Science AMA Series: I'm Nicholas Katsanis, a human geneticist at Duke, let's have a conversation about human genetic disorders: facts, dreams, and most definitely the eradication of unicorns, AMA!

$\mathrm{Dr}_{N}ico_{K}atsanis^{1}andr/ScienceAMAs^{1}$

[1]Affiliation not available

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Abstract

Greetings from sunny Greece, where I am taking a few hours to chat with you about human genetics on reddit. My name is Nicholas Katsanis, but please call me Nico. I am a human geneticist, and the Director of the Center for Human Disease Modeling at Duke University. My passion has always been to understand human genetic disorders all the way from the discovery of genes that cause them to dissecting pathomechanism and thinking about the possibility of developing new therapies. Over the years, my team and I have worked to identify genes that cause a range of disorders, with an emphasis on rare pediatric traits. As part of that journey, we have begun to appreciate how the context of the genome can alter the impact of deleterious mutations and impact clinical outcomes profoundly. In that context, we have also realized how the complexity of the genome poses a real challenge in understanding pathomechanism as well as predicting outcomes for patients; we are working hard to develop new biological tools that can help us interpret the functional consequence of genetic variation. In parallel, we are working to build a path towards integrating the research and the clinical enterprise as a way to improve the impact of genetics in health care. Today, I am happy to field any and all questions about human genetics, from why Mendel's peas are truly wrinkly to what the major stumbling blocks are to really accelerating the development of therapeutics. I'll be back at 1 pm ET (10 am PT, 6 pm UTC) to answer your questions, ask me anything!

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DR_NICO_KATSANIS R/SCIENCE

ABSTRACT

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Dr_Nico_Katsanis , r/Science , Science AMA Series: I'm Nicholas Katsanis, a human geneticist at Duke, let's have a conversation about human genetic disorders: facts, dreams, and most definitely the eradication of unicorns, AMA!, *The Winnower*

Hi Nicholas, and thank you for doing this AMA!

A couple of questions for you:

1. Prenatal testing has come to be seen as a logical extension of the genetic revolution because it can help prospective parents have healthy babies. Yet disability rights advocates raise important questions about this idea. Adrienne Asch, for example, argues *Do not disparage the lives of existing and future disabled people by trying to screen for and prevent the birth of babies with their characteristics.* The issue seems to get at the heart of what is 'normal' and what does it mean to be 'healthy'. What role do you think genetics has in this conversation? How do you think about this tension in your own work?
2. Human behavior. To what extent do you think human behavior is determined by our genes (if at all)? Our some types of behaviors more or less genetically determined (i.e. dietary preferences, predisposition to violence, humor etc)?
3. As a human geneticist, what does 'race' mean to you, and what value is there (if any) to dividing humans into racial groups?

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SirT6

Hi, these are really interesting (and tough) questions. Let me see if I can address them one at a time: 1. I think that disability is, in many instances, in the eye of the beholder and is not always seen as a deficit, just as a difference. People with deafness is one example, people who choose to adopt children with Down syndrome another--there are many others. In a, way, it's not just about what is "normal" but what one's expectations are of a quality of life. I think we would all agree that a genetic defect that kills infants qualifies as a "disability" across the spectrum, whereas a gene that causes progressive blindness may or may not. I think that the KEY role of genetics is to highlight our individuality and to help us make choices based on our individual moral compass (as long, of course, as our choices do not impact negatively societal laws or encroach on others' ability to likewise make their own choices) 2. This is the age-old nature v nurture argument. The bottom line is that we do not know exactly. There is no question that there are aspects of our personalities that are genetically determined, or at least their boundaries are. The rest, is the envirment (macro and micro) 3. I honestly do not know what race is. I guess we define it based on where people come from and what they look like. Yet here i am, born and bred greek, blue eyes and all and according to my genetic data, i am 12% middle-eastern. Humans are cocktails and the faster we realize, the better it will be for all of us. The main value is when we study disorders, the genetic background (that could be defined as "race") makes a difference in different susceptibilities to different disorders

How realistic is the possibility of one day being able to have your children "custom-made" on a genetic level? Most importantly will it be possible to "install" genetic predispositions for e.g. high intelligence, musical talents, language acquisition etc

olinko

I guess these thoughts have been on everyone's mind since we figured out what genes are. What has changed in our ability to access them. In some ways, many people have been practicing this already...think about individuals who choose to terminate female pregnancies for societal or economic reasons. At the level of gene editing, yes, i think that technologically it will be possible to do. Moral/ethical? Not so sure. Also, it is important that people recognize the major dangers of this. For all we know, removing an apparently "undesirable" mutation might protect from #1 but open up susceptibility to #2. I also think that some of the traits that we might desire for our children (such as you mentioned) are quite complicated, so we would need to be editing dozens or maybe 100s of sites to get a desirable effect. Not clear how feasible that is. I also need to mention that there is a clear tendency for our society to slip onto Orwellian fears about every new technology that comes along. That is NOT to say we should not be having a conversation, quite the opposite. But at the same time, we need to stay impassionate and realistic. Extreme uses of every technology will always happen, that is human nature. The other thing to say is that the bigger danger, right now, is not the technology but the charlatans who are promising snake oil cures to desperate people and exploit families ad libitum. I would want to guard against that first.

I'm a big fan of science, genetics in particular. That being said, my son has been found to have a muscular "disorder". Initially discovered due to slight developmental delays and blood CPK testing, they first told us he had muscular dystrophy, which was hard for the wife and I. After some genetic testing all we know is that he has an unknown variant (one nucleotide substitution) in a muscle gene. But no mention of this in the medical literature. His development is normal for a 1.5 year old at this point, so either it is a rare variant, or it is benign. So lots of uncertainty in the future.

My question is whether medical journals accept findings that are non-pathological. Perhaps someone has found this variant elsewhere but since it could be benign, it may not be reported.

A further comment. Years ago we would have been blissfully unaware. Modern medicine has made us worried.

closethird

The easy question first: most major journals do not have a good format for "benign" variants. However, we do have accrual databases that by now have upwards of 100,000 genomes archived, and the numbers are swelling all the time. Going a little deeper, you are speaking about one of THE major issues of modern medical genomics. This will get better as we accrue more data, but we are at a difficult time in which we just realized, as a community, that humans carry A LOT of rare genetic variation, most of which is benign. We are also beginning to realize that the effect of mutations can vary from person to person, so that one child with mutation "A" can develop a debilitating disorder, whereas another child might not. This is somewhat kin to having a full body scan; you are bound to find something "suspicious" that may turn out to be a normal (albeit rare) anatomical variant. However, once you have a database of 1 billion such scans, or more, then you get better a diagnosing. At the end of the day, it's all about risk-benefit. We are seeing cancer deaths drop in many cases because we have the ability to diagnose early from scans, but some tests are better than others. Same with genetic disorders. Some variants we understand well and we can predict what will happen with a reasonable degree of accuracy; these save lives. The rest of the time, we are left scratching our heads. But as i said earlier, things will only get better and better and better

Can you comment on the current situation on patenting of genetic information? And whether you think that this will hold back our abilities to use gene therapies? Do you think there need to be changes to patent laws where genetics are concerned?

I've read of companies trying to patent eg. Breast cancer genes. I don't really understand how that can work, since genes are naturally occurring, but at the same time if there are therapies out there that researchers come up with I can see that they or their employers might wish to patent and then profit from this.

BleachBody

i think that we need to take the long view on this. Humans have been around for 1000s of years; patents have a shelf life of 17 years. In other words: this too shall pass (and for the record, i see no value in patenting genes, to me it's like patenting the moon, fire or the shape of clouds)

Hi Dr. Katsanis, Thank you for doing this. A few questions:

1) How will all these pediatric diseases be treated if the pharmaceutical industry isn't interested in developing drugs for them? It seems like a classic market failure.

2) Why do you think the National Institutes of Health are so risk averse? Seriously, they only seem to fund work that leads to incremental advances.

3) Has Duke University implemented ANY additional requirements of their scientists to ensure that fraud like the infamous Potti case..(Info here for those who are not familiar. http://www.cbsnews.com/news/deception-at-duke-fraud-in-cancer-care/)

RobotsonRockets

You have hit the nail on its head. Market failure indeed. Well, at the risk of sounding self-serving, we are working on ways to overcome this problem, at least in part. We are working on a platform-based solution that will use essentially the same technology for a swath of genetic disorders, thus cutting down the up-front cost significantly. This, coupled with recent regulatory changes that will help with clinical trials for rare disorders, and i am actually optimistic! Now, for question #2, the NIH. I have noooo idea. Some might say it's because they need to report to congress and show progress. I mean, when you life is defined by 5-year periods of re-election, would YOU take the long view and be patient? At the end of the day it does not matter. The NIH is an imperfect system for imperfect humans and we have all adapted to deal with it. I do not think it's the main issue with academic science; i think that the US university financial model is deeply flawed...but that is another conversation

3: they have...more committees. Again, i need to be frank here: we were faced with a dishonest human who acted immorally. If we had to legislate an additional committee every time we met one of those, the entire planet would be nothing but committees. Also, just to be crystal clear, if a scientists wants to cheat and fake data, they will be able to do so, no matter what the regulatory environment. At the end of the day, independent replication is the alpha and the omega of the discipline and will always be the ultimate judge. I would argue that the major failure with the Potti thing was the tardy response to the concern raised by our peers, not the fraudulent act itself.

Have you come across any instances where the genes you thought were at fault were active but weren't causing the the disease for some unknown reason? What is the oddest finding you've had so far?

What's it like being a human geneticist at duke? Are the unicorn genetists getting on your nerves by poking holes in things with their horns?

hugemuffin

The oddest thing was an instance in which a mutation that is known to cause a genetic disease was actually exerting a protective effect for another disorder. Reminded me that nothing is as black and white as we sometimes think (or wish to be true). Re being a human geneticist @Duke. I love it, but it's very hard work. Choosing between work versus family time dilemmas every day. the unicorns are for the most part behaving.

Hi Nico! You sound like you have an interesting job! How do you feel about services offering personal genetic testing for ancestry and health arranged to anyone who wants it done? Could it be dangerous to give the average person their genetic information knowing they probably don't understand it well? Do you think the future of personalized healthcare lies with genetic testing?

I had my genetic testing done and found I have a few mutations which are known cause some symptoms/conditions I've been dealing with for years. I took my results to a few doctor and they all laughed at me saying we don't know enough about genetics to make those results matter. I've mostly been treating my symptoms with supplements from forum advice of other sufferers of these gene mutations and am feeling better. I was just curious if my doctors have a point and what I'm doing is mostly placebo effect or if this is all just so "new" the medical community hasn't really caught up yet.

gfjq23

Yes, i love what i do, it's a blend of satisfying intellectual curiosity wth trying to do the right thing (sorry if it sounds sappy). So, the whole genetics testing thing is, i feel, blown way out of proportion. For sure we have a moral and professional obligation to educate the population about the data, their dangers etc, but at the end of the day, I believe that freedom of choice and freedom of information trumps everything. Reasonable people will do reasonable things with the data, and they will be able to find the way to improve their understanding and interpretation. I work with families who have very young children affected by pretty serious, often life-threatening, genetic disorders. I can tell you that these folks are super-well read, super motivated and quite activist about having their information spread around the four corners of the globe. Others might feel differently, of course, and we must respect that. At the end of the day, data are data. As long as the accuracy of the information is high, true value will be found. At least that's how i choose to see the future, rose-tinted glasses and all...

What would a fantasy model be for preventing genetic disorders? Turning genes in parents off/on before conception? Or is it more realistic to develop therapeutics for disorders?

Pretentious_Cthulhu

therapeutics, every time. The reason is simple: it is dangerous to "predict" disease before it happens. Genetic disorders, some exceptions notwithstanding, are also highly individual and messing with the genome presymptomatically could be highly problematic, unless you had the means of ensuring that your prognostic power was 99.999%

Nicholas, thanks you for taking the time to speak with us. I want to know if you think the future of treatments for genetic diseases will continue looking for effective ways of modify genes or if you think we should focus on correcting the damaged proteins that are created by a damaged gene.

Vertex seems to have demonstrated that protein correction may be the future of medicine with their new CF treatments. While I am very happy that they may have found a different approach to treating genetic disorders, I feel that the model of only correcting the protein creates a lifelong patient requiring one of the world's most expensive drugs.

What are your thoughts on both the science and ethics of the two approaches?

cogitofire

Not to put too fine a point on it, but Vertex has als found a way to charge 300k per year for their drug. So, whatever happens, not only do we have to make better meds, we also have to make them cheaper, otherwise, what's the point??? To the heart of your question, though, i think that we need to be targeting processes, not necessarily the mutated genes. We have a few exciting leads in our own work wherein finding ways to restore balance/homeostasis by targeting a distal target seems to be having a beneficial effect. I suspect that this will be helpful for a whole host of disorders. This is not to negate, of course, the idea that for some genes/proteins, the mutated product is the best target.

Hi, from your point of view, how could the understanding or use of genes contribute to changing mental illnesses? By this i mean, after someone has had behaviour that harms or is at risk of harming themselves or others, and thus fitting the definition of a mental illness, how could understanding the role of genes lead to or benefit a therapy, cure, or solution?

motivationascending

Oh, i view a major role of genetics in this. Firs, i think that we will eventually be able to "bin" schizophrenia, autism, other neuropsychiatric traits, into functional modules based, in part, on genetic/genomic variation. From there, we might be able to gleam what treatment paradigms might suit best. I also think that we will have an outstanding opportunity to identify co-morbidities that will have direct clinical utility.

Do you think that cannabis will be a viable treatment for people with certain autoimmune disorders? Dr.

William Courtney's work suggests that cannabinoids have immunomodulating effects that can treat autoimmune issues.

Fractoman

I thik that these issues have become politicised and have been lightning rods where data have been cherry-picked to support a previous political viewpoint. I am not personally aware of the studies, but i am a bg fun of cold hard data and independent replication, followed by what happens happens. Unfortunately, I was reading somewhere that the vast majority of decisions, especially at the level of governance, are not taken based on fact, but on whim and emotion. No wonder...

We're starting to hear more about the ways in which trauma can alter a person in the generic level, and that the resulting alteration can be passed down hereditarily. How does this work? What are the effects? And lastly, if a person's genetics can change due to trauma, is there a chance we change our DNA at other times and for other reasons as well (eg pregnancy)?

book_smrt

I am not sure what trauma you are referring to. Exposure to carcinogens and others toxic chemicals can certainly change the composition of DNA, which is why they predispose to cancer, but beyond that, i do not know of other instances. Lysenco tried to get animals to "inherit" injuries. Didn;t work out so well fo rhim...

HI Nico, How far are we from realistically being able to offer a cure for people with inborn genetic disorders like muscular dystrophy, Li–Fraumeni syndrome or any other of these sort.

Also, we have seen BRCA come up in a big way for breast cancer. However, the treatment is mainly a prophylactic mastectomy. When can we expect to be able to actually alter these genes to reduce the risk?

sumvell

I think the issues that are preventing us are primarily a) financial; and b) regulatory. Intellectually, i see no major technological boundaries that cannot be overcome. then again, i'm an eternal optimist...

How much of a difference has the human genome project made to your research? If we were able to secure funding for a larger project how do you think it would best be spent?

drukath

The difference has been huge, simply because we have a pretty decent "parts list". I think the next frontier is to merge 1000s of genomes with phenotypes. I know Google and others are trying to do that. Finally, just betraying my own biases here, i think that we need to put together a MAJOR effort to functionally annotate genetic variants, maybe start with the rare ones that are more likely to be causally associated with disorders. I have the sense that we have a widening gap between data generation and our ability to interpret

How much truth is there is the statement that much of the scientific community are opposed to gene editing? Why do you think this is?

SerTinfoil

Editing in humans, yes, that is true, it is generally opposed (including by yours truly, at least for now). We just do not know enough about potential side effects or other detrimental issues. For example, there are whispers that some of our CRISPR mouse models are becoming sterile. May be an "urban legend" we just need more data. My general view is this: if we wait a little bit, we lose the possibility of helping people for a number of years. However, if we rush and screw it up, we push the field back decades and harm more people that way; look at gene therapy. In the late 1990s, it was all the buzz. Then several accidents and unfortunate events in the early 2000s and the field lost a decade. I just want us to be careful, thoughtful and methodical, not driven by the dual monsters of greed and desperation.

I recently learned about Androgen Insensitivity, and other sex chromosome disorders, that essentially make you develop as the opposite sex that you are. I'm curious if there are other, more subtle sex chromosome disorders? Do you think that gender identity disorder could be linked to a less detectable chromosome anomaly?

Prosth

I do not know but does it matter? I do not view gender identity as a disorder.

What is your most interesting finding so far?

Lysca

I think i responded earlier to this, but worth mentioning again: the discovery of pathogenic mutations that actually exerted a protective effect on other pathogenic mutations; opened my eyes to a possible path to therapies

Thanks for your time in doing this AMA. Two questions 1) I have early stage COPD and have seen ads touting Stem cell therapy. I have heard this is dodgy at best, but would it be worth a try at this point, or should I wait until desperation makes it necessary to try unvalidated medicine? 2) Mental disorders, genetic in nature, environmental, or experience based? In your opinon.

Lightningrules

I have heard the same things that you have re: stem cells for COPD. I am not an expert on this but i am deeply skeptical. I would find a professional that i would trust and discuss options, preferably an academician. Regarding your second question, i think all your statements are true, we just do not know the % contributions of this blend (which is is part why these disorders have been sooo difficult to study)

Hey Dr. Nico thank you for doing this AMA!

Heterozygous masking is beneficial to an individual in certain cases, i/e sickle cell's resistance to malaria. What are some lesser known examples of this? And do they have any known historical impact on our evolution?

Conquerofcephalopods

thank you for your interesting question. there are precious few of those, but we are beginning to discover them now. For example, our team, in collaboration with a group from Lausanne, reported a few months ago a mutation that in homozygosity causes a severe disorder but in heterozygosity looks to be protective of the 16p11.2 deletion associated wth autism. I bet you that there are scores more waiting to be found! No idea what the evolutionary landscape will look like though...

What's your issue with unicorns?

But seriously. I've had something bothering me for a while. I suffer with depression and anxiety, as did my mother. She actually had serious adverse side effects from medicating it. I'm scared that I'll have the same genetic issue that caused that. I mean, I'm half her. What should I do? Can they tell before medicating?

BloodBride

I have nothing against unicorns as long as we don't try to chase them to isolate all-healing elixir from their horns! But seriously, I am glad that you are thinking about this, since a lot of people are hesitant about confronting potential psychiatric issues. At this stage, genetics will not help you, we do not have definitive "a-ha" smoking guns However, a positive family history is important and potentially informative. I would strongly encourage you to talk to a doc about this, if you have not already done so.

What are your thoughts on those who blame their obesity on genetics?

BigIrishBalls

Well, it is absolutely true that some mutations cause severe obesity and that the person cannot do anything about it. I recall a book by a patient with Prader-Willi syndrome on the subject, describing himself as a prisoner in his own body in that regard. Having said that, that accounts for a minute fraction of the obese population. As a 200+ pound person who struggles with nightly trips to the fridge i can tell you this: my genes did not make me spoon the Nutella.

How do you think gene editing in model organisms, like zebra fish, has changed the path from gene discovery to bedside therapeutic? What new technologies for functional studies are you most excited about?

p1percub

In the research arena, gene editing is just another tool that has allowed us to scale our experiments (which is awesome) and also attack questions that we did not think were experimentally tractable. I am also excited about our ability (finally) to speed up our imaging. Even last year we would spend days and days imaging a clutch of embryos, we can now do as many as 500 in an hour. All sorts of new possibilities there!

Father of an amazing little girl with turners syndrome. no questions here, just wanted to thank you for your work.

MrNobles903

I am deeply humbled by your comment and i thank you for offering a continued reminder of why i do what i do.

How can we discover more about people with rare conditions better now that we have the Internet? My daughter has Cri-du-Chat syndrome (5p-), and while it's rare, we're now connected to many people across the world. Not much academic research is done on such conditions due to lack of funding, but might there be a way to improve outcomes by learning from a larger sample accessible via modern communications?

Floppy76

ab-so-lu-tely. About 2 years ago we were working with a family and diagnosed them with a mutation in a ultra-rare disorder. Through social media, the internet and whatnot, they quickly formed a support group. Through these conversations, not only did they find comfort and support, but they also started to piece together additional aspects of the clinical picture that had remained elusive.

Rare-diseases pose a large amount of genetic diseases (e.g. 3.5 million in Germany) and are often a blow of fate for the ones affected and their families. unfortunately there is low to non medication and funding available for this patient.

How do you assess the importance of a pan-European or a global association for research into this diseases and how probable would you rate the success of such associations? What are the difficulties in research with respect to the rare diseases?

Rash-Kah

I think aggregation of resources is critical. I think that patient registries are the first thing that needs to be done, so that the researchers can have access to motivated patients with good clinical data. Progression and disease endpoints are critical for designing clinical trials, while patient samples are an essential fuel for drug discovery

What is the most common misconception regarding your field that you would like to see be less common?

Guyuter

Oh, tough to pinpoint one, there are so many stereotypes. I think the biggest issue is one that some scientists are viewed as "knowledge at all costs" will less consideration for societal outcomes. My goodness, i cannot tell you how often i come across this and how far off the mark it actually is.

Precision medicine is going to rely heavily on patient genomic information. Given the state of disarray with electronic health records in the US, both with roll out and adoption, what steps do you think are needed to avoid a similar fate for precision medicine. Are we doing enough in your opinion to prepare our current and future physicians for this new medical model of care? Thanks for taking the time for this AMA!

DiscursiveMind

Thank you for your astute observation. I think that it's a shambles and that several european nations (Denmark, Sweden, Iceland for eg) are miles and miles ahead with regard to both the harmonization of EMRs and access to them. Regarding the latter part of your question, i know that several medical schools are working hard to revamp their carricula to respond to the changes, but we are in the midst of disruptive technologies and it will take at least a generation, in my assessment, for the rest of the educational and infrastructural environment to reach equilibrium

High school student here. How common are jobs as a geneticist? Is the job market good?

ladyroxannaz

Not the right question IMHO. If you have a passion, you will excel irrespective of market conditions. When i started, i had no idea what the market was, i just knew that human genetics was the coolest thing EVER!!!!!!!!

What's the number one genetic disorder that is your most favorite to research and why?

dont_judge_me_monkey

the ciliopathies. I started in the late 1990s chasing the genes for those and I have learned so much from these disorders, both as a scientist and as a human being. Now, 15 years later, some 100 genes identified, several pathways dissected and with excellent leads for clinical trials around the corner...wow!

What are the prospects for "fixing" genetic disorders in people who already have the conditions? I'm thinking of cystic fibrosis, in particular, but I would guess the prospects would be similar in many other cases?

brindlethorpe

I would say the odds are better than what they have been in a long time. We are finally beginning to see some cracks on the regulatory monolith and new technologies are likewise helping. I am genuinely excited about the future!!!

Hi Dr. Katsanis, Do you think it is realistic that someone could test during pregnancy to discover a disease like Down Syndrome in a fetus, and also have time to treat it before the disease phenotype develops? Do you think there will be a accurate genetic test for Autism in the future, considering how many genes can cause autism?

Goblue95

I think prenatal testing for DS is a full reality but prenatal treatment would be a tough thing, since it's really a collection of structural and progressive problems, that are also highly variable between individuals. I also think that there will be a subset of autism "genes" that will have high prognostic value but these will represent a small fraction of the whole, maybe 10% of AS patients?

Hello Nico! Thank you for doing this AMA. I am the mother of a toddler with Down syndrome but have no expertise aside from experience. I know recently the extra 21st chromosome was successfully turned off in a Petri dish using the XIST gene. What do you see as the implications of this experiment, both positive and negative? Do you see this developing into a actual viable treatment for Down syndrome? As a mother this is terrifying to me, so I would love to hear about how this could be used in more positive ways as opposed to just a dream to eradicate something I don't see as a problem. Thanks!

Tropicwhimper

I am quite aware of this study. Like everything else, it's about choice. I do see this as a potential therapy for DS. I actually did my Ph.D on DS genetics (many moons ago) and I am quite aware of and sensitive to the various views about the condition. I have to defer to my default position, which is all down to individual choice. I also do think that, in principle, the approach could be useful to deal with other aneuploidies that have much harsher outcomes.

As an undergraduate doing biology, and a dream of genetics, would a taught masters in genetics be preferable over a research masters?

con3131

research, every time. The experience is invaluable. However, i'm not sure you need to be doing a Masters. Perhaps you should find a job in a lab for 1-2 years, see how you like it, and then, if still interested, apply for a Ph.D. program

My mom just had her DNA analyzed for an ancestry report and for a medical predisposition report.

Have any suggestions or things to keep in mind?

Thanks.

Also, what companies would YOU choose to have these tests done?

anonymous_being

I cannot really go specific here, just to say that all these things are, at the moment, more fun that medicine. A few exceptions aside, they offer probabilities that do not impact our day to day life....all these tests do for me, at the moment, is keep reminding me the wonder of the genome, the complexity of it all and the true stunning diversity of humans.

What's the scariest thing about genetics!?

falconclutch

that you constantly fret about whether you got it wrong.

What can you tell me about rhett syndrome? I have heard tell that a lot of progress has been made, but how can such a syndrome be cured?

Livelyandweird

enormous progress, but no cures yet. there's LOTS of promise though. The trouble is that we do not know what the optimal intervention window might be; the guesswork say early years, beyond age 3 or 4, the nervous system wiring might set and difficult to intervene...

This may be controversial, but what do you think of scientists such as Bruce Lahn and their claims?

NeatAndProper

The data look solid. Whether the interpretation will stand the test of time remains to be seen, but i do not see anything overtly offensive in the findings, in that there is no evidence that brain size correlates with intelligence, no? That seems to be at the heart of the matter (I must admit i have a cursory grasp of that conversation)

What do you have against Unicorns? Seriously, is the use of the word Unicorn in the title supposed to be symbolic for myths?

On the other hand, How far are we from artificial Unicorns? Is it even possible? I am getting a vague notion of a rhino type horn on a horse as being the most realistic scenario. What's your opinion ?

TheWizardOfTomorrow

myths, particularly the one that fables their horn to cure everything. Too many quacks promising cures and preying on the desperation of parents. Closest we will et to is a fish, i think, that has an actual horn. Pretty sure NO medicinal properties though...

How do you feel about eugenics?

To me it sounds like the cure for civilization...

LordHy

eugenics was tried and failed a number of times. So, as a scientist i refer to Einstein whose definition of schizophrenia was to try the same thing again and again and expect a different result.

What do you think about the career of genetic counseling? How will it change in the future as personalized medicine becomes more common?

shmelody

that's a tough one. I think it might go one of two ways. At first, as in the next decade or so, i imagine a swell. After that, i imagine that people will use more and more crowdsourcing platforms to interpret their data