**How can we work in partnership towards a new era of genomic medicine and research?**

**Behind the Genes transcript**

**Helen:** Welcome to Behind the Genes.

**Rich:** There’s a whole new era I see coming in terms of the therapies that are directed at the causes of genomic conditions, both in rare conditions and in cancer, and thinking as we do that, about how we structure the system to generate evidence, and to respond to it, and have a conversation about what the right balance of evidence for patients to make a choice about their own care.

**Helen:** My name is Helen White and I am the Participant Panel Vice Chair for Cancer, at Genomics England. On today’s episode I’m joined by Dr Richard Scott, Chief Executive Officer for Genomics England. And today we’ll be discussing Richard’s recent appointment as CEO, lessons learnt from the last ten years in the evolution of genomics in healthcare, and how these learnings will be taken forward in the next ten years. And we’ll also visit the importance of keeping participant and patient benefit at the heart of research, as well as the ethical and safe storage of patient data. If you enjoy today’s episode we would love your support: please like, share and rate us on wherever you listen to your podcast.

Before we dive into the interview with Rich, I wanted to take a moment to share my story and tell you a little bit about myself. I have been a member of the Participant Panel at Genomics England since 2018. It was the year before that when I was diagnosed with endometrial, or womb cancer, and was offered the chance to join the 100,000 Genomes Project, which felt like something positive at what was otherwise quite a scary time. It turns out that I have something called Lynch syndrome, that’s a genetic condition that increases my chance of developing certain cancers, particularly womb and bowel cancer, which is actually a really useful thing to know as there are things I can do to reduce my chance of getting cancer; things like having regular colonoscopies and taking daily aspirin. I have now been on the participant panel for six years and one year ago I was appointed as Vice Chair for cancer. This is a new and developing role and I am excited to have so far helped recruit more people with lived experience of cancer to the panel and to be assisting Genomics England with connecting to organisations that advocate for people whose lives have been touched by cancer.

So that’s enough about me. I am delighted to be joined today by Richard Scott, and I am very much looking forward to our conversation. Welcome, Rich.

Thank you. So Rich, you’ve recently been appointed CEO of Genomics England. Can you tell me a bit about your background and what brought you to this role?

**Rich:** It’s a really good question and it’s one that doesn’t have a really very simple answer. I guess what it boils down to is I guess I’ve always had an interest, even as a child, for whatever reason, in genetics and genomics. I have also then always been drawn to things where I can have an impact and particularly the impact in healthcare and that’s what took me to being a medical student. And I guess it’s that combination of that particular interest in genetics and being able to see, even when I was at medical school I qualified in 2000 that this was an area of medicine that was going to be really important in the future. And then as I trained, as I did a PhD and as I saw the technology develop and change and then when I saw the UK government and the NHS investing in genomics in a really foresighted way, I found myself eight or nine years sitting at Great Ormond Street as a consultant in clinical genetics where I still practice, I still do one clinic a month there as a clinical genetics consultant seeing families with rare conditions.

But I could see when Genomics England was established that this was something, as I said, really foresightful where we could really collectively across the country make more of a difference together in terms of patient and healthcare outcomes. So I joined GEL eight or nine years ago initially in a subject matter expert role, and really found myself the more time it passed, understanding how working in my role at GEL and helping GEL be a really productive part of what is a busy genomics healthcare ecosystem in the UK, we can make a big difference, and that’s the thing that just wakes me up in the morning, is realising how much there is left to do, being proud of the stuff we’ve done, the difference we’ve made to participants in our programmes already, but realising that many of those still need our support to do better and the big distance left to go before we really deliver on I think the long-term promise of genomics, and I feel my mixture of skills and experience make me really excited to be in the middle of that.

**Helen:** Thank you. Yes, it sounds like you’ve brought many skills and experience, and interesting to hear that as a child you already had that interest in genetics and where that’s taken you. Can you tell me what being CEO Genomics England means for you? What are your aspirations for your first year in this position?

**Rich:** Well, I guess, as you can tell, I’m really excited to take on this role. As I said, as a doctor I’m always focused on the impact for patients and our participants and ultimately it’s the broader health of the nation. And the role I see Genomics England playing and being able to play in the future, sort of building on that, the leadership position the UK’s always had in genomics – you know if you look back to the discovery of the structure of DNA, the invention of sequencing technologies and also the clinical implementation coming from that government investment and the NHS investment, what excites me most about GEL is that we can be there, playing a critical role alongside others in that ecosystem, whether that’s in the NHS, whether it’s our participants and the patients who we’re aiming to support academia and industry, to create a whole that’s greater than the sum of the parts, and I genuinely feel that the UK remains uniquely placed to live out that potential that genomics has, engaging in the questions, not just you know, the scientific questions of: what could genomics test for? Or, how could this be implemented and is it cost-effective?

But also being able to have the nuanced conversation of what we all and our participants in the public and general, expect in terms of the care we receive or how our data is looked after, and getting that really balanced view on how we chart a path forwards where we can really see big differences being made in the future, and I think always being honest to ourselves about where we are today and that things don’t come in spotting some position a long time in the future that we want to navigate to, but also being really focused on the here and now and what is possible and what is evidenced, and what the next set of evidence or discussions or conversations in the public we need to have to help navigate ourselves there and that’s where at the moment our focus at Genomics England is both being very clear sighted on where Genomics could go, and also thinking very clearly about where we are today, and so very much at the moment for us it’s about focusing on the life service we offer to the NHS and we’re really proud to be part of a world-leading whole genome sequencing service, the first national health service in the world to be providing that in the context of cancer and rare disease, and so offering and providing our service that contributes to that.

Supporting researchers so that we can keep the flow of discoveries coming and also for example, making sure that our participants in existing programmes continue to get new answers as the science evolves. So, the last year more than 2,000 families had new findings fed back because of new knowledge that’s accumulating, keeping that flow going. And then we’ve got three big research initiatives going on at the moment where we’re really focusing on delivering around them. We’ve got a diverse data initiative where we’re really focused on making sure the research library, the National Genomic Research Library, our participants are representative of the UK population, so the discoveries that we’re supporting are relevant to everyone; our cancer initiative which is exploring the use of new sequencing technology in the context of cancer, and also looking at the use of image data and other modalities of data, alongside generic data to drive new discoveries.

And then the third initiative is our newborn genomes programme, where we’re asking a big question through a research study to generate evidence to ultimately answer the question: should every baby when they’re born be offered whole genome sequencing? Most pressingly to improve and broader the range of conditions that we can look for that are severe and treatable. So, this year we’re very much focused on delivering on those promises that we’ve made to our participants and our partners and through those programmes and very much with an eye to the future thinking about what we need to change in terms of the use of underpinning technology, so that we know that we’ve got the potential to scale, to think about the broader use of genomics in years to come as evidence evolves.

**Helen:** So Rich, there have been many advances in genomics in the last ten years. What do you think are the big lessons from those last ten years, and what do you think the next ten years will look like for the genomics ecosystem, what impact will this all have on healthcare as we know it?

**Rich:** So, genomics has changed extraordinarily in the last ten years thanks to shifts both in the technology, particularly the sequencing technology but also some of the computing technology that’s there to deal with the scale of data. Ten years ago we were talking about the 100,000 genomes project and beginning the project itself, but it was still very early in the use of whole genome sequencing, that’s gone from something where the big question around the 100,000 genomes project was: can this technology be used in routine care in cancer and for rare conditions, and if so, how do we do that?

And we’ve learnt both I think about that specific question and as I mentioned, we’re enormously proud to be part of enabling the NHS whole genome sequencing clinical service, so that has entered routine care. I think along the way the biggest lesson for me is actually one about this being about partnership and about working as a team across many different organisations and with our participants, and recognising that this isn’t just about one set of questions, or it’s not just about clinical or scientific questions, it’s about joining everything up together back to that point around, so a discussion about what people expect – this is about doing stuff together and learning often quite complex lessons about practicalities is one things, for example, one of the really big lessons we learnt around the use of whole genome sequencing in cancer are just practical lessons about handling of tissue samples and the need to make sure the right fridges are available on the right corridor of a hospital, with plugs available to plug them into, through to questions around, as I say, people’s expectations around how their data is stored, which it’s used for, which again there’s really strong precedent for, and as we explored, different uses of genomic technology, we shouldn’t just take those previous answers for granted, we need to make sure we validate and check with people what their expectations are.

So I think that’s the big one for me is sort of the number of different angles with which one explores questions and the fact that this is very much about doing it together. I think just one other piece which is so easy for us here to take for granted is that doing things at national scale with national scale investment from government, from other funders and from the NHS is absolutely critical and when you look across the world, we are in an extraordinarily privileged position here in this country because of that investment and because that investment recognises the need critically to join clinical care and research in a whole, where you recognise that you’re doing multiple things at once, but joining them up rather than them being two worlds, is really, really critical, and we’re really lucky to be able to do that at national scale.

So then thinking about what the next ten years might look like for the genomics ecosystem, I think lots of those things continue, so I think national scale and the need for ongoing investment to keep up our position at the forefront in terms of answering these big questions about the use of genomics in healthcare, and to where the evidence supports their implementation to roll them out and keep that link there between healthcare and research, and so making sure the systems talk to each other and I mean that in a digital sense as well as a human sense is absolutely critical.

And then, so in ten years’ time what are the areas of healthcare that will have been impacted, or could have been impacted by genomics, I’m really pleased that we’re doing a better job for families with rare conditions and people with cancer than we were ten years ago, I think there’s a long distance left to run even in those settings for us to do better and to continue to learn, so we expect our major focus to continue to be in those areas where we know they can have an impact and there’s more to do. We also then have the different areas where if the evidence pans out to support the use of genomics or if we can implement systems that can support it there can be a big sort of area of growth. For example, our newborn genomes programme is asking questions and developing evidence so that in the future policymakers can decide should that become part of routine care, and I think that’s something that could have become part of routine care in the next ten years if the evidence supports it and if that’s something that the public support.

If I were to pick one other area where there’s a real potential for growth in the coming handful of years it’s in something we refer to as pharmacogenomics. What that means is looking at your DNA code (genomics) to help make decisions about prescription of medicines and sometimes that’s about avoiding these medicines in people who are at a higher risk of having an adverse reaction, or it’s about tailoring the dose because of something about for example the way the person metabolises, chews up, the medicine and so can influence how much dose they need. That actually has an enormous potential; we all have variations in our DNA code that influence how we respond to or metabolise medicines. If you look across primary care, GPs and so forth, primary care physicians and in secondary care, hospital care, I think there’s good evidence that actually probably half of all appointments, interactions in those settings, if you were to have DNA data available that could influence how prescription choices are made; sometimes that’s about knowing that you’re doing the right thing, giving the normal prescription, but sometimes it's about modifying it, that’s an area where I think there’s a real potential for growth and that’s an area that the NHS also really recognise and we’re exploring ways in which we might look into that and think about how that might be implemented, because actually a lot of the questions there are about how you make sure the right data, the right information is available to clinical teams and patients at the time that prescriptions are being made.

There’s also real potential more broadly in thinking about more common disease settings, there’s lots of work going on from various research studies looking at the value of what people sometimes refer to as polygenic risk scores or integrated risk scores, where we use genomics as an element of estimating risk for common diseases like heart disease or cancer, that’s something where the evidence is being worked on and is developing, I think we’ll see a lot of evidence come out in the coming years and I think that will then influence how we implement genomics to help as part of that risk estimation process, which is routine now in GP practices where you go for an NHS health-check they do it with lots of complicated stuff, at the moment not genomics, and we’ll see how that plays out in the years to come.

So I think there’s enormous room for growth where genomics where at the moment it’s making an important difference to people with certain conditions that we can do better on. In the future I see it becoming very much more part of the routine day to day of healthcare. As we make that transition there’s lots to work through about the evidence, the order in which that’s done and the way in which we, for example, store data, and make people part of the choice about how their data is used and what I’m really excited about in Genomics England is the role we play in the middle of that, bringing our particular expertise around what we call bioinformatics, which is sort of managing genomic data at big scale, particularly national scale to support healthcare and research, generating evidence that can help inform policy, and also critically drawing things together into the conversation amongst different players in the ecosystem and participants in the public so that we can not just think about evidence in a sort of terribly scientific way but we think about it in the round.

**Helen:** That’s really interesting to hear you speak a lot about getting that evidence because that’s critical, but that takes a long time doesn’t it, so for example with the generation study, the newborn study it’s really important to measure the benefits of that if you’re testing young babies, newborn babies for diseases that if you pick up a condition that condition can be treated and something can be done about it early rather than poor parents going through this diagnostic odyssey, but also it’s that balance isn’t it with not leading to any harm, so if a number of parents come out of that thinking their baby might get a condition and it never happens there’s potential there isn’t there. But I think in terms of the public understanding of how long it takes to get evidence and everything else that needs to go on in the background I don’t think it’s always particularly clear that that’s a massive process that has to be gone through and there’s a lot of work going on behind the scenes – you can’t just do these things.

I think as patients/members of the public we’re eager to get on and for change to happen and things to be better but it’s a big, big process, but also good to hear that you talk about it being a collaborative approach, it’s not just Genomics England, it’s the NHS, it’s members of the public and patient voices, it’s other organisations working in partnership, it’s a big undertaking.

**Rich:** No, it is and I think that one of the words you used there was impatience, and I think that’s healthy and important to recognise, it can be easy, particularly for example as a doctor, sat in a clinic room to accept the status quo, and at the same time, one needs to recognise the complexity of the questions, the balance, the need to generate high-quality evidence to inform those opinions and I think combining both that sort of impatience and dissatisfaction with the status quo, and that mind-set about thinking really thoroughly and collaboratively about the right evidence that is needed to change policy.

**Helen:** Yes, really important that those patient voices are there from the beginning, from the planning of obtaining this evidence and that you’re measuring the things that matter most.

**Rich:** One of the areas where I think we’ve seen that play out, another area where I really see the potential for growth in the future is much more genomics-enabled treatments. We and you and the participant panel have helped us think about there’s a whole new era I see coming in terms of the therapies that are directed at the causes of genomic conditions, both in rare conditions and in cancer and thinking as we do that about how we structure the system to generate evidence and to respond to it and have a conversation about what the right balance of evidence for patients to make a choice about their own care, but also policymakers to make choices about funding, decisions and safety decisions, is really important and we’ve been supporting to a wider work in cancer in the UK called the Cancer Vaccine Launchpad, and likewise we’re part of something we call the Rare Therapies Launchpad, where in those two areas we’re exploring that, and that’s another area I think of real potential in the coming years, and also real nuance as we construct a way of navigating that together and making the most of the potential, but not just sort of rushing in and pretending we know all of the answers at the outset.

**Helen:** And those launchpads are of particular interest to participants in the wider patient population, there are a lot of people and children with rare, ultra-rare conditions who are desperate for treatments that just aren’t available right now, equally for cancer patients there’s a big need isn’t there for more effective treatments, fewer side effects, that target that person’s particular cancer, so it’s good news I think for the wider public.

It does seem that innovation and partnerships are crucial to Genomics England’s activities so how does Genomics England ensure that participant and wider patient benefit are at the heart of these activities?

**Rich:** I think one of the really important things is actually governance is sometimes a boring word, sounds like it, but I think thinking about how we’ve structured the organisation and placed you, as the participant panel, as part of our governance to make sure that when we’re thinking about for example access to data in the National Genomic Research Library, participants are sort of driving those decisions, it’s an independent committee that makes those decisions with representation from our panel. One of the things is thinking about the governance and making sure that you as our participant panel hold us to account for the decisions that we’re making, which I think is really critical.

I think then also as we’ve learnt a lot over the years, not always getting it right, about how we make sure that participants, or potential participants in the public are involved from the outset in the design of programmes because it always helps. I think certainly before I joined Genomics England I think I would have been unsure about the best ways of going about that and that brings with it sometimes a nervousness. I think the main advice I would say to people listening is to have confidence that just getting stuck in and have conversations is the way to do it. There are then also all sorts of expertise that we’ve really benefited from being to bear in terms of ways of doing that engagement work and that will come; the first thing is to have the confidence and the desire to put that at the centre of how you decide where your focus should be and how you design programmes.

**Helen:** I think Genomics England has been very successful with that by integrating that patient voice from the very early days and here we are what eight years on I think now, and yes, hopefully we’ll be there for some time to come yet, as long as Genomics England exists.

So Rich, with more and more health data being stored, how do we ensure that this sensitive personal data is stored and used safely and ethically across the genomics ecosystem. And actually while we’re on this question, can you just explain what genomics ecosystem means, because we use that term I think quite a lot, but I think it’s not necessarily understandable to the wider public?

**Rich:** What I mean when I talk about it is I mean the mixture of different people, whether that’s sometimes organisations, us, Genomics England, the NHS, the NIHR, National Institute for Health Research; industry partners whether they’re people who are from pharma companies or from biotech, academic researchers, participants in programmes – everyone who comes together to work on genomics in the UK and a bit like the word as it’s used in biology, it’s a sort of busy ecosystem with all sorts of people playing their own role and then working together, and so I think it’s a really important thing to recognise that we’re part of that and in fact it’s one of the things I love most about my role at Genomics England is thinking about all of the different partners that we need to work with and to those outside it I think it can also be a bit intimidating, because it’s hard to keep up with who on earth everyone is.

So then thinking about the question of how we make sure that data’s stored and looked after and used in the ways that people expect and safely and so forth, I think that’s absolutely at the heart of my role and our role. And I think one thing is actually always sort of starting at the: why are we doing this? What benefits are we seeking to bring to people? Is that what they expect? What have they signed up for if you like? But that’s in a research study or when they’ve decided to say yes to having a particular test, which is the same in any part of medicine. And if we use that to drive our decisions, that’s what’s so critical. And so that’s where thinking about programmes we run, and also the things that we think might be worth something that we should prioritise in the future is always first driven by the benefit that you might be bringing, weighing up the costs and the potential downsides and harm that might be caused by the use of genomic data in that way and that’s what should always drive things, and there isn’t a one-size-fits-all, you know, genomic data should be used and stored in this way and that’s one of the things that I think making sure that participants and the public are at the centre of the conversation is absolutely critical, it turns out that genomic data is very much like health data at large in many senses and it’s very precious for those reasons.

It is also special in a few ways. One of the ways that’s sort of peculiar if you like is that pretty much the DNA sequence, the genome, that you’re born with, is the same one that you hold throughout your life, that’s different from say if you do a blood count or something that varies for various reasons over your life and most things in medicine do change quite meaningfully over a much shorter time period. One of the things about the DNA code: A) it makes it more precious because it’s very much about you, your whole life; also it makes it more useful and reuseable in many ways, so one of the things that we think about a lot more in genomics is about the storage and reuse of data on an ongoing basis through the lifetime. And I do think that that model in certain settings and potentially more broadly as evidence accumulates, may well be the path that we take forward where you consider your genomic data part of your health record where it can be used and reused.

And what we need to do is explore why you would in the first case generate someone’s DNA sequence, and what sort of sequence, is it a whole genome or less than a whole genome? What would you use it for in the first place when you first generate it? And what other uses could there be to support the healthcare and have you involved them or the public more generally in decisions about how it’s used? Because we do, as I said, see the potential for genomics being just becoming part of the fabric if you like of healthcare, good healthcare, the best healthcare.

Linked to that is the point on research as well, like where people are happy for it, holding their genomic data and understanding how that impacts on longer term health outcomes, something we’ll continue to learn about for years and years. So I think the first point is about focusing on the why and whose data it is, one’s own genome belongs to you, it doesn’t belong to anyone else, what people are happy with and consent to and expect and then always holding that in mind as one makes the choices is critical. I’ve talked about how we think the governance and the involvement of the participant panel is really critical for that as well. And then it also comes down to doing in various ways, the job that people would expect in terms of, for example, that safety piece, using the very latest tooling to make sure that it’s held in a secure way, that it’s backed up so that it won’t be lost etc. and bringing sort of the right, very good minds around some of those more technical questions, but always with the expectations of the people whose genomes they are in mind and to say are we living up to their expectations, are we doing what they would expect?

So, Helen, I wondered if I could ask you a couple of questions. The first one I wanted to ask is what you’re hopeful for in the coming years as a participant panel member?

**Helen:** Thank you. I’ve actually already posed these questions to some of the other panel members, so I’ll try and make sure I include their responses here as well as mine, but I think it’s important to hear from everybody, not just me, Rebecca Middleton and Emma Walters have recorded their responses as well. I think the four main things that panel members are hopeful for is the coming years, the first is equitable access to whole genome sequencing, basically everybody who needs whole genome sequencing should get access to it regardless of where they live, their income, ethnicity or disability, so that’s something that we’re hopeful will get better over the years.

We know this is essential to improving healthcare, to improving outcomes for patients and generally for sort of greater inclusivity and in genomic research, we want as well as Genomics England, the data is the National Genomics Research Library to be representative of the population as a whole, not just the people who 1) are offered, and 2) agree to have their data in the library. And also, obviously the more data that is held in that library, the more opportunity there is for research across those rare and ultra rare conditions and rare and less common cancers, where it’s all about numbers, you need numbers of sets of data in order to draw things together and make conclusions to look for patterns.

And the other thing which I guess comes more under the umbrella of the NHS is that the panel is quite keen, they want everybody who’s undergoing genomic testing to receive good support and after care, I think regardless of whether that testing is via the NHS or as part of a research study, sometimes it will be both, but that’s for the patients at the coal face that is obviously critically important.

The second, I think broad theme, coming from the panel members’ responses is that I think you’ve mentioned this already, is increased understanding of genomics amongst the general public is really important – there’s a need to demystify genomics and to generally improve public awareness of its benefits and to get those conversations going around its regulation and its ethical use, but to do that you need to get meaningful engagement from a wide range of people, you know, that’s not always straightforward, there are lots of challenges there, it’s all about prioritising inclusivity, accessibility, to make sure you get diverse views and perspectives on genomics and on genomics research.

The other thing that came out very strongly from the responses which we have talked quite a bit about already is about this individualised healthcare. I think we as a panel are very hopeful that there will be this shift towards treatment strategies that are tailored more to the individual and their specific health condition, rather than a one-size-fits-all approach, we want effective treatments that will minimise side effects but also through the use of pharmacogenomics, to make sure if there’s a risk of a severe, sometimes life-threatening side effect that that can be identified and that individual doesn’t have that treatment either at all or has a lower dose, so it’s not so toxic.

And let’s hear from Emma who talks about this.

**Emma:** My hope is that we move to a truly individualised healthcare system and I’m really excited to see how in particular pharmacogenomics changes the healthcare landscape. For a long time we’ve gone with a one-size-fits-all approach, and that’s easy to deliver on a large scale basis that the NHS works on, but we know fundamentally that’s not how patients work, so to be able to consider individualising medication and knowing which won’t work, interests and excites me.

**Helen:** So the panel is also very hopeful about the development of those innovative therapies, and you talked about the rare therapies launchpad and the cancer vaccine launchpad, because those offer real hope for treating previously untreatable conditions and generally improving accessibility to treatments. And we’re also hopeful that there will be a much better understanding of diagnosis of cancer, through things like the multi-model programme, because although there’s lots and lots of research going on with cancer there’s still a long way to go to have more effective treatments and to improve diagnosis of cancer.

And then just finally just in response to your question, patient and public involvement, this is what the participant panel is all about, we are a group of individuals whose lives have all been touched by either a rare condition or by cancer currently, either we’ve had that condition ourselves or it’s affected our loved one, and we do bring these diverse views and perspectives to Genomics England and I think we have a crucial role in influencing its decisions about what it does with participant data and who has access to that data. It’s critically important that Genomics England listens to what matters to the people whose data it holds and who do that, as Rebecca here explains.

**Rebecca:** Genomics is a fast-moving science and it has the impact to change lives and healthcare for future generations, but genomics is a science of people and therefore the only way you can truly understand the limitations and opportunities of it is to talk eye to eye to the very people it will impact, and not everyone will agree on everything. But how we understand genomics and its power to transform healthcare, our own and that of our children and the ones we love, can only progress at the pace of the people that it will benefit. It’s a simple equation but it’s not maths and indeed not science: we are all different and unique, our emotions, experience and history will be wrapped up in our viewpoints and thoughts, and that’s where the panel comes in, representing and advocating for the very many different voices of genomic healthcare, ensures Genomics England is stronger, healthcare design is more meaningful and research is more impactful.

I have no doubt that the panel of the future will continue to be heard and understood at Genomics England, and I hope it continues to grow to reflect more diverse voices and experiences and continues to be the people inside the science.

**Helen:** Finally, the panel is also hopeful for increased public and patient involvement in genomics research, this is integral for shaping research both academic and commercial, it helps with identifying research priorities, developing new treatments, basically getting that voice of the patient in there to tell researchers what’s the most important and what matters to them.

**Rich:** So another question Helen, how do the panel feel about the changing genomics landscape?

**Helen:** A good question and I think overall it’s a balance between excitement and hope on the one hand, and a bit of apprehension and caution on the other. So the panel is really excited about the advances going on in healthcare, we’re entering an age now where we’re promised a much more proactive, as opposed to reactive approach to healthcare. You were talking earlier Rich, about having your genome sequence, and this is something that you have for life, it’s like your passport, your fingerprint, so from infancy to old age you’ve got this data which is held somewhere which holds so much promise of predicting if you might develop a disease, whether you might react badly to a drug, so ultimately it offers great potential to improve outcomes for patients, their families and the NHS. Again, we spoke earlier about this holds so much promise for producing the diagnostic odyssey that so many parents go through when the children are born with a condition that doesn’t have a diagnosis, potential to diagnose things like cancer a lot earlier where it’s more treatable and to prevent disease as well, I know that’s something Genomics England isn’t specifically looking at, but through screening programmes, using things for example like circulating DNA which may be able to pick up that there are things going on and picking things up earlier means that those things can be dealt with earlier.

I mean thinking of my own personal example, I know I have Lynch Syndrome, I know that I am at risk of developing bowel cancer now, but that means I can do something about it. So I have my colonoscopies every two years, I take aspirin every day because that reduces my chance of getting bowel cancers and I’m much more symptom-aware, so having that knowledge up front is very helpful in being able to move forward and reduce my chance of getting an advanced cancer.

The panel is also very excited about the ongoing collaborations and the novel therapies that are being developed through the rare therapies launchpad, these offer a lot of hope for treating previously untreatable conditions, and improving accessibility to treatments, and obviously more targeted treatments for cancer, you know, we’d need more effective treatments for cancer but with reduced side effects, so that in a nutshell, those are the other positive sort of things that the panel feel excited about. Where they’re slightly more apprehensive or concerned, I mean they do acknowledge that there are challenges ahead and there are big concerns about the NHS’s ability to cope with increase in demand for genomic testing and particularly worries about education and training of healthcare professionals in genomics, how do they effectively communicate research findings or results to patients if they don’t have a broad understanding of genomics?

And then finally, let’s hear from Emma.

**Emma:** I think I’m excited but cautious. I think it’s really important to acknowledge that the research being undertaken is groundbreaking and the vast majority of clinicians have very little to know genomics education, and translating these findings into tangible benefits for participants is so very important, and something I think we’ve really got to make sure we don’t lose sight of.

**Helen:** We talked earlier about awareness among the public about genomics and we do feel that there’s a need to drive education forwards, you know but this is challenging, given the rapid pace of developments that we’ve spoken about, I think even for the panel members who I would say are relative experts in genomics now it’s hard to keep up to date, so how do we do that moving forwards? We’ve talked about security of data, we understand there are moves to link more genomic data sets both nationally and internationally and that clearly has significant benefits because that brings bigger numbers of patients data together, but opens up potential risks in terms of security, so how do we make sure that the security of that data is as good as it is currently when it’s held in one pot in Genomics England Research Library.

And just a couple of final concerns that were flagged by panel members, there is some apprehension regarding potential misuse with genomic data by insurance companies; we’re given a lot of reassurance about that but there are concerns that could potentially lead to the most vulnerable in society being unable to get affordable cover if they’re found to have genomic changes that mean they are at risk of conditions or have certain conditions and there are also concerns about the ethical implications of AI in diagnosis and clinical decision making, you know, AI is obviously a fantastic thing for looking at patterns amongst a big lot of data, but how accurate is it and where does the human come in, in terms of decision making?

So those are, I think, the broad concerns from the panel. I don’t know if you have any thoughts on those, Rich?

**Rich:** I think the big thing to say is I think having the participant panel there, you said in the middle of that, become collectively quite expert and you recognise that. Having the ability to have these complex nuance conversations and have people share that and speak directly to us about it I think is the biggest thing – lots of those points there made by the panel, I think both things that we have very much in our mind about things that one needs to balance and focus on, and there are also things that we already talk about which is reassuring I think as well, we talk about with the panel. I think one of the things for us as well is sort of being clear on some of the things where there are really clearly well-established red lines, for example, that point on insurance, but that is very clear and part of our role is making sure that that is there and people can feel comfortable in that context to understand that.

I think the main thing that I would say is thank you to you Helen, and to all of the panel and all of our participants because I said earlier, this is a team thing and you are all very much part of the team and we would not be able to do our jobs in any way, I wouldn’t even say effectively, I would say with the relevance, which is the thing that we drive for, the relevance to have impact for people’s lives whose data we hold and will hold in the future. And so thank you for being part of the team.

**Helen:** Thank you. And I think thank you to Genomics England for having the foresight to create the participant panel in the first instance, it was there from the get-go and I think a really great opportunity for all of us to be involved in this, to have our voices heard and listened to, so thank you.

We’ll wrap up there. Thank you for joining me today and thank you for discussing your appointment as CEO for Genomic England, and your view on what the genomics ecosystem might look like over the next ten years. If you would like to hear more like this, please subscribe to the Behind the Genes, on your favourite podcast app. Thank you for listening. I’ve been your host, Helen White. This podcast was edited by Bill Griffin at Ventoux Digital, and produced by Naimah Callachand.