit access to the types of research affiliate groups that can access an individual's data?

I'm not convinced that my received results is everything that all research groups learned from my sample.

FreshExpress

Meeting with a genetic counselor before pursuing genetic testing can help you address these questions. Testing done on a clinical basis (sent to a CLIA lab, billed to insurance, and returned to your medical record) really isn't much different than other medical tests. The information is protected by HIPAA. States also vary with their privacy laws too. The consent process for genetic testing on a clinical basis should include what happens to extra DNA which can vary from lab to lab, but as the patient, you have input into the process (i.e. destroy or use for research with or without identifiers.)

The consent process for testing on a research basis should also contain information about how your privacy is protected under the IRB approved protocol and who may have access to the information. The research protocol can differ on how (and if) results are made available. Testing on a research basis can take a long time to complete.

Since you have questions about your results, perhaps visiting with a genetic counselor can help to determine the type of information that was meant to be gleaned by the test and address your underlying question about what you feel may be missing from your results that you thought might be available.

findageneticcounselor.com can help you find a genetic counselor near you.

Joy

Hi I'm an undergrad Biomedical science studying Medical genetics. I have a couple questions.

1. What is the career path from an undergrad to a genetic councillor?
2. What is your view on consanguinity and that in theory it can eliminate autosomal recessive disease but the disease frequency itself is not decreasing that much?

tallmasstang

Hi, Caroline Lieber, MS, CGC here. As a former program director of the Joan H Marks Graduate Program in Human Genetics at Sarah Lawrence College, I feel comfortable answering this question! Most genetic counselors have a undergraduate background in the biological sciences, usually biology and/or chemistry and some vertebrate development. Additionally, they have taken some psychology courses as well as some ethics or philosophy. Basic math courses are often required, including statistics. Generally a GPA of "B" is competitive, and most programs require GRE scores. Genetic Counseling training programs want to see volunteerism that includes direct encounters with clients making difficult life decisions, such as HIV/AIDS, birth control, domestic violence.

I am not sure I understand your question regarding consanguinity. Can you clarify it for me?

As a field, how fast is genetic counseling growing? What are some institutions in the country that have exceptional programs for this type of work?

Ingury

The genetic counseling profession has grown by 88% in the last 10 years with significant growth anticipated in the next 10 years! It is an exciting field with many opportunities to contribute across the healthcare spectrum.

The Accreditation Council for Genetic Counseling (ACGC) has a list of genetic counseling programs in the US. http://gceducation.org

Good luck! Joy

I'm currently in my 2nd year of working towards a GC career, and I just wanted to know if there is anything I should know heading into it? What aspect of the training did you find the most difficult, and how did you overcome the challenges that were involved?

I highly respect the line of work you two are in, and hope to be in your shoes sometime in the near future.

CamBamBoomSlam

Hi, Caroline Lieber, MS, CGC here. As a former genetic counseling training program director, I

appreciate this question. There are challenges to be sure, but it is an exciting and fulfilling field to be in. As the genomics world expands, one of the challenges is the amount of information that has to be learned. Secondly, the emotional impact of genetic information is powerful, and at times can feel difficult. No two families react in the same way, so it takes time to get comfortable with how different people respond.
Knowing that you make a difference in people's lives makes it all worthwhile. Good luck to you.

Is genetic counseling something that everyone gets some amount of benefit from? For example, does your work commonly help people find out conditions they may have or may be at risk to have?

Do patients have to come with specific questions such as evaluation for a specific condition or efficacy of specific treaments/drugs? Or Can I get genetic testing, send you the results, and find out some issues (or potential issues) with my dna?

kidneystonejones

Hi!

I often uncover other patterns in the family history beyond the reason why someone was referred. This often leads to good discussions about the patterns noted in a family tree. Sometimes it helps people understand that they may not be at increased risk. Since I specialize in cancer, let me give you an example. Sometimes a family history contains several relatives who have had cancer, but when we consider each person individually in the family tree (looking at their age at diagnosis, risk factors etc) sometimes there isn't a strong pattern and it may be a collection of cancers that appeared by chance or due to shared lifestyle or environmental exposures without suggestion of a strong inherited risk. This can be a valuable conversation to ease risk. Or, if the genetic counselor identifies a pattern, the conversation centers around the chances that you might develop cancer, genetic testing options and how the results could be used in your medical care, and the likelihood of finding an inherited risk factor.

Some people have very specific questions or their doctor had specific questions when they made the referral. It is important to consider what type of information you are seeking to ensure that the test you are considering answers those questions. Direct to consumer testing has a role for some people. Genetic counselors are available to help you understand the results especially if something unexpected is found. findageneticcounselor.com

Joy

As someone with a background in genomics and a PhD in a related field, but no medical or clinical experience, I sometimes wonder about going back to school to get a Master's in genetics counseling. It seems like a field that will only grow in important, and would combine my interests in science, communication, and helping others. For someone who already has a higher degree in a relevant field, would any of the accredited training programs be likely to abbreviate some of the required coursework?

neurobeegirl

Hi, Caroline Lieber here, Let me try to respond as a former program director. With your background and your interests, you would be a valuable asset to the GC community!
While every program is different, I feel comfortable saying that some programs would give credit for courses you have already taken that are required by the GC degree. You would have to investigate the programs individually. While the science is very important, the communication/counseling skills are equally as important. Introductory psychology courses are prerequisites for admission to most GC training programs. Here is a listing of the Accredited Genetic Counseling Training Programs: http://gceducation.org/pages/accredited-programs.aspx