

mutations always bad? If you're more like your mum, does that mean you've inherited more of her genes? And is there such a thing as a perfect genome?



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"It is certainly as true as the gospel that when a man sleeps with his wife or his mistress with dirty and smelly feet, if he fathers a boy, the child will have smelly and unpleasant breath. If he fathers a girl, she will have a stinky rear end."

Kat: There are lots of strange ideas about inheritance out there in the world. That one is from the [Distaff Gospels](https://books.google.co.uk/books/about/The_Distaff_Gospels.html?id=mDX2_0Qb03EC&redir_esc=y) (https://books.google.co.uk/books/about/The_Distaff_Gospels.html?id=mDX2_0Qb03EC&redir_esc=y) – a collection of medieval old wives' tales. Pregnant women were also advised to avoid eating hares in case they caused their baby to be born with a cleft palate and to skip snacking on fish heads in case their child ended up with a trout pout.

Fortunately, our understanding of inheritance has moved on a bit since then. Genetics and genomics are playing ever more important roles in modern healthcare. Genetic tests - and increasingly, more detailed genomic analysis - are providing an unprecedented amount of information about the underlying genetic variations and alterations that affect health.

Today's DNA tests can reveal a genetic diagnosis for an unknown developmental condition, identify family members with an inherited genetic risk factor for a certain type of cancer so they can choose whether to take up preventive measures such as surgery or screening. They can inform decisions about the best treatment for certain diseases, even help couples figure out their options for having a family, and much more too.

The pace at which genomic data and technologies are coming into the clinic is impressive - but at the same time, it can leave all of us - patients, the public and healthcare providers, not to mention genetics podcasters - feeling a little bit overwhelmed with all this new information and trying to figure out what it all means.

To help address the confusion, the Genomics Education Programme is running a week of action from the 16th to the 20th March, designed to raise awareness about the impact of

genomics in healthcare and what we can all do to tackle some of the myths and misconceptions that are out there.

You can get in on the action by following [@genomicsedu](https://twitter.com/genomicsedu) (<https://twitter.com/genomicsedu>) and [#GenomicsConversation](https://twitter.com/search?q=%23Genomicsconversation&src=typed_query) (https://twitter.com/search?q=%23Genomicsconversation&src=typed_query) on Twitter or head over to genomicseducation.hee.nhs.uk (<http://bit.ly/GeneticsUnzippedGenomicsConversationPodcast>)

So, harelips and trout pouts aside, let's find out some of the misconceptions that people believe about genetics in the 21st century...

- Mutations are always bad, right?
- There's nothing that runs in our family – clearly, I've got a perfect genome!
- What does it mean if I have the bowel cancer gene – am I definitely going to get cancer?
- There is a history of inherited breast cancer in the family, but I won't get it because I'm male.
- We're a heart disease family, so I don't need to worry about my cancer risk.
- If it's a 50:50 chance of inheriting a faulty gene and I have two kids, one of them will get it and one won't – that's how maths works.
- I've got a genetic condition – does this mean something bad happened back in our family tree or my parents did something wrong?
- I worry that because I have a genetic condition, my kids will definitely get it too.
- If I'm more like my dad that means I must have inherited more of his genes.
- It must be a really strong gene because everyone in our family seems to have it.
- Nobody in our generation has the condition that my dad and granddad do – that means we've bred it out so there's no chance my kids will have it.
- Once we know everything about our DNA, we'll have all the answers to illness and health.
- Your genes control everything about you – if it's in your genes, you can't do anything about it.
- One gene determines one characteristic, like eye colour or skin colour.

- Like Mendel found with his peas, there are only two versions of a gene: dominant and recessive.
- Changes in your DNA are rare. If you've got mutations in any of your genes it will be obvious because you'll look different or be ill.
- If we have a genetic test for one of our kids, then we'll know what they've got and we'll be able to treat it

Everyone wants to be a Superhero – Laura Boyes

Kat: Someone who's heard almost all of those myths and misconceptions at some point is Laura Boyes, Consultant Genetic Counsellor for the West Midlands. (<https://bwc.nhs.uk/clinical-genetics-staff/>) But where do they come from in the first place?

Laura: I guess people come to genetics with lots of different theories about inheritance and particularly about genetic conditions in their family. It feels like we're predisposed to make up theories of inheritance from a really early stage in life. Even when a baby is born, we talk about who they look like, who they take after.

Kat: Ooh, you've got your dad's nose.

Laura: Yes, absolutely. And that continues throughout life, doesn't it? Thinking about which side of the family we're more like, which of our parents we are more like. In my experience, people tend to apply that within a situation where they're thinking about inherited conditions or genetic conditions.

So, some of the things we hear really often is, "I take more after one side of the family than I do the other side of the family. I'm really like my father. Therefore I think I'm going to have this gene because he had this gene." That feels like a really human and natural kind of response to make.

Often it's in response to a 50:50 chance of inheriting an altered gene, but people can't always manage to make sense of the fact that they can seem to be more taking after one side of the family but they've still got 50 percent of their genes from the other side of the family as well. We're 50 percent our mother and 50 percent our father every time. People can't always equate that to what they're actually seeing played out in their family.

So sometimes that can present us with a bit of a challenge, because it means people come with a preconception about what their genetic test result might show and what their chance really is of inheriting an altered gene.

And whilst that's sometimes not particularly harmful - we don't necessarily need to be like computers, we don't necessarily need to process chance in the way that a computer would process chance - actually, if it means that we're only considering one possible outcome from a genetic test, then it starts to become a little bit more of a difficulty for that person.

I guess as genetic counsellors, part of the role that we're doing is to try and help people to navigate and process the idea of being at risk of having a particular genetic variant or genetic alteration.

Kat: And this is the principle of how casinos work. Humans are terrible gamblers; we have a really misplaced perception about risk. You say, "I'm feeling lucky, I've had a streak of four blacks on the roulette table so it's got to be red next."

So I guess if you are telling this story deep down to yourself - that you've got more of your dad's family side of genes, therefore you can't have a condition that affects your mum's family - maybe deep down that's going to be something really baked into you and it might be hard to hear the answer that says well, actually, you have, when you really have this hope that you didn't.

Laura: Yes. And sometimes I think it can raise other issues that have happened throughout life for people. So, if they have associated their relationship with one parent or the other as being more difficult, then they find that they've inherited the gene from that particular parent, that can make it more difficult to adjust to the fact that they have this altered gene.

Kat: One of the things that my mum really likes to do is look back into our family history. Do you ever get people coming in who have actually done part of the work for you, have gone back and traced their family trees?

Laura: Yes. People are generally really helpful about tracking back family history and we ask for quite a lot of it in genetics. We often ask for people to go back three generations, to their parents and their grandparents.

Sometimes you'll get people who are really avid historians and they'll come in having traced their family back to the 1600's in advance of your appointment, with big elaborate family trees

that they've drawn out themselves on both sides, and found out really interesting things about their history. All in preparation for helping to understand the genetic condition in the family.

Kat: Wow, that is dedication.

Laura: Absolutely!

Kat: What are some of the other types of misconceptions? Are there broad themes in the misunderstandings and myths that people bring to you?

Laura: Yes. Aside from those that draw on taking after one side or taking after the other side, people also sometimes carry with them - and this might be quite subconscious - ideas that carrying a gene alteration is something that is a response to something bad that has happened, or something that they've done wrong, or that it carries a sort of stigma.

One that we really commonly hear in families where there's a recessive condition like cystic fibrosis, is grandparents or great-grandparents saying, "That can't possibly have come from our side of the family because there isn't any history of that in our side of the family. It can't be us." I think that does two things.

First of all, it overlooks the fact that in recessive conditions you often don't see any history in the family. Because it takes two people that are carriers to have a child together for a child to be born with cystic fibrosis. That can happen to anybody at any time, even if there's no history.

Also, it gives a kind of judgement, if you like, for somebody being a carrier for cystic fibrosis, when in actual fact we've all got alterations in our gene. They give us differences; they make us unique. Some of them give us good attributes and some of them are more difficult and challenging to deal with. It's completely normal, we've all got some alterations in our genes and it feels quite important that we recognise that and we help people to understand that.

Because we can judge ourselves very harshly, particularly when people become parents. They judge their own parenting harshly; they take on a lot of responsibility for the things that their children have to deal with. If their child has a genetic condition that particularly shows, they can really feel like it's their fault that they are having to deal with all of this.

For some people, knowing that it's a genetic cause for that particular condition can help to alleviate some of that guilt. For other people it just radiates back in waves through the family. It

feels like this alteration being passed down makes something in their family a bit faulty. Really, we need to move away from that because it's normal and occurring in everybody that we have gene alterations.

Kat: I always like to say everyone is a little bit mutant! I'm aware that the word mutant is not terribly helpful, this talk of mutants and mutations.

Laura: It's often a term that we avoid because it can be heard in lots of different ways. Actually, some of it is about knowing your audience. I've had teenagers and young children particularly, when I've talked about genetics, say they've got really excited about the idea that they might be a mutant because they really like X-Men. So it suddenly means that they're a superhero.

Kat: Everyone wants to be a superhero.

Laura: Everyone wants to be a superhero!

Kat: So in your role as a genetic counsellor, encountering all these superheroes everywhere, what are some of the ways in which you try to figure out what are people's misunderstandings, the baggage that people are bringing when you're dealing with families who are looking into their genetics?

Laura: I think it's really about providing some time and space for that person to be able to really talk about what's been happening in their family. That's not something that we do in a lot of places within the health service, I don't think. We are under increasing time pressure as health professionals everywhere.

One of the real luxuries within genetics is that we get the time to be able to speak properly, to talk to our patients and our families and understand not just what the condition is that they are coming with, but also how all of their thoughts and emotions are being processed around it.

Actually, if you have a strong belief as I do, that wellbeing and health are intimately linked, it feels really important that people get that chance.

Kat: What's the impact of doing that well? What do you aim to send people away feeling and understanding?

Laura: I would be aiming to send people away knowing a little bit more, feeling that they have been understood, feeling that a lot of what they are feeling is normal and that they're not alone, so they're not so isolated with it. Hence, hopefully, feel a bit

more empowered to be able to deal with the condition and everything that comes along with it. Not just for them, but also for their family as well. In turn, that means that they then get to model some of that for their children, for their sisters, their parents, whoever comes next in the chain.

Kat: Is there one thing that you really wish that people could understand about genetics, about genomics, that would be helpful if generally everyone coming to you knew and understood this thing?

Laura: I think if there were one thing, it would be that variation is normal. That actually, we've all got many changes in our genes and they result in different attributes, but that's what makes us us. If we can accept those and accept that there are ones that are very positive as well as ones that cause us some difficulties, it would mean that we can accept genetic conditions more as a whole. That would help everybody to approach it in a slightly different light.

I don't think it's as important that people have a good understanding about a particular inheritance pattern or that they know how many genes we've got or what those genes do. Or that some of them protect us against cancer or that some of them make our heart work in the correct way.

Actually, those are things that are quite easy to teach. People don't need to know those before they come. As a society, if we could have a better acceptance that variation is completely normal, then that would be a nice place to start.

Genetics in popular culture – Anna Middleton

Kat: One common touchpoint for many people when it comes to learning about inheritance and genes is popular culture - whether that's the X-Men mutants, Sith dynasties in Star Wars, or the peculiar twist on hereditary memories in Assassin's Creed.

I asked [Anna Middleton, Head of Society and Ethics Research at the Wellcome Genome Campus in Cambridge](https://www.sanger.ac.uk/people/directory/middleton-anna) (<https://www.sanger.ac.uk/people/directory/middleton-anna>), whether these media portrayals of genetics are helpful, harmful or just... meh...

Anna: Well, after leaving school we know that most people get their genetics and genomics education, so to speak, from popular culture and the media.

Often you see there are opportunities for wonderfully imaginative discussions about conspiracy theories and things that have come from crime dramas and medical dramas. I actually don't see this as a negative thing, personally. I see this as a method to springboard a conversation when it's useful to have a conversation in the clinical setting.

I also feel, and know from our own empirical research that people understand that X-Men and mutated genes is not real, but the actual message behind that is that DNA exists and that if you make changes to it, it can alter a phenotype. That basic principle is quite useful.

Kat: So, I'm not actually going to become Spiderman if I get bitten by a spider?

Anna: No, but what the film has done is introduce the concept of DNA in a very socially acceptable way. Then it's in the lexicon and that's great. I think we should be celebrating that. I do know that scientists get very het up about the accuracy of how genetics is portrayed. Certainly in our group, we've done research about the interface between popular culture and genetics and how useful this is for clinic.

We feel very strongly that it's actually very, very helpful because it gives you a bridge to then build on. We do know that the vast majority of the public, 85% of the public in the UK, had never heard of the word "genomics" before, and why would they? But they may well have heard of X-Men, so that gives you an in, basically, for a conversation starter.

Kat: It is very sad to think that probably, very many more people have seen the X-Men films than have listened to my podcast. {Laughter}

How important do you think it is that the details are accurate? Because I know that some scientists get very wound up. You know, "The DNA helix is going the wrong way, they talk about inheritance all wrong. Grr, who was the science communicator on this film", or something like that. How important is it that the details are actually correct?

Anna: The technical details - well, of course you want the science to be as good as it can possibly be. But I wouldn't get too worried, like many scientists do, about always using the

technical terms. For example, if you use the term "pathogenic variant", it's not in common parlance. People are not going to really know what that means.

Unless you're wanting to create a power dynamic and show how expert you are, why not use the term "glitch" instead? People instantly get what that means. Why would you deliberately try and use a set of language that's going to alienate people?

So I don't feel as upset about the scientific accuracy as other scientists do. What I do feel is very important is just a general kind of engagement, awareness-raising, socialisation of all of these concepts. So that people can just engage when they're ready to engage.

Kat: And I guess as well, like you mentioned earlier, if there are wildly outrageous ideas about genetics and inheritance, surely that's a point for discussion about do you think this is true, do you think this could happen?

Anna: Absolutely. I think what we should all be aiming for is just a very broad general awareness of the discipline. So in the same way that climate change is now something that everybody has at least heard of - they may not know the scientific ins and outs, the uncertainties, the risks, the actual science behind it, but at least they've heard of it.

It feels as if genomics and genetics needs that same makeover in terms of communication methods, so that it's just part of everyday language and the lexicon. And people down the pub would feel comfortable just having a very basic conversation about the significance of that for themselves.

Kat: It's interesting you pick up on climate change, because there's some big climate disaster movies and sometimes, just looking at the news media, looking at things like the bush fires in Australia, looking at the climate change and the environmental impacts that are happening around the world, that can seem like a disaster movie.

Even from the news media, what we take to be factual and true, you hear stories about gene editing and all this scary designer babies stuff. How does that impact on the public consciousness, do you think?

Anna: Well, I think it does impact. It will feed in some level of messaging and I think that's a good thing. What's most important is that when people actually want to explore the facts, that they have really good quality resources to go to. That's really what's pivotal here.

Of course, the first thing you do when you hear about enormous climate disasters in certain parts of the world is you're Googling, "Is this due to climate change?", and you're looking to see what's online about it. You're trying to make a judgement on where you fit your opinion into this.

So for the genomics and the genetics and the healthcare space in relation to this, it's absolutely pivotal that there's a really good go-to set of resources to really help people.

It's not just about delivering the science, which is important, it's also about what does this mean for me now, and what can I do about it? And what's the relevance of this for my children? Those are the sorts of questions that people are asking.

Whose job is it to learn about genomics? Michelle Bishop

Kat: As Anna mentioned, regardless of the ideas about genetics and genomics that people have picked up along the way, it's important that they know where to get more information when they're ready - and that these resources are accurate and easy to find. That's true not just for the public, but for healthcare professionals who may also be feeling more than a bit bewildered by this fast-moving field.

Michelle Bishop is the Education Lead for the Genomics Education Programme (<https://www.genomicseducation.hee.nhs.uk/about-us/>)- part of Health Education England. I started by asking her whose job it is to get accurate information about genomics out there?

Michelle: I do think that everyone does have a responsibility to try and get as accurate a message out there, although appreciating that might not always work in some circumstances. In terms of healthcare, we know that the public will be getting their information from a wide variety of sources, whether that's popular media or family and friends, or from what they've learned at school.

Healthcare professionals I believe, do have a role to try and identify any of those misconceptions or misinformation that people have and to help navigate the conversation around that and to address that. So that individuals, when they're making decisions about a genetic or genomic test are doing so with correct information.

I also think that educators have quite an important role as well. So, thinking especially of the educators of the future workforce. This is an area where students will again come to the classroom or to the virtual classroom with their own prior knowledge or preconceptions about ideas.

Actually, the educator has a really important role, to try and identify those misconceptions and to correct that misinformation, so that in the future those healthcare professionals are not perpetuating these messages to the patients that they meet.

Kat: It is incredible thinking how much and how fast the picture is changing. I think back to the kind of stuff I learned at school, it was very much Mendel and his peas and all this kind of thing. Really, it wasn't until I was actually doing my PhD that we had large scale genome sequencing, still very, very slow.

Now you can get whole genomes done, we're talking about widespread, mass whole genome sequencing and then using that information to inform healthcare. It feels like stuff is changing very, very fast.

How can people on the frontline - health professionals, teachers, educators, nurses - start to think about how to get on top of this? It just seems like, wow, so much!

Michelle: It is so much! Not only is the information that we're understanding about genomics increasing, the use of it in popular culture is also increasing as well. So you're getting people having access to this information from a wide variety of sources, and perhaps not getting the correct information.

I think it's about having a place where people know if they're going there to get the information, it's correct information, it's up to date information and it's credible information as well.

It's also having to navigate the internet, where there's a wide variety of information out there and educational resources, not all of which have been developed with experts who have the most up to date knowledge.

So there's quite a lot that we have to navigate but I think if we can signpost these credible sources and make sure that experts are inputting where they can, then we're providing more accessible information in a timely manner, for people to be able to access when they need it.

Kat: So that's a nice segue into the work that you do at the genomics education programme. What kind of resources are you providing and who are they aimed at, who can actually get your resources to help them

Michelle: Our resources are available for anyone, but they are targeted towards healthcare professionals. Not only those that are currently working but also those who are going to be our future workforce as well.

We do work with educators as well and I think it's a really important point to make, that genomics is now being integrated into a large number of the training curricula, which means more and more healthcare professionals are required to know about genomics than they ever have before. That also means that a larger number of educators are having to teach about genomics, which they never have before.

This can be quite a daunting subject to start teaching, so we are doing a large amount of work to develop resources that are available for educators, and also to provide them with tools that they can use to identify misconceptions that might be there in their students, and also strategies to help them in actually dealing with these misconceptions and how to teach them in a way that the misinformation is being corrected.

Kat: So, what would be an example of a way of drawing out and addressing these misconceptions? Because we've talked already about some of the kinds of things that people might have myths and misconceptions. So how can health professionals find out what people think and then start working to address them?

Michelle: One avenue that say, an educator could take is to actually have a questionnaire that they've developed or adapt one that's already out there, that asks questions about some common misconceptions that are out there. You can get a baseline view about what your students or your student cohort are actually thinking. That would then allow you to target those in your teaching sessions.

One way to do that is to actually talk about the facts that are there about this particular area, then to actually address the misconception so you're actually bringing it up. You could perhaps phrase this as, "This is a common misconception that's out there in the public, so this might be something that your patients are coming to you with, this misconception."

Address it, talk about what might be wrong about the misconception but then come back to what the fact actually is as well. So, really getting people to understand the right information and the wrong information and be able to delineate between the two.

Kat: I always think it's very curious to find out why maybe someone has come across that. Maybe it was just something their mother always said or something like that.

Michelle: Yes, I think it's really interesting about the different ways that people have misconceptions. I've heard a variety of things where people have read something and they fully believe it. So the media does play a role in this as well. Although to give the media credit, I think they are calling on the experts a lot more, to be able to sense check the information that they're providing.

If we're considering what's available in popular culture then sometimes they might take creative license with what they're doing, just so it fits in with the storyline a bit better. But I know that some people do take that as actual fact, if they don't know anything else.

There's also a lot of family myths that are out there about how information is inherited in families and why some people might develop a particular genetic condition to another. So there's a whole range of ways in which people can actually gain their knowledge about genetics or genomics. It can be challenging to identify those misconceptions and those myths.

I do think it's the responsibility of the educators to do that, otherwise we are perpetuating those misconceptions. And healthcare professionals might be talking to patients about it, believing what they believe about genetics and genomics to be correct, if that hasn't been addressed in their education.

Kat: It makes sense that people like doctors and genetic counsellors, those on the front lines of genetic medicine, would hopefully be up to date on this and be able to communicate about it. But there's an awful lot of people who are intersecting with the health system - I'm thinking of nurses.



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work for a cancer charity, so those sorts of people who

are working with people who may have questions; the general

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public people with healthcare conditions, how can we try and

get more knowledge out there of where the resources are to

learn about genomics and to communicate it effectively?

Michelle: It is a big challenge and it's one that we're constantly working on. I think working in partnership with a number of different stakeholders is really important. Obviously this is our bread and butter, the business that we do every day, but we can't do it alone, by ourselves, so we need to work with different organisations.

Patient groups and charities are a really key part of that, because that's often where patients and the public are going to go first to find out the information. So we work with them, but also most of the patient organisations and charities will have an expert group that works with them to help write information.

I think the information that is written and provided on websites need to be very accessible for people, to break down the complexities of this subject in a way that is very easy for people to be able to understand, regardless of what their prior educational background is. And to make sure that we're consistent in the messaging that we're providing.

I think that's a really important thing because you don't want to have conflicting evidence from what seem to be credible sources. Otherwise it's very difficult for people to try and navigate that and to work out what is the correct information and what isn't the correct information.

Kat: So do you get wound up about dodgy DNA science in movies? Is your mother insistent that you're only good at podcasting because your distant relative was a preacher? (Or is that just me?) And what's the strangest misconception about genetics you've ever heard?

To get involved in the Genomics Education Programme's week of action you can follow them on Twitter, [@genomicsedu](https://twitter.com/genomicsedu), (<https://twitter.com/genomicsedu>) and get on the hashtag [#GenomicsConversation](https://twitter.com/search?q=%23Genomicsconversation&src=typed_query) (https://twitter.com/search?q=%23Genomicsconversation&src=typed_query) or head over to genomicseducation.hee.nhs.uk (<http://bit.ly/GeneticsUnzippedGenomicsConversationPodcast>)

That's all for now. Next time we'll be celebrating 35 years of genetic fingerprinting.

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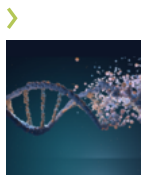
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(</blog/2020/2/27/s305-poop-pus-and-the-manhattan-project-genetic-alphabet>)

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