Science AMA Series: 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.

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[1]Affiliation not available

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Hi there! I have considered having my genome sequenced many times, but have often been discouraged by those who cite risks regarding life insurance and other policies that may deny or charge more based on my knowledge of possible future risks. Is ignorance best in these cases or is the margin of error of genome sequencing high enough that most insurance companies won't consider it? Thanks!

tadroqs

Hi! This is Erica Ramos, NSGC's Personalized Medicine Expert. This is a great, and very common question. As a few other posters mentioned, the Genetic Information Nondescrimination Act (GINA) is a national law that protects against genetic discrimination for employment and health insurance. But there are exceptions – most notably, the military is not subject to GINA. Life and disability insurance and long-term care do not yet have these protections nationwide.

Like with many cases in genetics, the risks and the benefits depend on the individual. The benefits are definitely higher for those who have a medical condition and are looking for answers or who have a strong family history of conditions that might be genetic. Even people who are presumed to be healthy can learn important information from their genomes, but the odds of us finding anything "health critical" in a healthy person right now is not that high. As we learn more about the genome and how it relates to medical conditions, responses to medications and other health-related issues, it will likely become more valuable.

One last thing – I definitely wouldn't say that "ignorance is best". There are lots of ways to determine if you might benefit from genetic testing, including knowing your family history! If there is something that you're worried about, a genetic counselor can help you to determine the value of genetic or genomic testing. You can find a genetic counselor near you at nsgc.org using the aptly named Find A Genetic Counselor page! ☺

Hi there! I have considered having my genome sequenced many times, but have often been discouraged by those who cite risks regarding life insurance and other policies that may deny or charge more based on my knowledge of possible future risks. Is ignorance best in these cases or is the margin of error of genome sequencing high enough that most insurance companies won't consider it? Thanks!

tadroqs

Hi! This is Amy Sturm, LGC, licensed genetic counselor, and NSGC's Cardiovascular Genetics Expert, as well as past Board of Directors member of NSGC. Great question, and one I hear so often from

patients in the clinic or when I give talks to the general public! Some others have posted helpful information below, including a link to GINA, the Genetic Information Nondiscrimination Act. The Genetic Information Nondiscrimination Act of 2008 (GINA) is a federal law that protects individuals from genetic discrimination in health insurance and employment. You can find great info here: http://ginahelp.org/. It is true that there is no current legal protection against life insurance discrimination. You can imagine, that with the patients and families I serve, that this question often arises, since they may be finding out they are at risk for heart disease. We do recommend that individuals consider securing insurance prior to undergoing genetic testing.

Is there any way to get DNA run anonymously? I am curious to get my complete genetic makeup, but don't want it saved or associated with me.

carver

Hi! This is Erica Ramos, NSGC's Personalized Medicine Expert. Great question – a lot of people want to know if they can be tested anonymously. If you are doing genetic testing for a medical reason and want your insurance to pay for the testing, then no. But there are other limitations to doing testing anonymously if it is done for a medical reason. Specifically, if the testing shows something of importance and your doctor wants to order any follow up testing, studies, etc. and have your insurance pay for it, the insurance company will typically need that to be justified by your test results.

If you are doing this for your own information/education and are paying out of pocket, then maybe. It depends on the lab that you would be testing through. It also depends on the physician or genetic counselor who is ordering the test. Your healthcare providers take on responsibility for your test results and they may have an obligation to put those results in your medical record. I would recommend having that discussion with your physician or GC before any test is ordered.

23andMe is one way to get access to some of your genetic information without a healthcare provider. They offer what we refer to as "direct to consumer" or "direct access" testing. 23andMe can currently provide you with your carrier status for specific genetic variations that can cause ~35 conditions and can give information around non-medical traits (like hair color) and your ancestry.

Is there any way to get DNA run anonymously? I am curious to get my complete genetic makeup, but don't want it saved or associated with me.

carver

Hi! This is Amy Sturm, LGC, licensed genetic counselor and NSGC's Cardiovascular Genetics Expert and past NSGC Board of Directors member. I concur with the great reply from genetic counselor Erica Ramos below. I think talking to a genetic counselor about this question might be helpful for you, so that we could better understand your motivation for not wanting it saved or associated with you. You can always find a genetic counselor at nsgc.org and http://nsgc.org/findageneticcounselor.

I have been waiting for a Genetic Counselor AMA for a while! It's such an interesting topic and there is definitely not enough information about it online! I'm bookmarking your blog for future reference.

I have a few questions, mostly as someone who is hoping to apply for a genomic counseling qualification next year (although I'm in the UK).

1. It's a career I find massively interesting, however I do have doubts that I would make a good genetic counselor. Is this a common thought? How would you improve upon the specific skills required for the job? What experience would you say benefited you most in preparing yourself for the role?

@Jehannine Austin

1. 100% of my siblings have been affected by mental health problems so I find the genetic link fascinating. How much of a role is there a role for genetic counselling in mental illness? If none then do you see it becoming more relevant in the future if/when we find specific mutations that influence

our mental health?

2. Where do you think we will be in say, 10 years, with regards to genomics and mental health? I know that there is much research being done into the genetics/mental health, but (as far as I know) we haven't as yet found the brain's equivalent of the BRCA1/2 mutations?

3. What would you say has been the biggest discovery in terms of the genetics of psychiatric disorders?

alldreams

I am president of NSGC and am NSGC's resident Psychiatric Disorders Expert! :) I'll tackle the answers to the two parts of your questions separately - first, the career bit. I'm thrilled that you feel its massively interesting, I agree (of course!) ;) I think that it can be a bit intimidating to know whether its a good fit for you. Broadly, I's say that to be a good genetic counselor, it helps to be a good listener who is reaaally interested in people and their emotions....and genetics. I'd suggest finding genetic counselors to have a coffee (or cup of tea, perhaps, if you're in the UK!) with, and talk with them about their work. I'd also suggest taking a look at the Prospective Students section of the NSGC webpage: http://nsgc.org/p/cm/ld/fid=43 In terms of what helped me most, personally, to prepare myself for entry into my genetic counseling training program....I think it was spending some time training and then on the phonelines as a crisisline volunteer....its really important to know that you actually like counseling (rather than imagining that it might be a good fit!), speaking with genetic counselors about their work, and having a decent university level training in human genetics. My genetic counseling MSc training provided me with the confidence, competence and qualifications to do my work well :) Good luck to you with your explorations of this as a career!

Do you recommend any of the services like 23 and me for people who are healthy and have no obvious genetic problems? I am always curious about my genetics, but don't know if these services are accurate, or if they are secure (from insurance companies).

Osiris62

Hi, Joy Larsen Haidle here, NSGC immediate past president and cancer genetics expert. People pursue genetic testing for a variety of reasons. Before getting tested, it is important to determine your goals, meaning, what information do you hope to learn and will this test help answer that question? Also ask, What will I do with my results, and will they help me make health decisions?

When considering direct to consumer genetic testing options, it is important to read the information provided by the laboratory to determine what is being tested, how results will be shared, and the educational background of the people who are analyzing the test.

Also think about the type of information that may be available to you. Do you want to know only information in which you could do something to help prevent a disease or delay onset of symptoms? If there is nothing that can be done to treat or prevent a condition, do you want to know about a potential risk over your lifetime? Will I find out extra information that I might not be expecting?

Meeting with a genetic counselor can be useful prior to testing as often a discussion about the family history can help put some health risks into perspective. It is also useful to put the test results into context for your personal situation. Some people will choose to do testing at home. If a test result raises questions for you, we are happy to meet with you to discuss them.

Here is a link to a blog that expands on some things to consider before pursuing direct to consumer testing.

http://nsgc.org/p/bl/et/blogaid=577

How realistic is it that in the near future humans will be able to chose the genetics of their offspring? Will it be possible to possible to omit genetic disorders that would be passed on etc.

Kureeru

Hi, this is Jen Hoskovec, NSGC's prenatal expert. As others have mentioned, there are options for preimplantation testing for certain genetic disorders (known as PGD) when the underlying genetic cause/mutation of the condition has been identified. In these instances, embryos can be tested prior to transfer to determine if they are affected with the genetic condition. Genetic counselors who specialize in PGD can help families asses their risk of passing on a hereditary condition and talk through the benefits and limitations of such testing.

Are panels such as Myriad's MyRisk really a good idea in the panic caused by a new cancer diagnosis. I have the seen drastic, and ultimately wrong, treatment decisions such as bilateral mastectomy based on a VUS in a minor gene that subsequently proved non deleterious. Armed with this misinformation, patients cannot be talked out of unnecessary surgery.

docroberts

Hi! Mary Freivogel here, NSGC President-Elect and cancer genetics expert. There's no question that patients with a new cancer diagnosis certainly have a lot to think about and process. However, it is important that genetic counseling is offered to them at the time of their diagnosis, with the understanding that some patients may decline. If a patient chooses to have genetic testing, the results can change their choice of surgical treatment and even possibly their oncologist's choice of chemotherapy. Of course, thorough genetic counseling is important so that patients are not caught off guard by what the results of genetic testing tell them. It's important that patients understand the implications for them, in the short term and long term, as well as the implications for their relatives. I would argue that genetic testing is only as useful as the shared decision making that goes along with it. Emotions are very high at the time of diagnosis and genetic counselors understand this. Go to findageneticcounselor.org to find a genetic counselor near you.

To what extent do you think knowledge of one's genetic disposition affects behaviour both positively and negatively? I can imagine that in some situations, knowledge of a likelihood to have certain illness would lead to better health choices but for others it may lead to "fuck it I'm going to die of X anyway".

ImNotJesus

This is Erica Ramos, NSGC's Personalized Medicine Expert. The REVEAL study, mentioned by u/cariaso, was the first of its kind to look at how people responded to finding out genetic results for Alzheimer's disease risk. The participants from this particular study did have a parent with Alzheimer's disease and sought out genetic testing, so one important point is that it's a self-selected population who will seek out this type of information. In other words - people who know that they don't want to hear the answer won't ask the question.

There have also been some studies from the same group (Dr. Robert Green at Harvard/Brigham and Women's) looking at people who seek out personal genetic testing. Their PGen study found that "56% changed a health behavior (diet/exercise/medications/vitamins or supplements) within 6 months of receiving personal genetic testing results." They also didn't see what Dr. Green calls the "jelly doughnut" effect, where people saw that they are LOW risk for something like heart disease and went crazy on the jelly doughnuts because they thought that it didn't matter.

http://www.nature.com/gim/journal/vaop/ncurrent/full/gim2015192a.html#t4

Another study from Dr. Eric Topol's group at Scripps looked at individuals who had direct to consumer testing after 3 months and after 1 year. They didn't find any difference from baseline in anxiety, fat intake or exercise after 1 year. However, 61.5% of the participants still thought that the test was highly useful to them.

http://www.ncbi.nlm.nih.gov/pubmed/23559530

So I think that we've learned that people who want this information will likely deal with it well and

genetic counselors can help to support them if they have questions or concerns. How health behaviors change in the very long term (>5 years) is still somewhat unknown.

Thanks for the question!

To what extent do you think knowledge of one's genetic disposition affects behaviour both positively and negatively? I can imagine that in some situations, knowledge of a likelihood to have certain illness would lead to better health choices but for others it may lead to "fuck it I'm going to die of X anyway".

ImNotJesus

Hi! This is a great question - thanks for asking! It's Jehannine Austin here again, NSGC's president and resident Psychiatric Disorders Expert! For sure, knowledge about genetics has the *potential* to influence people's behavior in both positive or negative ways .....this is why genetic counseling is so important. Our role as genetic counselors involves helping people to *adapt* - in a helpful or positive way - to their understanding about the role of genetics in the condition that they or a family member has. I think that mental illness can actually be used as a great example here. For example, you can imagine easily that some people might think: "If genetics plays a role in mental illness, then there is nothing that I can do, I am predestined to develop it, given my family history....I may as well just accept that its going to happen." Under those circumstances, you can imagine that people might be less inclined to stay away from drugs like pot, or crystal meth (things that we know can increase a person's vulnerability to psychosis). Genetic counseling in these contexts is about helping people to understand that genetics plays a role, but is NOT the sole cause, and that there ARE things that they can do that could make a difference to their health (like staying away from pot/crystal meth) ....we are actively working towards helping people to avoid feeling fatalistic about things....in the context of most common health conditions that humans have, genes are way less deterministic than we may believe!

For Dr. Hoskovec,

Why am I so tall when my parents are so short?

Also, what are the odds that my daughter be tall?

More details -- I am 6'4" tall, and my two brothers are 6'3, and 6'1" tall.

However, my Dad was 5'9", and my mom was maybe 5'2"

A couple of the uncles on my mom's side were tall. Like 6'4" or taller.

The only brother who has had children, had triplets, and the two girls are both 6'1" tall (and no, Reddit, I will not post photos of my nieces for you).

As for my daughter, she's 10, and loves playing basketball, and fortunately she looks like her mother, but she's huge feet for her age. She's always had long feet. My wife is short at 5'2" (she says she married me because when I'm around, she doesn't need a stool to get the items from the top shelf of the cupboard. But I digress).

My daughter is amazing whether or not she'll be tall, but if odds are, she's going to be a six footer, she needs to keep playing basketball and start learning volleyball.

Mongo1021

Hi! This is Jen Hoskovec, NSGC's prenatal expert. Questions about height in families are very common and I wish I had the answers for you! Height is a trait that is difficult to study because it is likely due to a combination of genes and environmental factors (we call these type of traits multifactorial). To make things more complicated, the genetic portion of height is also complex, likely involving a combination of many genes. For that reason, height can be unpredictable in some families - as you outlined in your question.

Many times pediatricians can provide some estimate of adult height during childhood using growth

charts and parental height information. But as you know, this approach is not exact!

Thanks for the question and enjoy those basketball games!

In my father's family, it turns out all the men from my great grandfather to me have the same mild hearing problem in the left ear. The women have not gotten it. That's five out of five males, and zero out of three women. I spoke to a biology lecturer in college about this, and he said it's very unlikely the hearing problem has to do with the Y chromosome. So... Do you guys think it could realistically be a gene on the Y chromosome impacting our hearing, or is it more likely a dominant bad gene just happened to get passed down only to sons?

Rhamni

Hi Rhamni! This is Erica Ramos, NSGC's Personalized Medicine Expert. Very interesting question. I'll preface this by saying that I'm by no means an expert in the genetics of hearing loss, but this is my take! There are a few different possibilities, none of which are very straightforward, and this is almost certainly not a comprehensive list. I actually did find 2013 reference for a Y-linked form of hearing loss, but it resulted from a piece of chromosome 1 that was stuck to the Y chromosome. So that's one (very rare) possibility. The more likely possibility, if we assume that it is genetic, is that it is an autosomal dominant condition and COULD be inherited equally by both men and women in the family. However, this form of hearing loss might show reduced penetrance, meaning that some women have the variant but will not have symptoms. It also could be that only men got the variant just by chance and that future generations of women could inherit the variant. It is a very interesting history and a genetic counselor that specializes in deafness and hearing loss could be very helpful in sorting through the family history, reviewing audiology reports (which can be very informative when assessing genetic forms of hearing loss) and guiding you on testing if your family is interested. Check out the Find a Genetic Counselor tool on nsgc.org!

Thank you for doing this AMA!

I recently read an op-ed article commenting on the small number of certified genetics counselors, and the impact that limited access to genetic counseling has on health care for people who want or need to be tested.

As medical research moves incrementally closer to achieving the vision of precision medicine, do you expect that the demand for genetics counselors will continue to increase? If and when the medical field shifts much more toward detecting heightened disease risk that can be managed effectively with moderate lifestyle adjustments (rather than mainly focusing on detecting high risk for conditions such as breast cancer or Huntington's) do you imagine the role and/or training requirements for being a genetics counselor will change?

Basically I am wondering how the field will shift to accommodate both increased demand and changing expectations, but if you don't agree that one or both of these things are likely to happen soon, it would be great to hear your views on that as well. Thank you!

neurobeegirl

Another great question - thanks! This is Jehannine Austin here again, NSGC president. To handle the part of your question that is about demand first - we currently have around 4000 board certified genetic counselors in the US, and our numbers have increased by >80% since 2006. The most recent survey that NSGC did (in 2014) on the subject we found that wait- times for people to see a genetic counselor actually compare favourably with wait times to see many kinds of MDs, and that urgent cases are typically seen within a couple of days. So, that is reassuring, but we are still working hard to ensure that we are able to continue to meet the growing needs for our services as a profession. The other part of your question is about whether training programs will need to change over time - and the answer here is both yes and no! I say "yes" in the sense that the curricula of the Genetic Counseling training programs have always had to be evolving, given that this field is moving so fast! So, there are constant tweaks that are happening to ensure that our graduates are current as they can be with their learning.

But the "no" part of my answer is that there are no real fundamental, paradigmatic differences between genetic counseling for a common complex conditions like mental illness, and genetic counseling for the conditions that are typically considered as being within the genetic counseling "wheelhouse", like huntingtons. Our goal with training genetic counseling students is NOT that they have a complete body of knowledge to last them the rest of their careers when they graduate (it wouldnt be possible, because the field is moving so fast!), rather, we are training them to have a really solid foundational knowledge base, and skills and expertise in counseling and communication, AND to ensure that they have the tools to be the lifelong learners that practicing in this field demands :) Thanks for asking!

As a scientist who participates in clinical trials as a way to earn an extra buck or two, I often try to persuade the trial runners to share my genomic data with me. I have yet to find one that will, typically because their IRB protocol forbids it.

Why is our current regulatory system so terrible with sharing genomic data to patients or customers? Do you think it needs to change, or does the policy do more good than harm?

Cersad

Hi, Joy Larsen Haidle here, NSGC immediate past president and cancer genetics expert. This is a tough question! Thank you for your willingness to participate in research trials! Research is just that… research. We need to learn more information to make sense of a theory before incorporating the information into routine medical care. This sentiment makes research testing much different than clinical testing. With clinical testing, the data is already available to demonstrate utility.

Many of the conditions we see are quite rare and I appreciate the research opportunities for the families. I look for protocols that have the ability to share results when possible and confirm the result in a clinical laboratory before using it in clinical care. If the IRB protocol does not allow sharing of the information, that should be a part of the consent process during enrollment so people know what to expect, meaning I'm contributing to the overall knowledgebase in hopes to help someone else or I'm hoping to get something back that is specific to me. Does the research study help reach my personal goals?

The speed of testing options is rapidly outpacing the regulations to help protect the public. Education is needed for the public and policy makers to help ensure that people have access to the information they wish to know about their genetic make-up without fear of the information being used against them in an unanticipated way.

Based on my knowledge, having your genome sequenced only allows you to know what statistical data tells us (i.e. A person with this gene is prone to heart disease, or cancer, etc). How accurate is this description?

For a second question, do you feel that things like insurance costs/eligibility should be based on what our genome tells us? Based off your first answer, is what we know now reliable enough for this to be utilised today in the market? Finally, do you feel that genes that are actually inactive in the body, or could become inactive, creates a major flaw in having the ability to use a personal genome as an insurance evaluation?

fastenedrex

Hi, this is Erica Ramos, NSGC's Personalized Medicine Expert. I'm going to try to answer your question and those brought up in the other replies. As u/TheLordB mentioned, the information that we can extract from your genome depends on the specific gene and genetic variant. Using the example of cystic fibrosis, genome sequencing would detect most variants and we do know what many of those variants do - whether they are benign (not related to disease) or pathogenic (disease-causing). However, any one person could have a variant that has never been seen before. If a variant is essentially new, we typically have to call it a variant of unknown significance until we have more data, either from more individuals with the variant or from studies that look at how the variant affects the function of a protein or changes an organism. That gets to the question from u/fastenedrex. We may be

very good at detecting that a variant or mutation is present but not very good at knowing how that variant will act in people. And honestly, we're learning more every day, even about diseases that we thought we understood very well. Most of the genetic testing that was done in the past was on people who had symptoms of a disease or on people in families with a disease. We weren't just out there testing healthy people. Now we are and we are learning that many "well-described" genes and diseases are not quite as clear-cut as we thought. So in many cases, our ability to predict risk and outcomes may not be as good as we wish it was or thought it was. Additionally, there are so many cases where genetic and environmental interactions must be evaluated together, our genomes will never be a "one stop shop" for answers about our health. If someone told me that they could assess my risk for lung cancer by looking at my genome, but never asked me if I smoked or had exposure to asbestos or worked in a coal mine, I would be pretty suspicious of that risk.

The insurance question is complicated and I'm definitely not an expert in the actuarial science that goes into calculating risk for insurance companies. Fortunately, there are protections in place that restrict the use of genetic information in health insurance (see posts about GINA above), but life insurance, long-term care and disability don't have the same protections. Insurance companies are all about risk and they have definitely expressed concerns that they will not be able to maintain their business if the buyers know something about their health and risk that the insurer doesn't. As genetic testing becomes more prevalent, I think that these are issues that will have to start to be resolved.

Thanks for the great questions!

Thank you for taking the time to answer our questions. I have a few questions about Mitochondrial Disease and mitochondrial DNA.

Why is the test for mitochondrial DNA (muscle biopsy) so much more invasive than normal DNA testing? Is this something that will likely improve as our knowledge and technology in this area improves, or is that just the nature of the beast?

I've heard that most mitochondrial disease is passed down from the mother through mitochondrial dna, but it's possible to get mito (via nuclear dna) if both the mother and father are carriers. Can you explain how this works? Do the specific complexes that are defective in the parent(s) dictate the complexes which will test defective in the child(ren), or can mito be passed on in a different type from generation to generation?

heiferly

Hi, this is Erica Ramos, NSGC's Personalized Medicine Expert. Fantastic question and it can definitely be a little confusing. In fact, I just ran this by a genetic counseling colleague to be sure I was describing it clearly! I find it easiest to think of it in the context of mitochondrial disease. You're correct in that you have mitochondrial DNA, which is only inherited from your mother, and nuclear DNA which is inherited from both parents. Mitochondrial disease can be caused by genetic variants in genes from both mitochondrial and nuclear DNA because both can affect the ability of the mitochondria to produce energy. There are many more nuclear genes that code for mitochondrial proteins, so most mitochondrial disorders are the result of variants in the nuclear DNA.

The specific genes and variants will determine which mitochondrial disease is found in an individual or family. However, mitochondrial disease are very well-known as having variable expressivity. That means that different people, even within the same family, can have different symptoms. This applies to mitochondrial conditions from both sets of DNA.

Thanks for asking!

I am a potential genetic counseling student (waiting to hear back from admissions next week!). Is there any advice or information about the field of genetic counseling you would have wanted to know when you first entered the program? What are you favorite things about the field and in what areas do you want to see improvements?

Also, where do you see the field heading in the next 5-10 years? Personally I'm interested in cancer and adult genetic counseling, but please feel free to discuss prenatal or pediatric as I find it all very interesting!

gudfred

Hi, Joy Larsen Haidle here, NSGC immediate past president and cancer genetics expert. What an exciting time for you! I remember the waiting period being nerve wracking too! Much has changed in the profession since I was in your shoes, but my students help me stay in touch with the questions those entering the field face.

Because the field has grown quickly and the opportunities to use your skill set are expanding rapidly, my advice would be to explore as many subspecialties and work settings as you can as a student. The training rotations in graduate school will help expose you to areas of genetic counseling that you may not have considered, but may really enjoy and choose to spend more time following graduation. And always keep your eyes open for new opportunities to use your genetic counseling skill set that didn't exist at the time you graduate or help create the opportunities too!

My favorite things about the field are the people and the science…meaning, I learn something from my patients every day through their experiences and their stories. I am also continually learning from amazingly talented colleagues whom I have met at the conferences or in my volunteer work with NSGC. Our understanding of the science is continually changing, which is exciting and offers new insight into cases which keeps us on our toes and does not allow one to get bored.

In the next 5-10 years: Wow, that is hard to predict with the explosion of genetic testing options and the greater comfort level with genetics in the general population. The field will continue expanding and we will need to collaborate with our healthcare colleagues to meet the demand and provide the best patient care. Varied service delivery models will be useful to provide flexibility for schedules and help people access our service regardless of their location. My goal is to help consumers understand their family history and genetic testing options as well as how to use it in their medical care…that is a key part of precision medicine.

Best of luck to you!

Are children of incest really "deformed" like commonly believed? If so, why?

pumpkinsnice

Hi. This is Jason Flanagan, the NSGC's Reproductive Expert. Thank you for the question.

There are greater risks for children who have parents of very close genetic backgrounds. The reason is that relatives share similar DNA. The closer the relative, the more alike the DNA. If a trait or genetic predisposition is inhered as a carrier (one gene works well, one does not), the trait is most often not detrimental. Since closely related individuals have common genetic histories, they are much more likely to have both inherited a trait that is harmful. From there, if both relatives are carriers, they have a 1 in 4 chance of having a child with two inherited non-working genes.

For example, let's say that a disease happens in in in 1 in 10000 people. In children born from very closely related individuals, the risk may be 1 in 64 or even higher. Taking it one step further, we believe that most of us have between 3-10 significant changes in our DNA. If we are closely related, it is much more likely that a child from closely related family members will inherit two mutations or changes that will result in disease.
In general, the problem is that there is limited diversity in the genetic makeup and therefore more risk to have a problem. That said, it doesn't mean there will be a deformity.

Obviously, there are more than just genetic risk factors but psychological risk factors. If you have more questions about this topic, please visit your local genetic counselor. The www.NSGC.org website is a good place to find a local provider.

Hi!

My question is, do you guys think the MTHFR gene is hype, or something to be concerned with?

My Mom is homozygous for the C667T allele, and she's had 6 pulmonary embolisms.

I was tested and am heterozygous for both the C667T and A1298C. The doctor that tested me said that I need to take baby aspirin and never use anything with hormones(birth control) to avoid a clot. A while later a psychiatrist put me on Deplin(l-mythlfolate) for it. It didn't really help, but I was being treated for the wrong thing at the time...

My older brother went to genetic specialists and they told him there was no point testing because it's all hype. He is now completely convinced it is useless information.

I know there are other factors involved, but just wanted to see if you guys have any opinion on the matter. Is it something people should look into?

Thanks!

berfica

Hello, this is Jay Flanagan, the NSGC's Reproductive Health Expert. Testing for MTHFR is a very frequent question that we discuss with patients as it has become a commonly ordered test.

If you look online, you will find a lot of information on MTHFR. Studies have linked MTHFR to blood clots, heart disease, breast cancer and many other conditions. When these studies were done, we didn't realize that MTHFR is so common. In fact, we now know that around 50% of Americans are carriers for this condition. Currently, we believe that MTHFR has very limited clinical utility and groups like the American College of Medical Genetics suggests that we should no longer be testing for MTHFR https://www.acmg.net/docs/MTHFR_gim2012165a_Feb2013.pdf

If you are worried about your genetic test, one thing you could do is a homocysteine level. If your homocysteine level is normal, there is no further treatment. If your homocysteine is elevated, your doctor may give you extra folic acid. That said, there limited data to suggest that treatment for elevated homocysteine levels will reduce the risk for heart disease, blood clotting, or any other associated problem.

One area of concern is that MTHFR has become a common test outside of the medical community. In this case, often the test is done to help sell expensive supplements. Considering over half of individuals who do the test will be positive, this has become a huge industry to break in to.

For those considering testing for MTHFR or have tested positive, it would be important to consider the benefits and limitations of this information. The best place to start is with your local genetic counselor. You can find your local GC at www.NSGC.org

For diseases that have both genetic and behavioral components (high blood pressure, many cancers) why is genetic screening not part of a standard health checkup? Are the genes really just not known? It would be great if those of us without a genetic susceptibility to high cholesterol (say) could chow down on shrimp alfredo.

IsThisNameTaken7

Hi! This is Amy Sturm, LGC, past NSGC board of directors member, and the NSGC Cardiovascular Genetics Expert. You're right, genetic screening is not part of a standard health checkup at this time for complex diseases like hypertension. This includes other common, complex diseases like coronary artery disease, diabetes, and many cancers, too. By complex, we mean that they are multifactorial in nature and there are many risk factors, including environmental exposures (smoking, poor diet) as well as genetic factors! Many genes and variants have been discovered in research that contribute to these types of diseases. The current issue is: does adding this type of testing 1) Add to the specific risk assessment for a patient? and 2) Help to motivate healthy behavior changes? Ongoing research will help us answer both of these questions, and if one or both answers are YES, then I think, in the future,

we may well indeed see genetic testing for common complex diseases as part of the screening process. It is also important to know that there are specific hereditary types of high cholesterol and cancer. There is a common condition called Familial Hypercholesterolemia. I've written an NSGC blog about it, which can be found here: http://nsgc.org/p/bl/et/blogaid=449. I've also written a blog about red flags that may indicate you have another type of hereditary heart disease: http://nsgc.org/p/bl/et/blogaid=568. And, I think shrimp alfredo - in moderation - might be okay!

It seems like genetic counselors have to wear a lot of hats as the bridge several different disciplines. What kinds of qualities would you say are important to develop for those considering genetic counseling as a career?

pensivebadger

Hi! This is Amy Sturm, LGC, NSGC's Cardiovascular Genetics Expert, and a previous Board of Directors member. I love this question! You are absolutely correct in that we DO have to wear a lot of hats! I think this is one of the reasons people want to go into genetic counseling as a career actually! You get to wear the clinician, scientist, and counselor hat all in a day's work! I think individuals who are considering genetic counseling as a career should be excited to be life-long learners and be adaptable to change! I have been a genetic counselor since 2002 and our field has changed and grown tremendously since that time! The genetic and genomic testing available today is amazing, but we need to have individuals who can stay current with cutting edge science and testing! Another very important quality is empathy. As a genetic counselor, we work with patients, clients, research participants, colleagues, customers, etc. from a wide variety of areas, and empathy is a key quality that needs employed on a daily basis. For specifics on prerequisites for applying to a genetic counseling graduate program, there is great information that can be found on nsgc.org and abgc.net (http://www.abgc.net/Certification/become_a_genetic_counselor.asp). This is also a great site and resource about becoming a genetic counselor! http://www.becomeageneticcounselor.org/faces_of_genetic_counseling

Hi,

I'd like to ask about pre-diagnosis for couples considering to have children: Two of my brothers (out of 9 siblings) have a rare genetic disease (malignant osteopetrosis; one died, the other is in quite bad shape). It is recesive and very rare, but also fatal. I know that I carry the gene. How and where can I test for a specific condition like that, to assess the risk that both me and my partner are carriers, if we ever want to have children? Is this kind of testing accessible to the general public?

Thanks a lot for your work!

caracatrepa

Hi, Jen Hoskovec here - NSGC's prenatal expert. Many have already chimed in with some great information. I will reiterate that general population expanded carrier screening can provide couples with a lot of information for family planning. However, for those who have a very specific family history such as yours, I would agree that a formal genetic counseling consultation would be helpful so that the testing is most appropriate and meaningful for you. I imagine this is something you have thought a lot about given what you have gone through with your brothers and I wish you the best of luck in this process. www.NSGC.org has a "find a counselor" tool that may be of help.

I'm interested in possibly joining this field. I'm already doing genetics studies, and I was wondering if a psychology degree is required to get certification?

kaspookaboo

Hi! This is Erica Ramos, NSGC's Personalized Medicine Expert. There are a few answers to this question farther up the page, but I wanted to quickly answer yours. It's not necessary to have a

psychology degree, but psychology and counseling coursework may be helpful to have in your background when applying for genetic counseling programs. Check out the Prospective Students page on the NSGC website for more info: http://nsgc.org/p/cm/ld/fid=43.
Good luck with your studies!

This question is, I suppose, for Mary E. Freivogel.

I'm a adopted pediatric cancer survivor which means I obviously have some risk factors for cancer but I also have essentially no family medical history.

I've got three kids and I'd like to get some idea of what their risk factors are for cancers, specifically pediatric cancers. What, if anything, can I do to get this information?

Killfile

Hi, this is Erica Ramos, NSGC's Personalized Medicine Expert. And more importantly to this question, I spent six years working as a clinical cancer genetic counselor. I would recommend a consultation with a cancer genetic counselor in your area. You can find one at www.findageneticcounselor.org. The likelihood that your cancer was related to a hereditary cancer variant depends on the type of cancer and that directly impacts the risks for your children. When you have your counseling appointment, bringing copies of the pathology reports for your cancer and other medical records if possible. It will be very helpful to your genetic counselor and will help you to get the best possible information. Best wishes to you!

Hello, thank you for taking time out of your busy days to answer questions.

I'm a Caucasian of Mediterranean decent who lives in Asia. I'm 13w pregnant with my Asian husband's baby.

I just found out I tested as T/T for MHTFR. The doctor here pointed to the results and said we need to be careful and take extra folic acid supplements and then... No other explanation.

I've been googling and it seems this homozygous mutation causes... Everything.

In the near term, I'm worried about how to keep my baby healthy. I read to not take folic acid supplements, but instead consume as much Folate as possible.

What are the risks of complications for my mutation and how do I manage them?

For the long term, my father's mother had alzheimers and my mother's side history in not sure about. My mother is someone easily addicted to meds, gambling, alcohol, etc. Depression is pretty bad with her.

What are the risks that I will go down these paths myself and how do I avoid it?

Thank you so much and I'll be sure to spread any information I recieve to others who might benefit.

lili_misstaipei

Hello, and thanks so much for raising this! Its Jehannine Austin here again, NSGC President and resident psychiatric disorders expert. Actually, we get questions so often about MTHFR and what it might mean, that one of our members did a FANTASTIC blog post about it for us...(although it specifically mentions a different variation in the gene, the concepts can be applied to the variation you mention too) its lighthearted in tone, but the content is real....take a look and see what you think: http://nsgc.org/p/bl/et/blogid=53&blogaid=613 You used the word "cause" - you read that it seems to cause everything...and I can absolutely appreciate that this must be a terrifying idea. So, I want to reassure you! This is one tiny variant that has been *associated* with everything....and associated in this case does not mean cause. We know that the things you are worried about - addiction, depression, alzheimers - these are complex disorders....that means that they arise as a result of the combined effects of genetic variationS (plural! And probably lots and lots of them!) acting together with our

environment....AND (though you will absolutely find lots of scary articles around the internet, for sure) the MTHFR variation has not been definitively agreed upon by the scientific community as playing a role in any of these conditions. Here is an article I wrote from the NSGC blog that covers some of the questions you mentioned about depression and addiction: http://nsgc.org/p/bl/et/blogaid=357 Hope this helps.

My son has tuberous sclerosis, a condition he inherited from his mother. Both function normally physically and mentally. What can be done so that my son does not pass this trait to his children?

luvmynutbag

Good afternoon! Jen Hoskovec here, NSGC's prenatal expert. For individuals who have an inherited condition, there are options such a preimplantation genetic diagnosis (PGD) paired with in-vitro fertilization (IVF), that can be utilized to assess the genetic makeup of embryos prior to pregnancy. A genetic counselor who specializes in PGD can be a resource to answer questions and discuss the feasibility, benefits and limitations of this option when the time is right for your son and his partner.

Heart disease runs in my family (just about every male as far back as anyone can remember has had some sort of heart problem); is there anything i can do to prevent my children from inheriting it?

FutureStan2616

Hi! I am Amy Sturm, LGC, NSGC's Cardiovascular Genetics Expert, and previous Board of Directors member. Please check out my blog for information on whether you might be at risk for hereditary heart disease! You can find it here: http://nsgc.org/p/bl/et/blogaid=568. Heart disease is very common, and it does sound like you have a strong family history. I would start gathering your specific family history information. For example, did you relatives have coronary artery disease, heart attack, arrhythmia, stroke, high cholesterol, aneurysm, cardiomyopathy, or some other type of heart disease? This type of information will help you and your doctor figure out if there may be a concerning pattern in your family. If so, I would recommend meeting with a cardiovascular genetic counselor. It can be determined whether genetic testing might be right for you, and your family. If your family has a specific type of hereditary heart disease, children can undergo genetic testing, too, to see if they inherited the risk variant, or not. If your children are already born, then you cannot currently change their genetics. If you want to have future children, and a known disease-causing genetic change is running in your family, specialized reproductive technologies like in vitro fertilization and preimplantation genetic diagnosis can be done, and have been utilized successfully for people with certain types of heart disease in their families. You can also help your children by getting them started with healthy lifestyles now! Your pediatrician can help with this, too, and there is great information on websites such as the American Heart Association.

Is there any correlation between physical appearance that is similar to a relative and a genetic predisposition to illnesses.

In other words, if I look like an uncle, will I be more/less or not correlated at all to have the same genetically related illnesses?

Hvitacristr

Hi! This is Erica Ramos, NSGC's Personalized Medicine Expert. VERY common question and this can definitely be the source for a lot of misconceptions about how conditions are inherited in families. There are some physical traits that are correlated to risks of genetically-influenced conditions. For instance, certain genetic variants in the MC1R gene make it more likely that you will have red hair, freckling and sun sensitivity. Variants in this gene have also been associated with an increased chance of developing melanoma, a type of skin cancer. There are also genetic conditions that have very specific physical traits associated with them. Individuals in a family who have Marfan syndrome may all be tall and thin individuals with long arms and similar facial features. So it really depends on the trait

and the condition. On the other side of that, when I was a cancer genetic counselor, I would often talk with patients who said "I don't look anything like my mom, so I'm sure I didn't get her BRCA mutation." or "I KNOW that I have the same genes as my aunt because we're so much alike.". In those cases, the condition and the trait are not linked and that can lead to some unpleasant surprises when it comes to discussing results. That's one of the things that genetic counselors really focus on with patients - what are their beliefs about the conditions in their family and whether those beliefs are consistent with the science/genetics.

These questions are directed mostly at Erica Ramos, however I hope everyone weighs in.

- What are the implications of genetic engineering as it interacts with epigenetics? Would using techniques such as CRISPR, obliterate or damage epigenetic markers on the manipulated segments? Could such markers be included in the engineering process in order to further influence the expression of recombinant DNA?
- The CRISPR method has brought diy bio to the masses (sort of) and proves to be effective. Are their any other developing gene - manipulation methods that are similar to it in terms of accessibility and efficiency?

Thank you for your time

Umbrifer

Hi, this is Erica Ramos, NSGC's Personalized Medicine Expert. Excellent and very heady questions! I'm not an expert in CRISPR or genetic engineering, but "off-target consequences" are certainly a big concern and one that is being researched extensively. It has already been described that CRISPR can introduce unintended double-stranded DNA breaks outside of the target, resulting in the introductions of indels. The location of those indels have the potential to result in frameshift mutations if they occur within genes or they could impact gene expression if they occur in a regulatory region. I found at least one study that used CRISPR to modify methylation, which is a common epigenetic mechanism. Using CRISPR to edit the epigenome is also being studied and that could impact gene transcription. So I think that all of these things are possible and all need to be researched more thoroughly! I'm not familiar with methods that are similar to CRISPR from the perspective of accessibility or efficiency.

On a side note, if you would like to hear from people far more versed in CRISPR than me, I would highly recommend a video recording from the 2015 National Society of Genetic Counselors Annual Education Conference. The session was called "Hope, Hype and Horror Movies: Contemplating Human Germline Modification" and journalist Carl Zimmer and genetic counselor Laura Hercher discuss the development of the technology and all of the MANY issues that surround it, from practical to ethical (https://www.youtube.com/watch?v=0gUO2Qupzeo). Carl also has some fantastic Radiolab episodes and other articles focused on CRISPR.

Thanks for the question!

Hello! Genetic Counseling is one of the things I thought of doing after college. Currently a sophomore studying biology.

What are some things I can do to better prepare for a masters program in GC?

Will having a chemistry minor make or break me for the field or applications? My major requirement is up to Organic I, and I'm not sure if I should keep doing more chem for the minor or do something I love (a music minor) or something maybe a little more applicable (computer science minor).

Jaesch

Hi, this is Erica Ramos, NSGC's Personalized Medicine Expert - and 15+ year GC. :) So great to hear that you're considering a career in genetic counseling. I've worked in many areas, from clinical to lab to business, and loved them all. NSGC President Jehannine Austin answered some of this above, so I'll just reiterate that there are resources for prospective students available on the NSGC website -

http://nsgc.org/p/cm/ld/fid=43. As for the best preparation, reach out to genetic counselors in your area and talk with them! This is a great, but challenging field so it's really important to know that you really want to be a GC when you're applying to programs. Good luck!

I've heard of a recent genetic breakthrough called CRISPR that allows scientists to genetically engineer a variety of animals. How soon will parents be able to genetically engineer their children? To what effect will this change our society? Will only the rich be able to afford designer babies? And if so will that doom children of parents not rich enough to genetically engineer their children to a life of poverty and underperformance compared to genetically designed children?

PurpleMufin

Hi, this is Erica Ramos, NSGC's Personalized Medicine Expert. Excellent questions about a complicated topic. I don't think that I can do justice to this topic in a few paragraphs so I would highly recommend a video recording from the 2015 National Society of Genetic Counselors Annual Education Conference. The session was called "Hope, Hype and Horror Movies: Contemplating Human Germline Modification" and journalist Carl Zimmer and genetic counselor Laura Hercher discuss the development of the technology and all of the MANY issues that surround it, from practical to ethical (https://www.youtube.com/watch?v=0gUO2Qupzeo). Carl also has some fantastic Radiolab episodes and other articles focused on CRISPR. Thanks for the question!

Do you agree with the FDA decision to limit access to services that provide direct-to-consumer personal genetics information? For example, 23andMe was recently barred from releasing health-related SNP screens to consumers, and must now stick to lineage analysis. From a big picture perspective, do you support patient access to direct genomics information, or should this information be "hidden from view" to protect laymen from self-diagnosis?

whiteknight521

Hi, this is Erica Ramos, NSGC's Personalized Medicine Expert. The National Society of Genetic Counselors has a position statement on DTC testing.

DIRECT ACCESS TO GENETIC TESTING: The National Society of Genetic Counselors believes that people interested in at-home DNA testing (also known as direct-to-consumer, DTC, or online genetic testing) have a right to make an independent, informed decision about whether to pursue this form of testing. Companies that offer direct access to genetic testing have a responsibility to offer consumers easy access and/or referrals to appropriate resources and qualified genetics professionals, such as genetic counselors. http://nsgc.org/p/bl/et/blogaid=370

There are always going to be companies that do genetic testing well and provide quality information and companies that do not. The FDA's decision to get involved highlighted a desire to ensure quality testing when other regulatory bodies, like those that govern clinical laboratories, are not involved.

How's the job out look there? I thought this was something 8 years ago that would be taking off, but there's still doesn't seem like demand has increased as predicted. Is there a glut of graduates?

applebottomdude

Hi, this is Erica Ramos, NSGC's Personalized Medicine Expert. Answering u/Zazetsumei question as well. :) There are several answers above that talk about the growth of the genetic counseling profession. Here in the US, the job market is excellent. One recent article highlighted the fact that there are fewer than 300 counselors that will graduate this year compared to about 650 job openings around the country. Jobs in industry and laboratories are growing at a particularly rapid rate as more genetic tests are being offered and the skills of genetic counselors are being used more broadly. There is some great data around job opportunities and salary that is summarized in the NSGC Professional Status Survey here: http://nsgc.org/p/cm/ld/fid=68. I couldn't be more excited about the opportunities

available for genetic counselors!

Two months ago I had brain surgery for a brain tumor removal. Last week I had a thyroidectomy for thyroid cancer. I am relatively young. Would genetic counseling benefit me?

Technologian

Hi, this is Erica Ramos, NSGC's Personalized Medicine Expert. And more importantly to this question, I spent six years as a clinical cancer genetic counselor. I think that genetic counseling could be very useful and it is standard to recommend genetic counseling in most cases where one person had two separate cancers, particularly if they are young. I would suggest looking for a cancer genetic counselor using the Find a Genetic Counselor tool at nsgc.org. When you have your counseling appointment, bringing copies of the pathology reports for your cancers and a detailed family history will be very helpful to your genetic counselor and will help you to get the best possible information. Best wishes to you!

How serious is a mthfr c667t gene mutation?

What tests and/or interventions are worth trying to mitigate the effects?

How would you tell if they were working?

thisjibberjabber

Hi! This is Erica Ramos, NSGC Personalized Medicine Expert. To echo u/GoodMutations, this is VERY common. You can tell because at least four other people have asked about it in this AMA! NSGC President Jehannine Austin addressed this perfectly above. :)

https://www.reddit.com/r/science/comments/4fmwrj/science_ama_series_were_experts_with_the_national/d2ap1dv

I am actually applying for genetic counseling programs next year!

I was wondering if any of you had any tips for strengthening one's application or any recommendations on any particular programs that you think would be most beneficial for a budding young GC. What should you look for, what experience do you need, what would make an applicant stand out, etc.

Thank you very much for your time!

badwolfettee

Hi badwolfettee! This is Erica Ramos, NSGC's Personalized Medicine Expert. Very excited to hear that you're applying for programs - it's a great field! NSGC President Jehannine Austin had a great response to this above - https://www.reddit.com/r/science/comments/4fmwrj/science_ama_series_were_experts_with_the_national/d2amyb1

Good luck with your applications!

So if my father has dimentia.what's the chance that I will get it.(son if that matters)

thisMFER

Hi, this is Erica Ramos, NSGC's Personalized Medicine Expert. Dementia is a symptom of several different conditions, including Alzheimer's disease and others. The chance that you would develop dementia depends on the cause of your father's disease. A genetic counselor that specializes in neurogenetics can help to review his history and to determine if any genetic testing might be helpful in assessing your risk. You can search for someone in your area at www.findageneticcounselor.org.

What is your opinion on the future of personal genome sequence privacy? Who should have access to the information, and where should the line be drawn? Should everyone in your family have access, perhaps it will be a legal right? What about insurance companies?

Its_Your_Father

Hi! This is Amy Sturm, LGC, licensed genetic counselor and NSGC Cardiovascular Genetics Expert, as well as past NSGC board of directors member. Number one, I think that each individual's personal genome and who has access to it should be the decision of that specific person. Informed consent, a PROCESS, not just a signature on a form, should be undertaken, so that individuals really know and fully understand who can access their genomic sequence information. You should have access to your personal genome information. If you want it shared with researchers, either currently or in the future, official consent should be obtained from you to do so. I'm not sure if it will be, in the near future at least, a legal right for everyone in your family to have access. There are benefits to this. However, not everyone in families want to know, for example, whether they might have a genetic predisposition to something like dementia. The Genetic Information Nondiscrimination Act of 2008 (GINA) GINA makes it against the law for health insurers to request, require, or use genetic information to make decisions about your eligibility for health insurance, your health insurance premium, contribution amounts, or coverage terms. Based on this, healthy insurance companies should not be able to request or access your personal genomic sequence without your permission.

Are any of you in the NYC area and accepting new patients?

funklordtoejam

The National Society of Genetic Counselors has a "find a counselor" tool on their website www.nsgc.org

There you can find certified genetic counselors by specialty in your area.