

Hi Reddit! We're experts with the National Society of Genetic Counselors and are here to answer your questions about the sometimes-complicated world of personal genetics.

GeneticCounselors¹and r/ScienceAMAs¹

¹Affiliation not available

April 17, 2023

Abstract

We're genetic counseling experts with the National Society of Genetic Counselors. Genetic counselors receive special training in two areas: genetics and counseling. We use our advanced training to guide and support patients seeking more information about how inherited diseases and conditions might affect them or their families, and to interpret genetic test results. The genetic counseling process integrates: Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence. Education about inheritance, testing, management, prevention, resources and research. Counseling to promote informed choices and adaptation to the risk or condition. Helping patients and families prepare for or navigate at-home genetic test results. NSGC serves as an integral resource for patients, prospective students and healthcare providers interested in learning more about genetic counseling. We're doing this AMA as part of the National Human Genome Research Institute's National DNA Day Reddit AMA series! Ask us anything! Here's a bit about those of us answering your questions today: Erica Ramos, MS, CGC: I am NSGC's President and Personalized Medicine Expert. I can discuss next-generation DNA sequencing technologies such as whole genome and whole exome sequencing, and how these technologies are impacting healthcare and benefiting patients. Amy Sturm, MS, CGC, LGC: I am president-elect of NSGC and NSGC's Cardiovascular Expert. I have more than 14 years of experience helping patients with a higher risk of genetic heart disease understand their familial risk and genetic testing results. I am a nationally recognized expert on familial hypercholesterolemia and can also discuss other hereditary forms of heart disease, including cardiomyopathies, arrhythmias, familial aneurysms and others. Joy Larsen Haidle, MS, CGC: I am a past president of NSGC and an NSGC Cancer Expert. I can discuss hereditary cancer syndromes such as Lynch syndrome and hereditary breast cancer. I am an active public policy advocate for genetic testing. Jason Flanagan, MS, CGC: I am NSGC's Reproductive Health Expert. I can discuss preconception and prenatal genetics, such as how genetics affect infertility and miscarriage, as well as the process and ethics surrounding preimplantation genetic screening. Ana Morales, MS, LGC: I am NSGC's Cardiovascular Genetics and Spanish-Language Expert. I specialize in genetics and heart conditions and I'm a nationally recognized expert on cardiomyopathy, a common condition in which the heart muscle's ability to pump blood is diminished. I can also discuss how I've worked to expand access to genetic information in the Spanish-speaking community. Brianne Kirkpatrick, MS, LCGC: I am NSGC's Ancestry Expert. I can discuss the use of genealogy and DNA testing for exploring family connections and genetic health risks. I can also discuss limitations and benefits of the popular at-home genetic tests. Blair Stevens, MS, CGC: I am NSGC's Prenatal Expert. I have 10 years of experience counseling patients and their families about their risks to have a baby with a genetic condition as well as testing options for conditions such as Down syndrome, cystic fibrosis, sickle cell anemia and spina bifida. I have a passion for helping families who carry a pregnancy diagnosed with a genetic condition or developmental difference. Trish Brown, MS, LCGC: I am NSGC's Policy and Nutrition Expert. I have more than 20 years of experience in clinical genetics and can discuss DNA testing for nutrition and fitness, at-home genetic tests, the study of pharmacogenetics, and policy issues. If you would like more information about genetic counselors and the role we can play in your healthcare, visit our website: aboutgeneticcounselors.com. Updated: Thank you all for participating in today's AMA! We've enjoyed answering your questions. You can find more easy-to-understand genetics information on our website AboutGeneticCounselors.com. If you're interested in genetics and infertility and have more questions on the topic, tune

into a free webinar tonight at 7 p.m. CT. Sign up and see future webinar topics here: <https://goo.gl/ZDFTrM> Thank you, Reddit!

[REDDIT](#)

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GENETIC_COUNSELORS [R/SCIENCE](#)

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Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence.

Education about inheritance, testing, management, prevention, resources and research.

Counseling to promote informed choices and adaptation to the risk or condition.

Helping patients and families prepare for or navigate at-home genetic test results.

NSGC serves as an integral resource for patients, prospective students and healthcare providers interested in learning more about genetic counseling. We're doing this AMA as part of the [National Human Genome Research Institute's National DNA Day](#) Reddit AMA series! Ask us anything!

Here's a bit about those of us answering your questions today:

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Trish Brown, MS, LCGC: I am NSGC's Policy and Nutrition Expert. I have more than 20 years of experience in clinical genetics and can discuss DNA testing for nutrition and fitness, at-home genetic tests, the study of pharmacogenetics, and policy issues.

If you would like more information about genetic counselors and the role we can play in your healthcare, visit our website:

aboutgeneticcounselors.com.

Updated: Thank you all for participating in today's AMA! We've enjoyed answering your questions. You can find more easy-to-understand genetics information on our website AboutGeneticCounselors.com.

If you're interested in genetics and infertility and have more questions on the topic, tune into a free webinar tonight at 7 p.m. CT. Sign up and see future webinar topics here: <https://goo.gl/ZDFTrM>

Thank you, Reddit!

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Hi and welcome!

1. What are your experiences counseling pregnant mothers with profound fetal genetic abnormalities?
2. What are your thoughts on information security of private genetic testing companies like 23andMe?

[PHealthy](#)

Great questions! I will tackle #1. This is Blair Stevens, NSGC's Prenatal expert, and I have counseled countless families who have carried a pregnancy with a severe genetic condition. A genetic counselor's role becomes even more important in these pregnancies as patients are often shocked and saddened by the unexpected news of a severe genetic condition. The first step is to determine whether we are certain of the diagnosis. Genetic counselors can speak with families about the accuracy of genetic testing. We then aim to ensure the patient is well informed about the nature of the condition, understand the long term effects of the condition and what the chances are of it happening again in future pregnancies. We also ensure patients understand their pregnancy management options and refer them to other specialists, when needed. It is incredibly important to support families during these difficult times, explore their needs and values and help them in any way we possibly can. Genetic counselors have access to many resources including written information, support networks, grief counselors and perinatal hospice groups, when patients are in need.

Hi and welcome!

1. What are your experiences counseling pregnant mothers with profound fetal genetic abnormalities?
2. What are your thoughts on information security of private genetic testing companies like 23andMe?

[PHealthy](#)

Hi and thanks for your question! This is Trish Brown, NSGC's experts on Policy, Nutrition, Fitness and DTC testing. I am going to address your second question about information security of private genetic testing companies. It has been my observation and experience that most genetic testing companies have been upfront about how they plan on protecting your genetic data and how they intend to use it, and they are employing industry best practices to de-identify data, so if there is a breach, your personal information is kept separate from your genetic results. However, there are a couple cautionary notes. First, not all companies approach information security the same way, so it is important to learn as much as you can about a company before entrusting them with your data. Ask how long they have been around, how are they funded, and who are the people running the company? Read everything they give you, including the fine print. Although from my perspective most companies are upfront about their data policies, that is because I am looking for those policies. They might not always be easy to find. Sometimes the details of how a company will use and protect your data are buried in the "terms of use" and many of us are in the habit of just agreeing to them, and not really reading them, when creating an online account for the first time. Look for that information. If you can't find anything on data security, privacy, or how your data will be used in the future, ask or go find another company. The

NSGC has a number of genetic counselors that are experienced with the various private genetic testing companies, and they can help people find the right company for their needs. To find a genetic counselor, visit <https://www.nsgc.org/findageneticcounselor>. Thanks for participating today!

Thank you for doing this AMA! Here are my questions:

1. There is a vocal minority of physician-scientists that are critical about the hype around precision medicine (e.g. Dr. Michael Joyner and Dr. Vinay Prasad).

How do you feel about the rapid approval of genomic technology in the common disease setting (e.g. FoundationOne)? Do we currently have enough data or expertise to actually use these well?

2. What's one way the genetic counseling field might change in the next 10 years?
3. Do your experiences support the recent GIM study that showed up to 40% of results from third-party tools on raw DTC data were false positives? Any examples of how this actually played out?

[secret_tacos](#)

Happy to be here! This is Erica, NSGC's President and Personalized Medicine Expert. I also spend what is probably too much time on Twitter (@ERamosSD) so I've seen all of the discussions that you're referring to critiquing the hype around precision medicine. And yes - there is a lot of hype. But it is important to not throw all applications of precision/personalized medicine together. The application of genome and exome sequencing in rare and undiagnosed disease has been a game changer. In my backyard of San Diego, Dr. Steven Kingsmore and his team are using rapid genome sequencing to diagnose the youngest and sickest among us and they recently published on their successes - 43% of babies receiving a diagnosis, many of whom had changes in their care. Population studies are generating large amounts of data that can be used in the interpretations of genetic data and are helping us understand what genomics looks like in a relatively healthy adult population. There are definitely areas that have fallen short of our expectations - which are pretty high - but I disagree with the idea that these applications aren't making a difference.

Genetic counseling is definitely a profession poised to make big changes in the next 10 years and genetic counselors in industry and in patient care roles are innovating already. I expect that we will see at least some degree of population screening for well-understood adult conditions like BRCA1/2, Lynch syndrome and familial high cholesterol syndromes, which already flagged by the CDC as public health concerns. That means that GCs will be integrated into more areas of healthcare, like primary care and internal medicine. Those GCs won't likely be seeing every patient but they'll be building programs to make sure tests are implemented responsibly. There will also be more access to genetic counselors through technologies like video genetic counseling. Overall, I see our role over the next 10 years as leading the way to ensure patients benefit from the responsible implementation of genetic medicine.

The GIM paper that showed a 40% false positive rate was eye-opening but I've seen several genetic counselors report the same things in their patients. It was a small study and it would be great to see more data around confirmation but it reinforces that the raw data from at-home tests are not held to the same standards as if they were performed in a clinical lab, either for the genetic data that is generated or for the interpretation. Any test that is used for clinical decision-making should be performed in a clinical lab. Period. A genetic counselor offers additional value by putting clinical test results in the context of the individual, maximizing the utility of test results. Therefore, it's important to involve a genetic counselor before undergoing or during the process of genetic testing.

Thanks for your questions!

What genetic trait most people believe passes down the family free but actually doesn't?

[bman_78](#)

Hi, this is Erica, NSGC's President and Personalized Medicine Expert.

I was going to say stubbornness but now that we are learning more about the genetic influences of personality traits, I'm not so sure any more! ;) Nature and nurture probably...

What genetic trait most people believe passes down the family free but actually doesn't?

[bman_78](#)

Hello, Ana Morales, here, NSGC cardiovascular and Spanish language expert. Great question! Genetic traits are in our DNA, and DNA is passed down from parents to children. We have 46 chromosomes: 23 come from our mother and 23 come from our father. Within each chromosome, we have genes coding for many body functions and traits. Because we inherit half of our DNA from each parent, we have two copies of each gene. All genetic traits therefore pass down the family tree. The way in which some of these traits are transmitted may give the impression that they may not be passed because they may not appear in every generation. Traits that don't appear in every generation are most commonly known as autosomal recessive traits, in which two copies of a specific gene are needed for a trait to show up. In other words, you need the same gene copy for a particular trait inherited from each parent for the trait to show up.

How do you think we will proceed as a society when selective screening for embryos is so commonplace that we transition from "we don't want to have a child with Tay Sachs" to instead "we don't want a child with autism" or "I just really love blonde hair"?

[ninjakitty117](#)

This is such an important question for us to ask, as a society. I'm Blair Stevens, NSGC's Prenatal Expert. As genetic knowledge and technology advances, it is the role of organizations such as the National Society of Genetic Counselors to promote these types of questions and ensure that the ethical and social aspects of innovation are examined just as closely as the medical and technical components are.

At this time, we are still far from testing for most traits and conditions as we do not yet even understand the genetic component to them. However, with rapid advances in technology, such as CRISPR, we need to be thinking about the ethical implications now. NSGC recently published a position statement on gene editing embryos and it can be found here: <https://www.nsgc.org/p/bl/et/blogaid=826>. NSGC also recognizes and celebrates a person's inherent value including differences in one's physical, cognitive, or psychiatric functioning and we feel policies should be enacted around technologies to ensure safeguards protect the rights of those with these differences. Thank for you asking thought provoking ethical questions!

Hi! I have a Bachelors degree in genetics. I'm not interested in going into medicine, and extremely hesitant on grad school. I live genetics, and I love teaching other people admit how amazing it is. Any suggestions on jobs I could/should look for? Everything I find needs a PhD and 12 years experience.

[ninjakitty117](#)

Hi! This is Erica, NSGC's President and Personalized Medicine Expert.

Well, first I'll make my obligatory pitch for genetic counseling! :) Really though, I may have been in a similar boat. I got my Bachelor's in Cellular and Molecular Biology and was headed to a PhD program. Then I realized that I wasn't that excited about doing research and I didn't want to go to med school either. I got very lucky and got information about genetic counseling programs along with some of the PhD material. After talking to a few counselors, it seemed like a perfect fit - no med school, no rat research and I got to combine genetics with medicine. And now I can't imagine a better profession. GCs are in demand with increasingly growing opportunity. Genetic counselors also report very high job satisfaction and competitive salaries.

But it does require grad school so I'll throw a few other things out there! Many labs and clinics are now hiring genetic counseling assistants. GCAs do not have specific training at this time, but typically have Bachelor's in genetics or related fields and then get on-the-job training. The roles depend on your work setting but generally entails supporting the genetic counselors to prepare for patients, gather literature, etc. Clinical labs also have a lot of opportunities for individuals with Bachelor's and then sometimes additional technical training, if you like being in the lab. If you're a great communicator, some companies will look for people who can translate complicated information into more simple education or marketing materials or will have science writers.

The last thing that I'll say is that there is no way that I would have predicted that I would be working in market development in a big biotech company. So my best advice is to reach out and make connections where you can, learn about the other things that your company/employer does and stretch out of your comfort zone if something seems interesting. One of my favorite quotes that has served me well is from Richard Branson - "If somebody offers you an amazing opportunity but you are not sure you can do it, say yes – then learn how to do it later!"

Good luck!

Genetic chimerism seems to occur much more frequently in humans than what was previously believed, especially in multiple births, but research on its frequency in humans is still lacking ([this Polish study from 1996](#) is the most frequently referenced). Are there any ongoing studies to get a better sense of what the frequency of chimerism occurrence in humans truly is, and have you had these internal discussions on future protocol should you come across such a case?

[ComeyDontPlayDat](#)

Hello, my name is Jay and I am NSGC's reproductive expert. I am not an expert in chimerism, so I do my best to give you information from my experience.

As the article you referenced indicates, the true incidence of chimerism is not well known. In my experience, we have had prenatal testing come back showing two distinct cell lines. Some with 46XY and some with 46XX. When we didn't have appropriate molecular tools, it was hard to figure out if this was due to a male and female embryo fusing to become a single embryo. Over time, with the ability to do more molecular testing, we have determined that most of these are due to a culture of maternal cells and fetal cells. So in these cases, it wasn't due to chimerism.

That said, there are now a number of reports on blood cell chimerism. For example, DOI: [10.1002/pd.1503](#) and PMID: 10783379 discuss how blood chimerism can happen between twins or triplets by blood vessel connections that allowed for blood stem cells to move from one baby to the other. This is likely much more common than previously thought or understood. This could have important consequences for example in pretransfusion testing PMID: 17430077 .

The more uncommon cases are the ones with chimerism that isn't confined to just the blood. For example, this article DOI: [10.1111/j.1749-6632.2008.03570.x](#) discusses individuals with multiple cell types which did lead to other concerns.

With modern molecular testing, it seems that everyday we are coming across things that we previously had not anticipated or planned for. I think that is what makes the world of genetics both very interesting and very humbling. As soon as you think you have a handle on it, things change. That is why I think it is important to have a guide whenever a genetic test is done. The outcomes may not be what a person anticipated when they did the testing. For example, we can run across mosaicism, findings unrelated to the disease we are testing for, or even non-paternity. In these cases, genetic counselors (<http://www.aboutgeneticcounselors.com/>) are trained to walk a family through what this information means and explore emotions, science, and opportunities around them. So, genetic counselors are well versed in the unexpected.

Thanks for asking, it is a very interesting topic and one that we likely will gain a greater appreciation for as more molecular tools are employed.

Jay

How are GCs getting and giving information about polygenic risk scores for complex diseases? What does this look like in a GC session with a patient?

[genomegal](#)

Hi! Amy Sturm, MS, LGC, here, NSGC's President-Elect and Cardiovascular Expert. Polygenic risk scores take information from multiple single nucleotide polymorphisms, or SNPs, and put them into one combined risk score. GCs could receive polygenic risk score information from clinical or research genetic testing. This type of testing may be useful for figuring out which patients may have the highest chances for common diseases like cancer, heart disease, and obesity. Polygenic risk score information could also help us figure out which patients with inherited conditions like familial hypercholesterolemia might be at the highest possible risk. There are multiple ways that GCs could provide this information to their patients. I've provided this type of information to patients in a research study I worked on, and we used both a website portal for patients as well as paper reports that were viewable online or printed. When talking about polygenic risk, I think it's helpful to show patients how their increased risk relates to others without that risk.

@Trish Brown - what do you think about the legitimacy of the nutrition/fitness genetic tests?

[genomegal](#)

Thanks for your question! Trish Brown here, NSGC's expert on Policy, Nutrition, Fitness and DTC testing. I personally believe there is a role for genetic testing in nutrition and fitness, but it is a rapidly evolving area with mixed scientific support. The field is at a very early stage so legitimacy is in the eye of the beholder. Most of the scientific evidence to date is based on small studies in populations that aren't necessarily diverse, and they associate a trait, like obesity, to one or more genetic variants. Often there is no scientific research on what to do about the finding, so recommendations are generic and are often things people already know to do. With obesity testing, for example, you may learn from a test that you are more likely to be overweight and have a reduced ability to know when you are full, so you overeat. You may be recommended strategies on how to eat slowly and chew your food thoroughly before taking another bite. Some people are very excited by this information because it is validating their experience and they find it useful, and for some reason the genetic data makes them take actions that they didn't before. This is why I believe in this area, there are people who will get some benefit from this basic information. The other side of the coin is that just as many people are disappointed in the results. Either they haven't experienced obesity so they don't trust the result, or they already know they tend to overeat because they don't feel full. The test doesn't feel very helpful or legitimate to these folks. Finally, there are a few companies that are overemphasizing the impact of

genetic information in an attempt to sell additional products, such as vitamins or food, that claim they are going to somehow compensate for some genetic issue. There is simply no data that these things work. Therefore it is important to carefully research any company that you might get testing from, and involve an expert, like a genetic counselor or other care provider with the right experience and background, in the decision to get genetic testing for nutrition or fitness purposes. These experts can help an individual determine which tests have good scientific evidence, if the results have value for them personally, and if the resulting recommendations have scientific merit.

Why are there so few available academic programs for genetic counselling? My Canadian classmates all had to go abroad to Europe or the US because there were only 8 seats total in Canada (at the time).

And a fun question from someone who has also graduated from genetics (but only a BSc): do you prefer Bladerunner or GATTACA?

[huaer](#)

Hi Huaer! This is Erica, NSGC's President and Personalized Medicine Expert.

You are right, there are fewer programs in Canada than in the US, but that number definitely seems a bit low. There are currently 5 programs in Canada (<https://www.cagc-accg.ca/?page=117>) including one at UBC, home of NSGC Past President Jehannine Austin! (@J9_Austin on Twitter).

The number of genetic counseling training programs is growing quickly, although much of that growth is in the US. There are currently 43 accredited programs who are training students or will be accepting their first classes in the next few years. There are another six that have submitted letters of intent to build programs, with several others in the early planning stages. As importantly, the geographic distribution of programs is getting broader, the class sizes are getting bigger and there are more options for students. The Bay Path University program is offering a hybrid program with a mix of on-campus and online classes and Boise State University just announced that they will be the first online program.

With all of the genomics efforts in Canada and as the GC workforce gets bigger (so there are more GCs to supervise and train students), I'm optimistic that more programs and more slots will follow.

And while GATTACA has been the bane of my existence many many times, I have to admit that I'm a fan!

Thanks for your questions!!

23andMe recently received FDA authorization for reporting BRCA1/BRCA2 variants, but also provides genetic health risk reports for other genes and diseases. Has the company received FDA authorization for all of its genetic health risk and carrier status reports? When clients come to you for counseling based on data from 23andMe, do you recommend they get a confirmatory test using a different assay method?

[johndiegosan](#)

Hi, there! Amy Sturm, MS, LGC, NSGC President-Elect and Cardiovascular Genetics Expert here! The U.S. Food and Drug Administration approved the marketing of 23andMe Personal Genome Service Genetic Health Risk tests for 10 genetic diseases or conditions on April 6, 2017. These included Parkinson's disease, Late-onset Alzheimer's disease, as well as a number of other conditions: <https://www.fda.gov/NewsEvents/Newsroom/PressAnnouncements/ucm551185.htm>. Yes, when

patients come to genetic counselors for counseling based on their 23andMe BRCA results, we do recommend that they get a confirmatory test. As taken directly from the FDA's website on this topic: "Consumers and health care professionals should not use the test results to determine any treatments, including anti-hormone therapies and prophylactic removal of the breasts or ovaries. Such decisions require confirmatory testing and genetic counseling."

Are there studies on changing genetics of cancerous cells in order to make them kill themselves or of non-cancerous to make them stronger against cancer?

Is Genetic mutation on alive cells possible?

Is it true that Cloned beings tend to get cancer and die quickly?

[ardicli2000](#)

Hi! This is Joy Larsen Haidle, NSGC's cancer expert. Thanks for your interesting question. I had to do some digging to answer your questions as they are a little outside of what I do on a regular basis in clinic. I found a recent article by the National Cancer Institute that reviews some of the recent data and offers a nice summary relevant to your questions. The link is below:

<https://www.cancer.gov/news-events/cancer-currents-blog/2018/oncolytic-viruses-to-treat-cancer>

Hopefully you find the summary useful for your questions.

What are the career prospects for students coming up? I know GCs are in high demand, but there are only so many accredited schools - what advice would you have for students interested in this profession?

[genomegal](#)

Great question and an incredibly important topic. This is Erica, NSGC's President and Personalized Medicine Expert. I'm hoping some of my fellow NSGC Experts will jump in with advice since we have such a wide range of experiences, but I can share some of what I've seen around career prospects.

First, the number of genetic counseling training programs is growing quickly. There are currently 43 accredited programs who are training students or will be accepting their first classes in the next few years. There are another six that have submitted letters of intent to build programs, with several others in the early planning stages. As importantly, the geographic distribution of programs is getting broader, the class sizes are getting bigger and there are more options for students. The Bay Path University program is offering a hybrid program with a mix of on-campus and online classes and Boise State University just announced that they will be the first online program.

There are a lot of great resources for students at [aboutgeneticcounselors.com](#) and I encourage anyone thinking about genetic counseling as a career to visit [findageneticcounselor.com](#) and find GCs who will talk to students about the profession. Talk to a variety of GCs so that you have a complete picture of what the profession is like. Follow GCs on Twitter, including #GCchat and @GeneticCouns (NSGC's Twitter account). We're a pretty friendly bunch and we love engaging with prospective students! Participate in our consumer webinars and read our blog posts about genetic counseling (<https://www.nsgc.org/p/bl/et/blogid=53>). There are also some labs and universities that are hosting sessions on genetic counseling as a career so keep an eye out, especially if universities in your area have GC training programs.

Hope that helps [u/genomegal](#)! Thanks for your question!

Thank you all for taking time to answer our questions.

Do any of you have an opinion on, or experience with, targeted, pro-active outreach for at-risk families.

For example, one might consider providing information to females in families that have incidents of Fragile-X syndrome.

[Wrathchilde](#)

Hello, my name is Jay Flanagan, I am NSGC's Reproductive Expert. Thank you for taking the time to write in and ask a question.

Pro-active counseling and testing does have precedence in history. In the 1970s, it became possible to test for a disease called Tay-Sachs. Tay-Sachs is a significant condition in which babies appear normal for the first few months and then there is a rapid decline which includes blindness, deafness, and early death.

This is a recessive condition, so both parents have to be carriers. In most populations, it is fairly rare, but in persons with an Ashkenazi Jewish heritage, the risk to be a carrier is higher than seen in other heritages.

In the 1970s, it became possible to screen for this condition. Because of the significance of the condition, Jewish communities around the world began to encourage couples to seek out testing prior to having children. The population-based program has been so successful, it is estimated that the number of babies born with this condition has been reduced by 90% in some countries.

Carrier testing is now commonly woven into the care of patients who are of reproductive age in the United States. A number of conditions, including conditions like Spinal Muscular Atrophy, Cystic Fibrosis and Sickle Cell Anemia are recommended to be offered to couples considering having children by organizations like the American College of Obstetricians and Gynecologists and the American College of Medical Genetics.

Today, with modern approaches, most carrier screening tests include conditions like Fragile X and may include 100s of other conditions.

But that isn't necessarily the question you asked. While there have been approaches to pro-active testing, in many cases we fall short of actively helping family members who are at risk to have a genetic disease or have future children with a genetic disease. The primary approach has been by offering testing, but there is so much more to the story than just testing.

Genetic counselors are uniquely trained to explore with individuals what it means to have a genetic disease or at risk to have a child with a genetic disease. It isn't enough just to offer testing, it is more about exploring how knowing this information will impact them or change their future decisions. For some, the thought of knowing is more concerning than not knowing.

In our society, we often associate asking if someone wants to be tested for a condition as the same as asking if they want to know if they have a condition.... If we offer genetic testing and someone declines... they must not want to know. That isn't ultimately true though, offering a test does nothing to understand the psychosocial and physiological concerns a person has with knowing this information. In fact, this is a common reason that individuals do not make it to see a genetic counselor, they have been offered testing and declined or did testing, so the discussion ends with the test.

To find the answer to your question, it will take more than just one approach. I think we need to do a better job throughout society in engaging families. We need more support groups, more online communities, and more opportunities for education. We need to be active and more purposeful in our

education for sure. But we also need to make sure families know there is a safe place to go where they can to discuss what this all means for them. We need to make is "personal" to them.

I think we can take a lot from the history of testing for Tay-Sachs. The story is a success because families were educated about the topic, they had access to testing, but also that individuals had appropriate and meaningful counseling.

I always say that it takes a village.

To find a genetic counselor in your area, please visit <http://www.aboutgeneticcounselors.com/>

Thanks for asking,

Jay

Thank you all for taking time to answer our questions.

Do any of you have an opinion on, or experience with, targeted, pro-active outreach for at-risk families.

For example, one might consider providing information to females in families that have incidents of Fragile-X syndrome.

[Wrathchilde](#)

Hi! This is Erica, NSGC's President and Personalized Medicine Expert.

One of the unique aspects of genetics is that when you identify a genetic risk for disease in one person, many other people in the family could be at risk. This is especially important in conditions that we call dominant disorders - where the disease risk is passed from parent to child and all first-degree relatives of someone with the risk (parents, children, siblings) have a 50% chance of inheriting that risk.

To address this, genetic counselors always discuss the risk to family members with patients and encourage the patient to educate their families so that they can get tested. We also use a strategy called cascade screening to get very accurate and targeted testing. My fellow AMAer and President-Elect Amy Sturm just wrote a blog post on this that you can check out!

<https://www.nsgc.org/p/bl/et/blogaid=1024>

One really interesting opportunity that we have now that we live in a connected world is using technology to make those connections. A genetics lab just presented data at a big genomics meeting about their success offering cascade testing to their customer's families.

[https://www.genomeweb.com/molecular-diagnostics/researcher-presents-promising-early-results-color-cascade-testing-uptake-acmg?](https://www.genomeweb.com/molecular-diagnostics/researcher-presents-promising-early-results-color-cascade-testing-uptake-acmg?utm_source=Sailthru&utm_medium=email&utm_campaign=GW%20Seq%20Tues%202018-04-24&utm_term=Sequencing%20Bulletin#.Wt-CxNPwagw)

[utm_source=Sailthru&utm_medium=email&utm_campaign=GW%20Seq%20Tues%202018-04-24&utm_term=Sequencing%20Bulletin#.Wt-CxNPwagw](https://www.genomeweb.com/molecular-diagnostics/researcher-presents-promising-early-results-color-cascade-testing-uptake-acmg?utm_source=Sailthru&utm_medium=email&utm_campaign=GW%20Seq%20Tues%202018-04-24&utm_term=Sequencing%20Bulletin#.Wt-CxNPwagw). Both the lower price of testing and using better ways to reach families is a huge benefit that we will have to take advantage of!

Thanks for your questions!

Hello and thank you! I am just wondering how you picked your specialties? Was it something you kind of fell in love with or was it something you grew up loving?

[oif3gunner](#)

Hi! Amy Sturm, MS, LGC, NSGC's President-Elect and Cardiovascular Genetics expert here!

Regarding my specialty of cardiovascular genetics, I kind of fell into it, and now love it! I was recruited by a physician who saw a need in the area of cardiology and genetics and wanted to recruit a genetic counselor to build such a program. I had no specific training in cardiology or cardiovascular genetics specifically, but I was excited by the possibilities of using genetic information and genetic counseling to help prevent heart disease, the #1 killer of men and women in our country, so decided to go for it and learn on the job! It was very challenging but extremely worthwhile and I'm still working in this area and bringing it to the population screening phase! Thanks for the great question!

Hello and thank you! I am just wondering how you picked your specialties? Was it something you kind of fell in love with or was it something you grew up loving?

[oif3gunner](#)

Hi, this is Erica, NSGC's President and Personalized Medicine Expert. I'll echo Amy, I fell into my area and love it! My first job as a genetic counselor was seeing patients for preconceptional and prenatal genetic counseling and after about 4 years, I was ready for a change. I had an opportunity to help start a clinic that was focused on using genetics and medical imaging in adults at risk for genetic diseases (cancer, heart disease, etc) and I was very excited about the opportunity to build programs for those individuals from scratch. Unfortunately, that clinic closed but I had some great experiences that trained me in using software tools to improve processes, the business of health care and building products and programs - and I had the pleasure of being the first cancer genetic counselor in Las Vegas! I took all of that to a biotechnology company called Illumina and have spent most of the last six years building educational programs and working in market development around proactive and predictive genetic testing so that we can prevent disease rather than treat it. And my experiences in hereditary cancer helped to make that a passion project. We've gotten a little better at offering genetic testing to some high-risk patients but we have a long way to go. With sequencing technology getting better, faster and cheaper, we've never had a better opportunity to prevent disease!

Hello and thank you! I am just wondering how you picked your specialties? Was it something you kind of fell in love with or was it something you grew up loving?

[oif3gunner](#)

Hello, this is Jay, I'm NSGC's Reproductive expert. I also agree with Amy and Erica. I didn't even know about reproductive genetic counseling until I was hired by a Women's clinic. Originally, I was seeing patients in many different areas including oncology and prenatal. In our organization, we have a reproductive endocrinologist that loves genetics and genetic counselors and wanted us to be part of the clinic. Over time, I realized that I had a great passion for couples who were going through this challenging stage in their lives. As new tools became available, there were more and more options that we have to assist couples on their journey to having families. And genetics is playing a significant role in helping these families. That said, the most rewarding part of my job is not the genetics, but the support and encouragement that I can give them. It is so awesome when parents bring back their little ones and thank you for being part of the journey. Thank you for asking!

What is the professional view on prenatal screening and eugenics?

[TromboneEngineer](#)

This is Blair Stevens, NSGC's Prenatal Expert. The purpose of prenatal screening is to provide expectant parents with information about the health of their pregnancy. The National Society of Genetic Counselors advocates for informed choice and support our patients' right to choose how they

utilize information obtained from screening. The most important part of this very complex and difficult decision is to ensure patients are making decisions based on accurate testing (which means additional testing is often needed), up to date and balanced information and their decision is based on their own values and desires. While some women and couples may elect to end a pregnancy when a genetic condition is identified, many women and couples value the information because it can help them prepare them for a condition that is often unexpected. Pretest counseling about the benefits, risks and limitations of prenatal screening is highly recommended to help couples consider what they would do with information they may receive from a prenatal screen. To find a genetic counselor near you, please visit <https://www.nsgc.org/findageneticcounselor>.

First of all, thank you for doing this AMA!

I have a couple questions for Trisha Brown or Joy Larsen Haidle, but others are welcome to answer. I am currently an undergraduate student who is planning on applying to multiple genetic counseling schools next year. I've noticed that during my time with my undergraduate years and exploring the genetic counseling field, I've become very interested in the public health and policy topics involving genetics and the genetic counseling field in general. So I have a couple of questions:

- 1) What do you typically do on a daily basis that involves public health genomics, advocacy or policy? Do you feel as though you have room to be involved in that role and practicing in a clinical role as well?
- 2) What are some advice that you can provide a prospective student who is intersted in the genetic counseling field and public health genomics?

[Zevadis](#)

Hi Zevadis. This is Joy Larsen Haidle, NSGC's Cancer expert. Thanks for your question! Exciting that you are considering a career in the genetic counseling profession. The field is expanding rapidly and it is an exciting time to be a part of the profession. The skills of a genetic counselor are highly transferable to many work settings so that you have opportunities to continue personal growth throughout your career. I am a clinical provider in a community cancer setting. However, I have spent time educating and being an expert resource for payers and legislators as they try to set policy. I advocate for my patients to have access to appropriate care and coverage for tests or surveillance based on their mutation status or family history. My clinical experience is useful in adding a voice for the needs of families such as cascade testing, working on the Cancer Moonshot, or considering issues related to general population screening for cancer risk. My patients share their stories and experiences and it helps me have a broader view on topics. There is definitely room for both public policy efforts and clinical work. Many genetic counselors work in public policy to help optimize patient care.

As you contemplate graduate school, try to gain experience through job shadowing, volunteering and following topics impacting genetics/public health to help with your interviews and round out your experiences. During graduate school it is a good idea to gain experience in a wide variety of subspecialties as you may find an area that interests you that you had not considered. Plus it helps you be well rounded and flexible to join the work force and balance your personal/family needs too. Network and talk with colleagues to learn about opportunities that interest you throughout your career.

Hopefully this helps. Good Luck!

First of all, thank you for doing this AMA!

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[Zevadis](#)

Hi and thanks for your question. This is Trish Brown, NSGC's expert on Policy and Nutrition/Fitness. I am so glad to hear of your interest in policy. I specifically work with third party payers to help them with coverage policies for genetic services and genetic testing, and it sounds like you are thinking of public policy. In general, policies are very important in order to create a framework of communication between an entity, like a payer or a government body, and the people they are impacting. Both payers and the government are stewards of our healthcare dollars, and in addition, the government plays the role of ensuring a healthy and functional society. That includes programs beyond healthcare and moves into education, therapies, access to resources, and much more. The fiscal and financial responsibility is huge, and it is important to get the right balance of implementing rules based on scientific evidence while embracing innovation. There is definitely room to have both a clinical role and a policy role. Many of the members of NSGC have worked in the clinic as well as created policies at their own institutions, influenced third party payer policy, and even worked on legislation for genetic counselor licensure, Medicaid coverage, newborn screening, and other exciting topics. The clinical perspective is invaluable in order to accurately represent patient and healthcare provider concerns. As a student, you will find that there will be more than enough opportunities to learn about public health genomics and policies at local, state and Federal levels. When you interview at genetic counseling programs, definitely ask about your interests, and learn more about the clinical rotation sites. Some may have more opportunities than others that are aligned with your interests, but I know you will find something no matter where you end up. Thanks again for participating!

How do you address the ethics with the common practice in genetic counseling of not mentioning "incidental" or untreatable findings with patients, when those seemingly benign issues could actually be the cause of patients symptoms? This negatively impacts patient care- not only by gaslighting them into believing there is nothing actually wrong with them when there is and making them feel crazy, but also impacts the level of care they get for their health issues as docs then say "well you had a genetic test and all was normal so it must be in your head".

[Krabuki](#)

Hello! Amy Sturm, MS, LGC here, NSGC President-Elect. It would be helpful for me to better understand your question. Often times in genetic counseling, we do actually share information with patients that may not be treatable today. One example of this is increased genetic risk for adult onset neurological disorders. Also, genetic testing is not a perfect technology. There are many limitations. In fact, depending on what condition you're testing for, even when you know it's inherited, there may be a relatively low likelihood that today's available genetic testing and interpretation of testing results can give a definite "answer". Let me know if this helps or if you have follow up questions! Thanks!

What's the primary concern people have when first working with a genetic counselor?

[adenovato](#)

Hello, Ana Morales, here, NSGC cardiovascular and Spanish language expert. In my experience, the

primary concern people have when first working with a genetic counselor is thinking that the genetic counseling visit equals a commitment to pursue genetic testing, and that is not the case. Genetic counselors will gather medical and family history and let you ask questions so you can decide which risk reduction option fits your personality and needs. I wrote about this topic in a blog titled What to Expect When you Meet with a Genetic Counselor. The blog is written in English and Spanish (scroll down for the English version!): <https://www.nsgc.org/p/bl/et/blogaid=651>.

Two questions:

First, do those ancestry DNA commercials showing 1st world people going around and experiencing other cultures as if they're meant to be a part of them make you feel uneasy, too?

Second, I have read articles about viruses (?) being developed that attack specific types of cancer, and I'd love to hear your perspective on how promising this is for the near (10-year) future of cancer treatment?

[MarginallyCorrect](#)

MarginallyCorrect

Thanks so much, Reddit user, for raising your first point and sharing a feeling others likely have experienced as well. I'm Brianne Kirkpatrick, and in addition to being a licensed and certified genetic counselor, I'm a consumer of many different ancestry tests and genetic genealogist by hobby and passion. I can appreciate and somewhat recognize the uneasy feeling you describe from watching TV commercials about DNA testing.

'Cultural appropriation' is the term for what you're describing as happening in some of them. I am not an expert in sociology or psychology (those experts might best comment on reasons why a DNA commercial might lead us to experience discomfort), but I CAN comment on what I've seen and heard from others who have described ancestry testing having had a profound personal impact, and what makes me uneasy about the commercials.

It's a natural instinct and desire to want to belong to a group. For some, that desire for belonging is quenched by having living family and friends to relate to. For others, the results from a DNA test that tie their origins to a particular part of the world can hold a lot of significance. One person I spoke with who was adopted as an infant said that after doing an ancestry test, she didn't "feel like a space alien anymore." A DNA ancestry test helped her feel human and connected to others and to other cultures in a way people who are not adopted may never fully understand.

For me, what causes unease when I view most commercials regarding DNA ancestry is the failure of most commercials to show the work that goes into figuring out your actual ancestral origins. The commercials don't show that a lot of "discovering ancestry" comes from doing genealogical research... researching traditional paper records or microfiche documents (like birth certificates and immigrant ship rosters), for example.

DNA is only one tool used in putting together the pieces of someone's past, whether a recent or distant one. It's a misunderstanding to think that you'll do a DNA test and have your genealogical family tree unfurl the way a TV commercial might suggest. I wish it were so easy! In reality, genealogy requires a lot of time, work, effort, and research know-how.

A DNA result can mean a lot or very little, depending on the person; it's insightful of you to have noticed that these TV commercials led to strong, negative feelings in yourself, and to want to understand more of the 'why' behind it. Thanks for posting your question today during our DNA Day Reddit!

Side note from Brianne: [This article](#) posted on TheTempest.co comments on cultural appropriation and DNA ancestry testing. It hasn't been reviewed by the NSGC and therefore should not be seen as the opinion or stance of the Society, nor mine. I'm including it because it relates directly to this topic and gives more food for thought on cultural appropriation and DNA testing.

Some questions for Brianne Kirkpatrick, Trish Brown, and possibly others:

1. I understand it can have a significant dietary consequences if one's ancestors thousands of years ago were mostly farmers, hunter-gatherers, herders, foragers, or otherwise. For example, descendants of herders (like Northern Europeans and some East Africans) tend to have lactose tolerance. Descendants of farmers or foragers who ate mostly grains and/or vegetables (like South Indians) tend to be more primed to digest vegetables than descendants of hunter-gatherers, who are possibly more attuned to digesting meat. Do you have any other high-level dietary advice based on ancestry?
2. One's DNA becomes increasingly predictive of IQ as one gets older, and ends up explaining 70% or more of one's IQ late in life (which struck me as remarkable when I first learned it). See <https://www.sciencedaily.com/releases/2013/10/131001141218.htm>. Do you think it will be feasible for at-home genetics tests to check for the relevant variants and make reasonably good predictions of late-life IQ in the coming years? Or do you think at-home tests will just keep testing for a relatively minor handful of variants associated with intelligence for the foreseeable future?
3. At-home genetics tests frequently test for a variant associated with having fast-twitch muscle. There is also evidence that genetics might help explain why East Africans are disproportionately represented among winners of long-distance races and West Africans among winners of sprints, and I've heard it suggested that the same might be true of the Icelandic population in the realm of weight-lifting. Does recent research show any other notable ties between ancestry and specific types of athletics? Do you think at-home tests will start testing for such variants more expansively in the near future?
4. At-home genetic ancestry tests sometimes have trouble dealing with recent admixture between two fairly distant groups, and will often place people with such admixture among an "intermediate" population that is (to some extent) on a cline between them. For example, if someone's ancestry derives from Northern Europe and the Levant, he might be assigned to Italy or Greece, despite a lack of actual ancestry from those areas. Do you know how, if at all, popular at-home genetic ancestry tests are working to address this problem?

[Volucra](#)

Hi and thanks for your question! This is Trish Brown, NSGC's experts on Policy, Nutrition, Fitness and DTC testing. I am going to address your Q3 on the link between ancestry and types of athletes, first because that is one of my favorite topics. Research has definitely shown some association between different types of athletes like endurance and long distance running and ancestry groups, but for me it feels like an artifact of how those studies are conducted. Most studies to date are examining a specific population and group, such as Polish rowers, or South African soccer players. As a result you get data that supports the kind of muscle types and metabolisms that are good for that sport only and you aren't looking at the whole population. These studies also don't take into account culture and other environmental influences that might make region specific athletes. In David Epstein's book *The Sports Gene*, he addressed this question, and specifically commented on long distance runners that come from African countries. In some of these regions, running is a constant and normal part of society for entertainment, competition, and travel. Everyone runs. If you live in such a society, are you a great runner because you are very experienced at it, or your genetics evolved to support that, or both? For me, personally, after reading a lot of research in this area, the answer is that while genes (and

therefore ancestry) may influence whether or not a person is better at an endurance or power sport, it is exposure to a particular sport and a developing passion about it that makes one an athlete. More population data is needed to truly understand how the various athletic traits are distributed around the globe. Many at-home testing companies are now including information now about elite athlete genes, if you are more likely to be endurance or power, or how to modify your workout based on your muscle type. They will likely continue to expand in this area in the future. If you are considering at home testing for athleticism or fitness reasons, please consult a genetic counselor or other professional knowledgeable in this area to make sure that you are going to get the answers you are looking for before buying a test. <https://www.nsgc.org/findageneticcounselor>.

Some questions for Brianne Kirkpatrick, Trish Brown, and possibly others:

1. I understand it can have a significant dietary consequences if one's ancestors thousands of years ago were mostly farmers, hunter-gatherers, herders, foragers, or otherwise. For example, descendants of herders (like Northern Europeans and some East Africans) tend to have lactose tolerance. Descendants of farmers or foragers who ate mostly grains and/or vegetables (like South Indians) tend to be more primed to digest vegetables than descendants of hunter-gatherers, who are possibly more attuned to digesting meat. Do you have any other high-level dietary advice based on ancestry?
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[Volucre](#)

Hello Volucre, this is Brianne Kirkpatrick, NSGC's Ancestry Expert. I've seen your questions, and it is taking a bit of time to develop a response. Mainly because you have asked about complex topics, responses to which could fill a book chapter (maybe an entire book)!

The first three questions you posed have much to do with traits and body functions known to be associated with complex gene/variant/environment interactions. Variants that have to do with diet, IQ, and muscle twitching are not "predictive" in the same way that many cancer risk variants and variants conferring risk for genetic disease (like Huntington's disease) are. You can have a variant associated with a certain trait and commonly not show the expected trait, for example.

How demands for this type of testing will change over time (in comparison to testing for more traditional "predictive" testing of medical significance) will depend on how much value individuals & society place on information that isn't going to lead to firm recommendations. There are some great articles (in Genome Magazine, I believe) related to these topics that I will look for and post later as a follow up.

As for the fourth question, I am going to encourage you to direct it towards ancestry testing companies themselves. I am not privy to their priorities for addressing the various short-comings of their ethnicity assessment algorithms. The genetic genealogy community has representatives who share concerns with the companies, and I encourage you to join the [International Society of Genetic Genealogy](#) if this is an area of interest and you are not yet a participant in that society/community yet. That is the best way to have your voice heard on topics related to ethnicity calculations.

Thanks for posting!

Any of you familiar with Fructose 1 6 bisphosphatase deficiency? Would it be possible to do keto diet with it, or would it be too dangerous to try? What should you do if you have the mutation along with type 2?

[MsArtistick](#)

Hi, this is Erica, NSGC's President and Personalized Medicine Expert. It's fantastic that you're asking the question, but unfortunately we're not able to offer medial or dietary advice during this AMA. There are many genetic counselors and clinics who work with people who have genetic conditions that impact metabolism and can give informed advice about diet and nutrition. To find someone in your area that may be able to give you the appropriate guidance, you can visit [findageneticcounselor.com](#) and look for the Metabolic specialty.

Good luck!

What is a common misunderstanding people have about genetics?

[Sentry459](#)

Hi, this is Erica, NSGC's President and Personalized Medicine Expert.

DNA isn't destiny! Although there are a handful of genes that will cause disease 100% of the time, we've learned that other genetic markers may be protective for disease. So if you have a disease risk, you might also have something protective in your DNA. As we start to test more and more people, we'll not only have a better sense for how all of these genes work together but what they look like in the general population. It's exciting but it does mean that DNA and genetics and genomics are not going to be perfectly informative for awhile!

Great question, thanks!