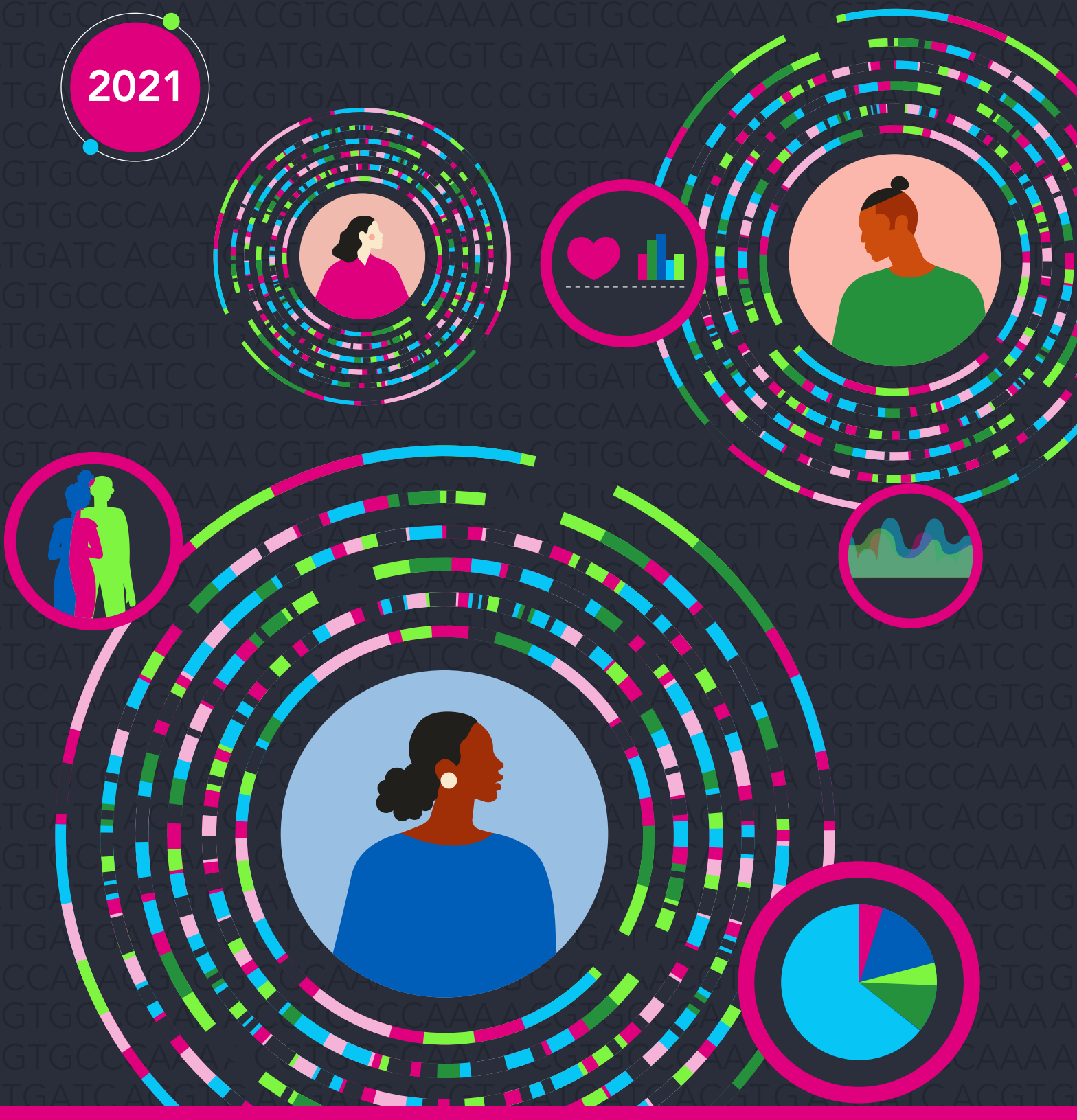
Annual Report

Genomics England

genomicsengland.co.uk

2021



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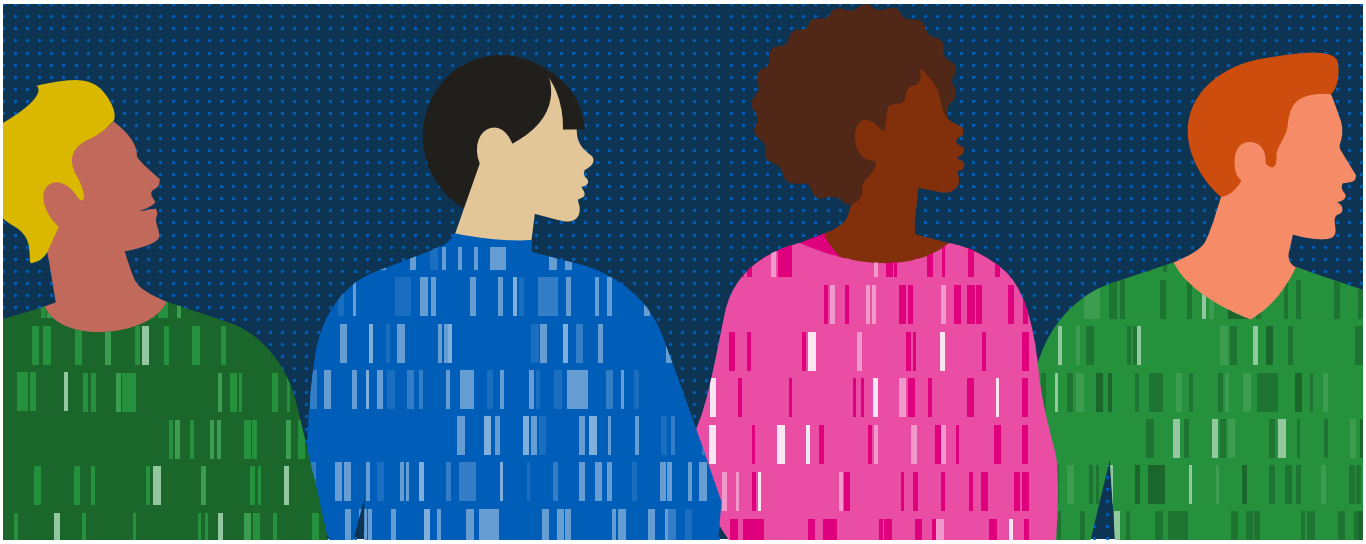
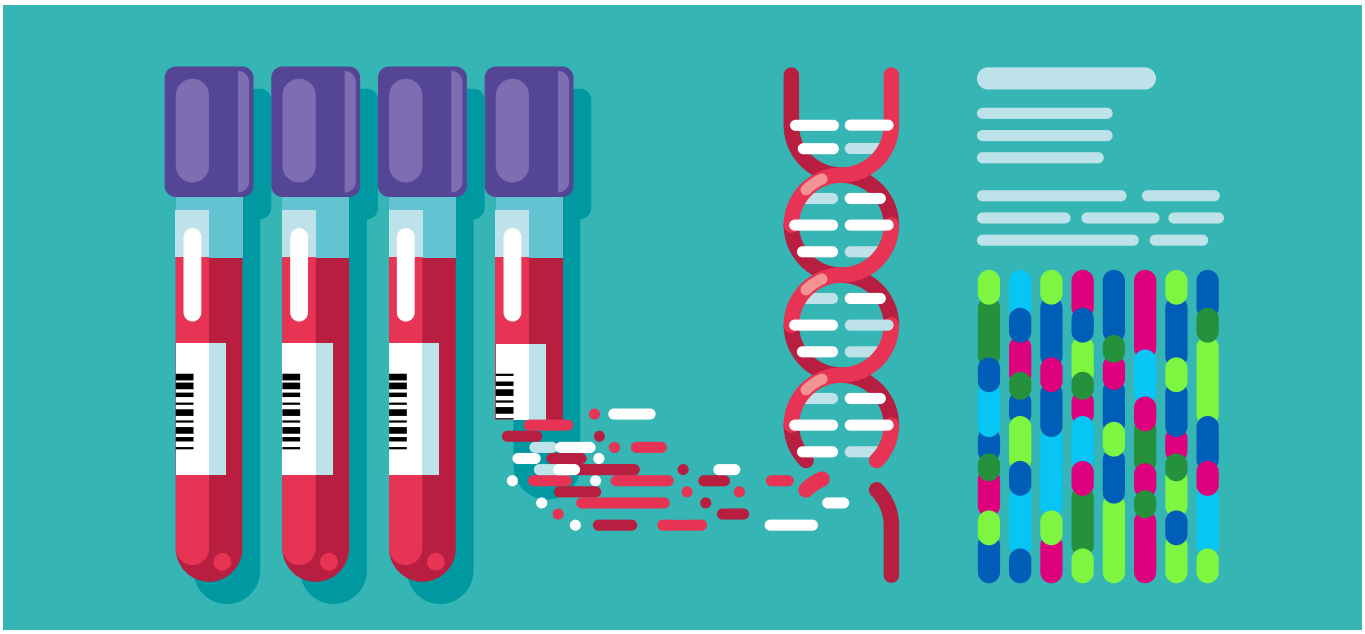


About Genomics England

Genomics England, with the consent of participants and the support of the public, is creating a lasting legacy for patients, the NHS and the UK economy. Initially focused on sequencing 100,000 genomes, we are now building on that foundation to enable faster, deeper genomic research, bringing the benefits of genomic medicine to all who need it.

Genomic healthcare superpower

The UK is a global superpower in genomics with a history of fundamental scientific discoveries and translation into clinical practice and improved patient outcomes. The UK has some of the richest genomic datasets in the world and the world's first nationwide whole genome sequencing (WGS) implemented in the NHS in England. This success is in large part due to the Government's ongoing investment and support in Genomics England, the NHS' investment in the NHS Genomic Medicine Service (NHS GMS), and the wider genomics ecosystem.



Through years of investment in landmark infrastructure projects and world-leading healthcare initiatives in addition to significant investment in the NHS infrastructure, the UK has become a clear front-runner in genomic healthcare.

We are now at a point where the readiness of the NHS coupled with our ability to sequence and analyse the genomic data at scale creates the opportunity to bring the benefits of WGS into mainstream healthcare.

Throughout the pandemic we have also seen the wider benefits of having a strong genomics capability in the UK which revolutionised the ability to track the spread of disease and to design more effective treatments.

Since its creation over 8 years ago, over £350m has been invested into Genomics England's unique mission, which has enhanced the UK's Genomics Ecosystem. This has reaffirmed the UK's position as a leader in a sector valued at over £5bn with growth forecast to be over 400% in the next 30 years.

Reviews

Ministerial review

Department of Health & Social Care

Sajid Javid

Secretary of State for Health and Social Care

> I am so proud of the role Genomics England has played and I'm pleased to see it go from strength to strength. It is now clear that so much of the future of healthcare will rest on our genomic capability, and I'm determined that Genomics England and the UK should continue to lead the world.

Nearly 70 years have passed since the double helix structure of our DNA was discovered here in the UK, and the 'secret of life' was unveiled. In the decades that followed, the UK has continued to lead the way with revolutionary discoveries in the field of genomics, including our pioneering 100,000 Genomes Project. That powerful partnership between Genomics England and the NHS has shown the world the value that whole genome sequencing (WGS) has to transform lives, especially those with rare diseases and cancer.

The UK's scientific response to the global pandemic has been another important milestone, underlining the importance of genomics in protecting and improving the health of every one of us. When COVID-19 emerged, we were one of the first countries in the world to scale up genomic sequencing and deliver ground-breaking programmes that have turned the tide of this pandemic, both here in the UK and around the world.

I am so proud of the role Genomics England has played and I'm pleased to see it go from strength to strength. It is now clear that so much of the future of healthcare will rest on our genomic capability, and I'm determined that Genomics England and the UK should continue to lead the world.

Today, the NHS in England is the first healthcare system in the world to offer WGS as part of routine care. WGS is helping more and more patients put an end to years of uncertainty by giving them a diagnosis for the very first time. I am also proud that my department is funding the latest research project between Genomics England and the NHS, which is assessing the benefits and considerations of using WGS to screen for rare inherited conditions in 100,000 newborn babies – another world first.

Since becoming Secretary of State for Health and Social Care, I have made tackling historic disparities in our healthcare system a central mission. Genomics is a key part of this. Equity of access to genomic medicine is one of the most important ways we can close historic divides in healthcare. The ever-lowering costs of sequencing mean that bringing the benefits of genomic medicine to all who need it is not just possible, but essential. I was proud to sign up to Genomics England's 'Diverse Data' programme, which is showing the way forward for how we can reduce health inequalities through genomics.

Genomics not only represents the future of UK healthcare but will play a crucial part in our economic future too. The sector already contributes around £2.5bn a year to the economy, creating many high-skill high-wage jobs of the future. Genomics will remain central to our Life Sciences Vision, and Genomics England will be central to its continued success.

This Annual Report underlines just how far we've come in this incredible field, and I'm proud to support Genomics England's life-changing mission to affirm the UK's role as a global genomics superpower.

Sajid Javid
Secretary of State for Health and Social Care

Chair review

Baroness Nicola Blackwood

Chair

Having spent the last six years supporting the UK technology and health sectors, I am honoured to serve as the Chair of the Genomics England board, a position I took up in May 2020.

Genomics has been used in healthcare for decades. Reaching mainstream scale has always been a case of "when", not "if", and the answer to that question seems to be "right now". With the advances in science, technology and clinical application over the last few years, we have begun to see an acceleration in the use of genomic medicine and its benefits to patients and populations worldwide.

Genomics England has been and remains central to this acceleration. In our support to the NHS, for example, we have been helping the NHS GMS launch its WGS offering. At the same time, we have been investing in our technology and partnering with organisations such as Amazon Web Services and LifeBit to continue to improve our genomic research services.

However, our role is to be more than a service provider. As an organisation, we act as a convener of communities: we start and support conversations between the public, participants, healthcare workers, and private and academic researchers. The reason is simple: it will be these communities who will drive our vision of genomic healthcare available to everyone, everywhere.

This vision won't come without its challenges. For example, whilst COVID-19 highlighted the value of the Life Sciences sector, it also reinforced the importance of public trust. Genomics England will continue to put ethics, information governance, and the participant panel at the heart of everything we do.

Alongside trust, our focus has shifted from the 100,000 Genome Project to delivering user-friendly platforms for clinicians and researchers at scale. This is why we are investing in our technology, to include our new cloud-based Research Environment and an informatics service to support WGS in the NHS GMS. This investment will also ensure that we can adapt more swiftly to this fast-moving sector, and that the data we hold is used for the maximum benefit of the UK population.

Any commitment to delivery comes with change, and this applies no less to our structural organisation. Our aim is to be more user- and service-focused, and to that end we have refreshed our executive team and made new non-executive appointments to support our governance and bring on further expertise.

As I mentioned in the opening, I am honoured to be serving as Chair, and here I would even say that I'm proud. The organisation we're building is transparent, trustworthy with the public's data, and able to meet our country's ambitions to transform healthcare for all.

Baroness Blackwood
Chair of Genomics England

CEO statement

Chris Wigley
Chief Executive Officer

This has been an extraordinary year for the genomics community globally, and a year of transformation for Genomics England. We are proud of the impact we've had in partnership with the NHS and many others, and excited about the road ahead.

Looking first at the big picture: in the face of the pandemic, genomics has come into the global conversation in a way that few would previously have thought possible. When the virus was first sequenced and the sequence posted online, it unleashed a global scientific effort of unprecedented scale and pace.

We're proud to have played our part in that effort through our work to understand what our human genome can tell us about the "COVID puzzle" of why some people respond so severely to the disease and others are asymptomatic. Our collaboration with the University of Edinburgh (GenOMICC) was the leading contributor to the global COVID-19 host genetics initiative programme, just as the Covid Genetics (COG UK) consortium was the leading contributor to global efforts to sequence the virus and identify novel variants.

Reflecting on my time at Genomics England, since arriving as CEO a few months before the pandemic, we have truly transformed as an organisation, and yet our founding spirit and sense of mission endure.

Considering that transformation, the most profound aspect is probably the answer to the question: "why does Genomics England exist?" Back in 2013 we were set up as part of the Olympic Legacy to deliver on a moonshot project – sequencing 100,000 genomes. At the time this was widely seen as impossible, not least because of the cost of sequencing then. But (like many plucky British science stories) the team, in partnership with the NHS and thanks to the efforts of the participants, achieved the impossible. The 100,000th genome was sequenced in December 2018, and work is continuing to further analyse the genomes and return new findings to participants, and advance the scientific frontiers on the back of this extraordinary dataset. Building on that foundation, we now have a much further horizon in our sights: ensuring that everyone benefits from genomic medicine. We do that both by working with the NHS to deliver genomics-based insights for patients, and by working with researchers in academia and biotech who develop novel understanding, diagnostics and treatments that can enhance healthcare. We think of that as a mutually-reinforcing infinity loop, as laid out in the strategy section of this report.

"We're proud to have played our part in that effort through our work to understand what our human genomes can tell us about the "COVID puzzle" of why some people respond so severely to the disease and others are asymptomatic.

CEO statement continued

Chris Wigley
Chief Executive Officer

To look at each of those areas in turn, in our healthcare work with the NHS we have…

- Supported the launch of the WGS Service in the NHS GMS.
- Provided genomic analysis for first 1,400 patients,
- The Department of Health and Social Care has approved our plans for further investment in the National Genomics Informatics System.
- Started the service design for the Newborn Sequencing Initiative announced in the October 2021 Spending Review, and completed a public dialogue on the initiative with our partners.
- Launched clinical proof of concept work for new cancer technologies such a long-read sequencing aimed at testing their efficacy at scale to support faster and more accurate diagnosis.

In our research work we have…

- Launched a new cloud based Research Environment in collaboration with Lifebit, a UK based startup to upgrade on our existing platform which was one of the first in the world.
- Sequenced over 28,000 COVID-19 patients in partnership with the genomics ecosystem.
- More than 4,250 researchers from 193 research institutions and 30 companies (including 8 of the 10 biggest pharma companies) have signed up to our research environment.
- Using our data set, scientists around the world (mainly the UK) have published 181 scientific papers in leading journals
- Published papers using the data we hold in the New English Journal of Medicine, Nature Cancer, and Nature Genetics in Medicine.
- Started collecting new forms of data, such as clinical imagery, which will soon be used to support new types of research in the Genomics England Trusted Research Environment.

In transforming Genomics England's structure, leadership and capabilities, we have…

- Welcomed extraordinary leaders and contributors at all levels
- Built new capabilities in service design, product management, user research, and cloud engineering.
- Restructured our teams around the services we deliver and convened multi-disciplinary science-tech squads to build the products that underpin those services.

However, this is only what we have accomplished so far. In the coming years we are focused on mainstreaming and scaling these services:

- We, in partnership with NHS England and NHS Improvement, will add new functionality, improve the user experience, and increase the number of cancers and rare diseases eligible for the WGS service diagnoses.
- The Genomics England Trusted Research Environment will be improved with new tooling and new sources of data (such as clinical imaging) to deepen our research collaborations with our partners and academic researchers.
- We will commence new programmes to address major health and research questions focusing on: newborn sequencing, diverse data, and trialling new technologies to diagnose cancer.

We are proud and excited that Genomics England headlined the £5bn healthcare research and developed investment in the October 2021 Government Spending Review.

If we step back and think about our goals with newborns, in cancer and diversity in genomic medicine – we have the opportunity with our partners to help everyone live longer, happier, healthier lives, served by the smartest healthcare system in the world.

That's what gets us out of bed in the morning. We hope you're excited by it too.

Chris Wigley
CEO Genomics England

Chair of Participant Review

Jillian Hastings Ward

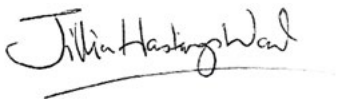
Chair of Participant Panel

The Participant Panel celebrated our 5th birthday in April 2021 and we're busier than ever, representing the interests of participants whose data is held by Genomics England. Our quarterly meetings are an opportunity for us to ask the senior leadership team anything, and we've been working hard in between to make sure that participant voices continue to be heard. Our members speak up for the participant community at the Access Review Committee, Ethics Advisory Committee and GeCIP Board and we have a regular slot at the induction sessions for all the new staff who join Genomics England. Our Vice Chair, Rebecca Middleton, recently joined the steering group for the (newborn screening pilot study). Several Panel members have also been interviewed for Chris Wigley's G-Word podcast (www.genomicsengland.co.uk/news/podcast), and have enjoyed sharing our perspectives with a wider audience.

The coming year will see the return of 'Additional Findings' (indicating a predisposition to certain forms of cancer or other treatable conditions in the NHS) to participants in the 100,000 Genomes Project. We've been shaping the way that this important activity will be carried out. For many of the participants who asked to hear about these, they will be very useful, but we need to make sure that they are delivered in a sensitive and appropriate way.

We're also keen that Genomics England puts in place a structured reanalysis programme for 100k participants who did not receive a diagnosis in the first round of analysis. This should just be the start, not the end, of their opportunity to benefit from genomics.

As recently reported in the Genome UK Implementation Strategy (www.gov.uk/government/publications/genome-uk-2021-to-2022-implementation-plan), we'll continue to speak up for the people in Genomics England's databank [/NGRL] in perpetuity. In the coming year, we're looking forward to welcoming new Panel members representing COVID patients and those opting in to research via the NHS GMS. We're also sharing our expertise to help the development of patient and public voice (PPV) roles in the GMS Alliances across England.



Jillian Hastings Ward
Chair of Participant Panel

Origins

Our history

DECEMBER 2012

Announced by former Prime Minister David Cameron – an Olympic Legacy

JULY 2013

Genomics England formally launched by then Secretary of State for Health during NHS 65th Anniversary Celebrations

JULY 2017

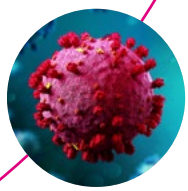
Chief Medical Officer launches Generation Genome and the Life Sciences report

DECEMBER 2018

Genomics England reaches goal of sequencing 100,000 genomes

JANUARY 2019

Long Term Plan "an NHS where access to secure linked clinical, genomic and other data will support new medical breakthroughs and consistent quality of care"



JANUARY 2020

COVID-19 cohort launched to identify biomarker for mild and severe patients

OCTOBER 2021

The Government continues its support of genomics through announcing Newborn Sequencing pilot and Diverse Data programme which will be implemented as part of £5bn Healthcare R&D package as part of 2021 Spending Review.

Original moonshot

In December 2012, the 100,000 Genomes Project was launched by then-Prime Minister David Cameron, with the goal of harnessing whole genome sequencing technology to uncover new diagnoses and improved treatments for patients with rare inherited diseases and cancer. It was also a "moonshot" programme that if successful would make the UK a world leader in genomic medicine and showcased the clinical benefit if scaled. Genomics England (GEL) was established to deliver this project in July 2013 through funding from the National Institute for Health Research (NIHR), and continues to operate today as a Government Company fully owned by the Secretary of State for Health and Social Care.

The 100,000 Genomes Project served as the basis for the NHS GMS being the first health care system in the world to offer WGS as part of routine clinical care. In building the evidence for WGS, Genomics England collaboration with NHSE/I was vital in creating 13 NHS Genomic Medicine Centres (GMCs). The 13 NHS GMCs enabled the recruiting and consenting of participants from routine care and to return the results for clinical care purposes; establishing a state-of-the-art sequencing centre run by Illumina, Inc.; building an automated analytics platform to return whole genome analyses to the NHS; and hosting a world leading genome data set for academic and commercial research.

The technologies and platforms pioneered and developed through the 100,000 Genomes Project and NHS GMS put the UK in a strong position to lead the world in the rapid application of genomics to tackle the COVID-19 pandemic. Whether that was in sequencing the virus to detect and track new viral variants to understanding how a patient's DNA impacts their response to SARS-CoV-2 infection. As we will articulate through this report, Genomics England's unique position at the intersection of frontline healthcare and cutting-edge research puts the UK in a perfect position to continue our proud leadership in genomics over the next decade and beyond.

"The technologies and platforms pioneered and developed through the 100,000 Genomes Project and NHS GMS put the UK in a strong position to lead the world in the rapid application of WGS to identify vulnerable individuals during the COVID-19 pandemic.

The 100,000 Genomes Project impact

The 100,000 genomes project is a ground-breaking programme designed with the goal of harnessing WGS technology to uncover new diagnoses and improved treatments for patients with rare inherited diseases and cancer, an ambition completed within five years, at the level of sequencing, but whose scope is ongoing.

Patients

Over 85,000 participants benefited from the 100,000 Genomes Project receiving genomic sequencing which could lead to diagnostic results, change of treatment course or genetic counselling – often the genetic diagnosis participants received, ended many years of "diagnostic odyssey."

Research platform

A dedicated research platform where clinicians and researchers work together to interpret and analyse the data from the 100,000 Genomes Project to provide better patient outcomes and clinical understanding.

World-leading

Over £100m of UK investment, created over c.500 science and tech jobs and hundreds of scientific papers cementing our world leadership position and providing a catalyst for the entire genomics industry (now worth £5bn[1]).

Return on investment

During the last decade the cost of sequencing has dropped significantly which propelled the scale up of a world-leading clinical genome sequencing service for patients. Ending long diagnostic odysseys through genetic diagnosis, it is estimated to have significant economic benefits as set out in a ground-breaking paper in the NEJM[2]. Prior to diagnosis, patients in the study, which involved 4,600 participants, had a median of 68 hospital visits, with a combined total cost of £87m. Following the project 25% of the 533 genetic diagnoses made had immediate clinical impact which reduced these costs significantly.

The 100,000 Genomes Project harnessed the information patients hold within them to create a rich and novel data set that could be used by researchers and clinicians to answer some of the biggest questions in understanding human health. Participants of the project gave consent for their genome to be sequenced, and their genomic data to be linked to their health records which included information about their medical conditions. The coalescence of genomic data and health data could then be anonymised and shared with health professionals to improve knowledge of the causes, treatment, and care of diseases which in turn will streamline the diagnostic process and enable tailoring of care to individuals (Barwell et al., 2018).

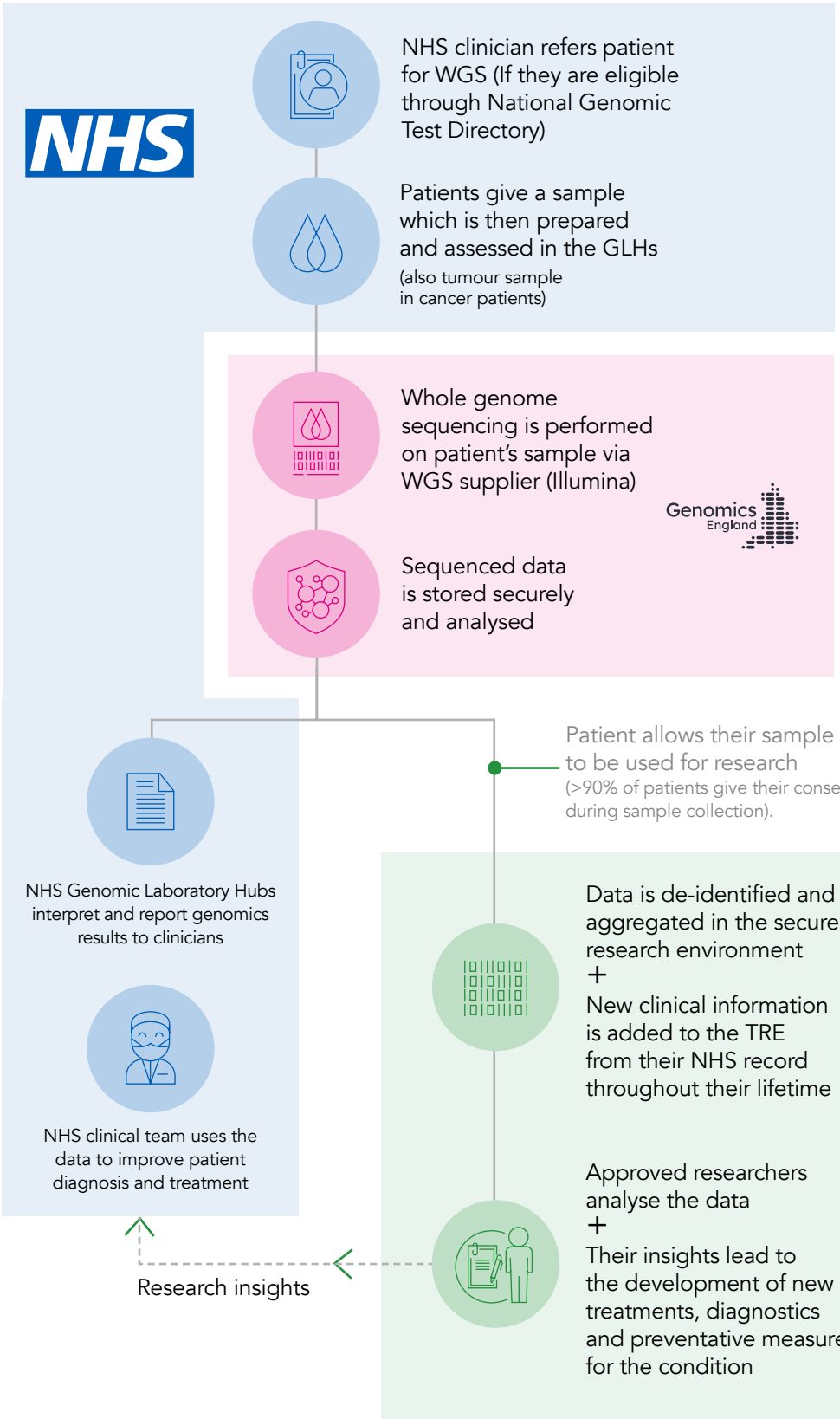
> "Participants of the project gave consent for their genome to be sequenced, and their genomic data to be linked to their health records which included information about their medical conditions.

[1] https://www.bioindustry.org/uploads/assets/2b60cf38-020b-4a97-8d8f84bb464b8b7d/BIOJ8942-Genomics-Report-210728-WEB.pdf

[2] 100,000 Genomes Project Pilot Investigators, 2021. 100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care—Preliminary Report. New England Journal of Medicine, 385(20), pp.1868-1880.

Genomics England at a glance

Whole genome sequencing - patient journey



How researchers access the National Genome Research Library

What we work on

117,000
genomes in our Reading Library

53
petabytes genomic and medical data

95,600
participants recruited (in 100k)

8/10
top pharmaceuticals as partners

21K
cancer genomic signatures

84
academic institutes

160+
publications using Genomics England Assets

550
research projects in our environment

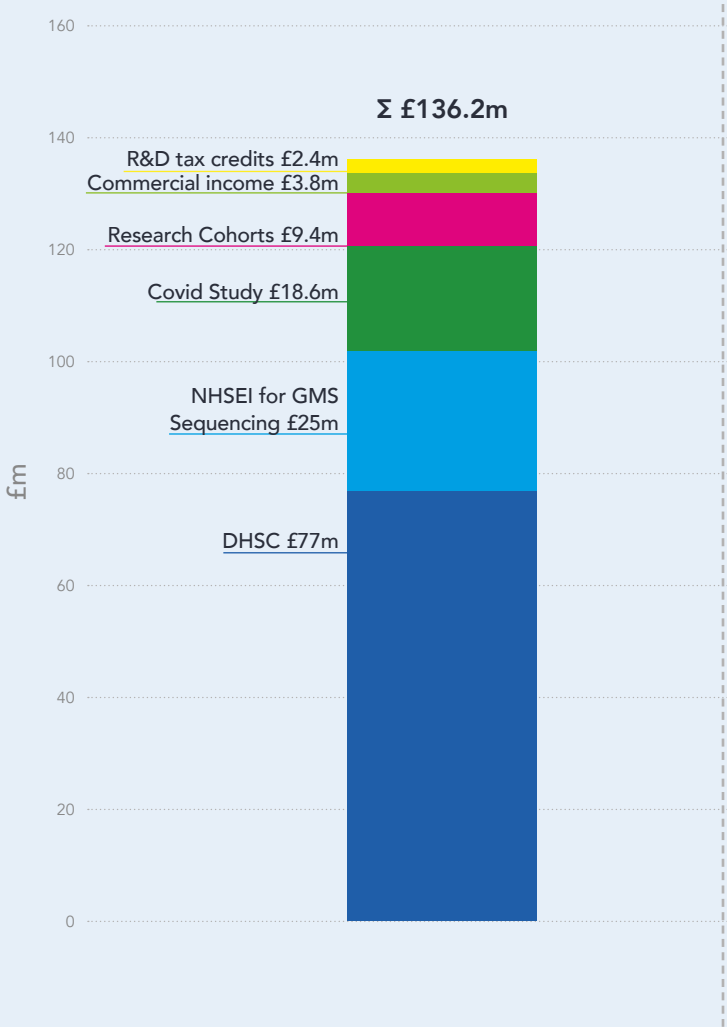
3,680
registered academic researchers

32
bioinformatic pipeline workflows

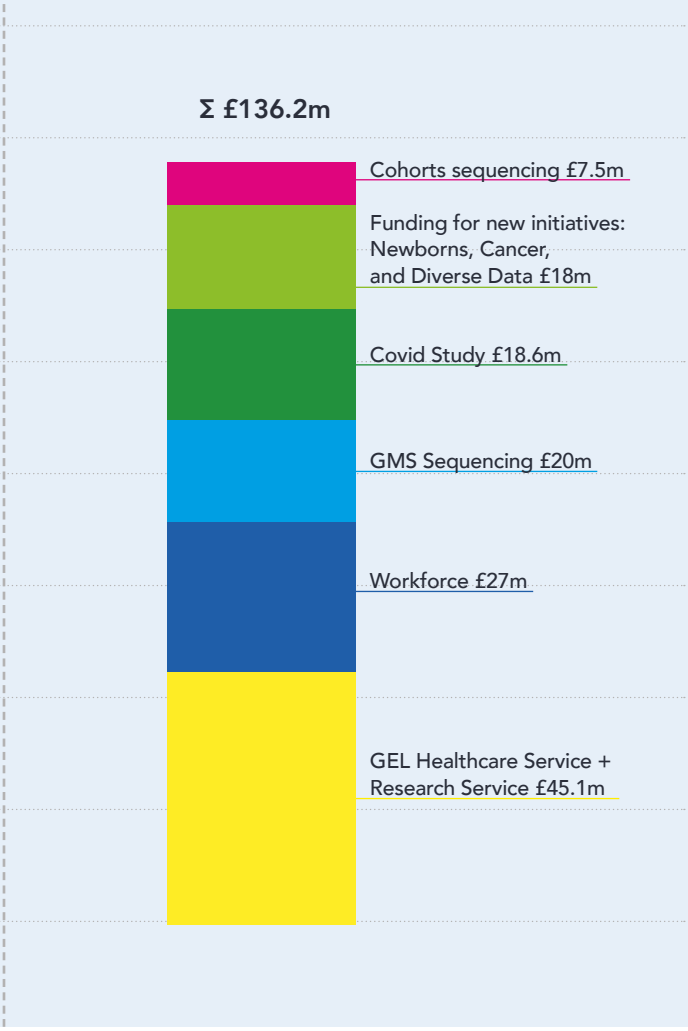
Over
94%
of patients who were offered WGS consented for their data to be used for research

Source and Application of Funding – 2021/22 Plan

Where does our funding come from?



How do we spend it?



Our virtues

Speed

we are conscious that our deliverables impact real people and even save lives – we need to work robustly and we also need to work fast

Focus

We prioritise what's important and finish what we have started before moving on

Curiosity

We recognise that curiosity and experimentation are good, and we learn from evidence and experience

Integrity

Both internally and externally; we are responsible stewards of patient data; we're straight with each other and our partners – we keep our word

Impact

We deliver services that enable the NHS to deliver better outcomes for patients and our partners for the long term

Connection

We are not an island, we are part of UK Health system, the global technology and life sciences community and will work with others to achieve common goals

Empathy

Patients are the priority, we ensure quality and relevance in our outputs, we are open and honest with them, and we put their interests ahead of our own

Our team

109
New joiners this year

39 nationalities

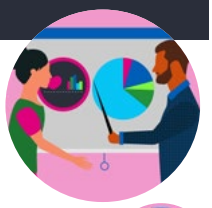
Gender identity

Women	Men
43%	57%

Average tenure

2.3 years

2021 survey results



96%
of Genomics England Employees believe in what the organisation is trying to achieve

84%
Agree transparency to the public is important to leadership at Genomics England

94%
Agree our purpose will be as relevant (or more) tomorrow as it is today

68%
Would recommend Genomics England to friends as a great place to work (up by 26%)

Vision

Turning science into healthcare, together

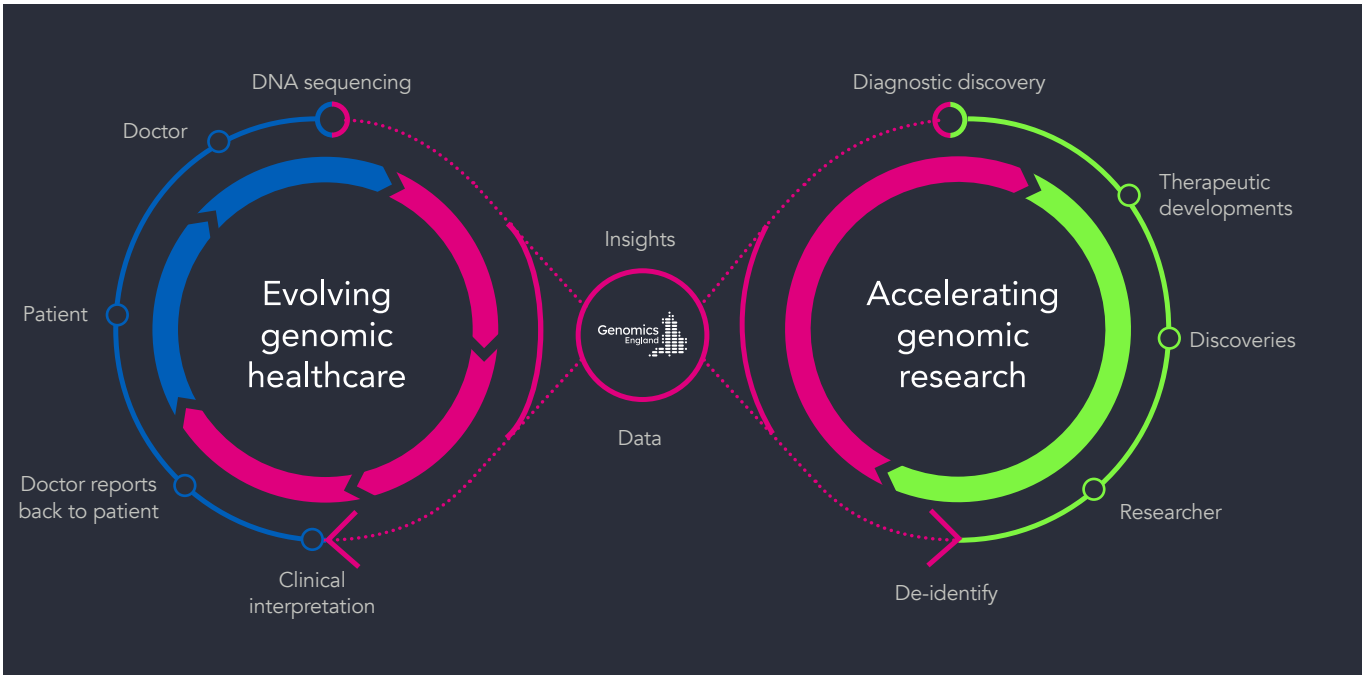
By providing researchers with access to large datasets, genomic research can be accelerated significantly, enabling new insights and scientific discoveries. Furthermore, patients will benefit from improved care, by identifying genetic illnesses earlier, and developing personalised healthcare tailored to a patient's genome. Enabling NHS teams to achieve better patient outcomes via genomic insights that will inform more precise diagnoses, through stratified therapeutics and personalised interventions. This is possible due to the creation of an ethical and transparent framework which will ideally form the foundations of a UK genomics industry.

Genomic, clinical and real-world datasets are increasingly linked and accessible. In this world, our goal is to be a "national champion" for genomics and bioinformatics: using machine learning and expert teams to deliver deep insights, at scale. In this way, we will work in partnership to ensure that everyone benefits from genomic healthcare: better outcomes for NHS patients, in a more productive health system, with clear pathways to bring the latest research to bear in mainstream health.

Current genomic clinical diagnostic methods analyse the genomic information from patients with suspected rare diseases and cancers by analysing one segment of a gene at a time. This type of testing can lead to very long wait for diagnosis if the suspected mutation was not found and it limits the volume of clinical testing.

Genomics England have been using new technology (WGS) that made it possible to sequence millions of fragments of DNA simultaneously, tipping the scale to enable clinicians to offer genetic testing to many more patients, and test one patient for a large number of genetic diseases with one diagnostic test.

Infinity loop



Using the infinity loop model, we can support NHS teams to achieve a better patient outcome by accelerating genomic research using our trusted data environment and work in partnership to augment an ecosystem-wide approach to clinical trials, target discovery and therapeutic development, which will feed back to clinicians to inform more precise diagnoses. This feedback system will therefore generate efficiencies by linking and curating relevant datasets for system-wide optimisation. Building a next generation of genomics technology infrastructure within a coherent clinical and research architecture.

And when we talk about doing all this work "together" – that's core to the origins and culture of Genomics England. We work closely with our Participant Panel to ensure that the patients and research participants that we exist to serve have a real voice in shaping our priorities and work. For instance, their representatives sit on the Access Review Committee that decides whether researchers from academia or industry can gain access to the Research Environment. Look out for the review from Jillian Hastings Ward, the chair of the Panel, which is a few pages back in this report.

Impact

Impact

Patient stories

100,000 Genomes Project

Jessica's story

Jessica was born with GLUT1 deficiency syndrome, which affects her movements, general development, and causes epilepsy.

"If we had this done when Jessica was born and found out the results straight away, we would have been on the right track immediately."

Jessica's Mum

Diagnosis of rare diseases in children through the 100,000 Genomes Project

The first children to receive a genetic diagnosis through the 100,000 Genomes Project have been given their results at Great Ormond Street Hospital (GOSH), part of the North Thames NHS Genomic Medicine Centre.

Both Georgia Walburn-Green and Jessica Wright had rare, undiagnosed, genetic conditions when they joined the Project. WGS pinpointed the underlying genetic changes responsible for their conditions.

As well as removing a large amount of uncertainty for the families, the results stand to have a major impact on many areas of their lives including future treatment options, social support and family planning. They also have the potential to help many more children with undiagnosed conditions who may be tested for these genetic mutations early on and be offered a diagnosis to help manage their condition most effectively.

Jessica's story

Jessica Wright was also recently diagnosed through the 100,000 Genomes Project.

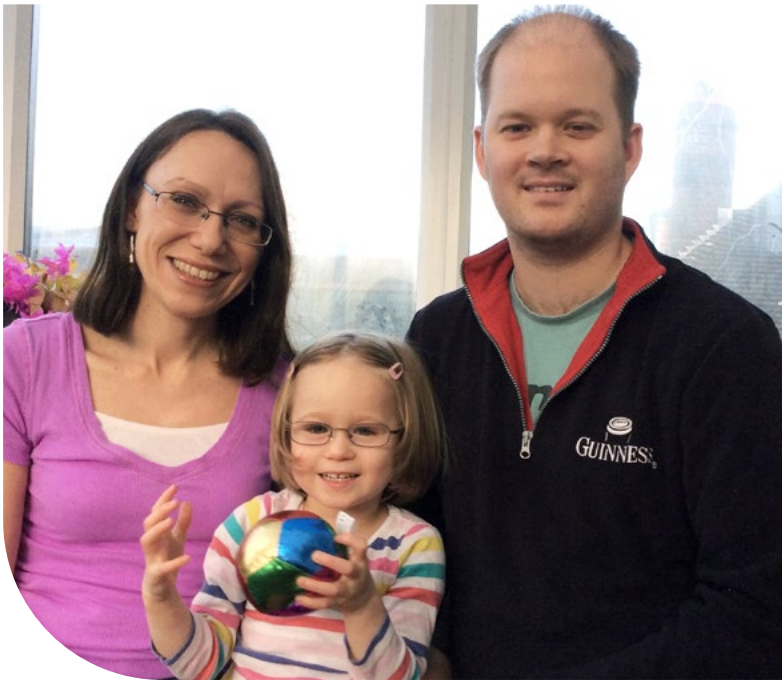
Mum, Kate Palmer talked about what the diagnosis means for them: "Now that we have this diagnosis there are things that we can do differently almost straight away.

Her condition is one that has a high chance of improvement on a special diet, which means that her medication dose is likely to decrease and her epilepsy may be more easily controlled. Hopefully she might have better balance so she can be more stable and walk more. She's now four years old and still looks like a wobbly toddler trying to move around!

A diagnosis also means that we can link up with other families who are in the same boat and can offer support. The condition is still quite rare but there are definitely other children out there who have it. I'm really looking forward to saying 'We are one of you, we have this problem too!' More than anything the outcome of the project has taken the uncertainty out of life for us and the worry of not knowing what was wrong. It has allowed us to feel like we can take control of things and make positive changes for Jessica.

It may also open doors to other research projects that we can to go on. These could be more specific to her condition and we are hopeful that they could one day find a cure."

GOSH Consultant in Clinical Genetics, Professor Maria Bitner-Glindzicz says: "With undiagnosed genetic conditions it really is a case of the more families we test, the more we can diagnose. In order to confidently say that a particular gene is likely to be the cause of a condition and not just natural variation that we see in everyone's genes, we have to match up gene mutations and symptoms across several children to find common features. The more children we therefore have to cross check against, the more likely it is that we can find these common features and give a diagnosis."



> Her condition is one that has a high chance of improvement on a special diet, which means that her medication dose is likely to decrease and her epilepsy may be more easily controlled.

6,625
Diagnosed with rare diseases

Creating an impact, changing lives

Unique participant influence model

NHS
transformation of pathways and skills

50%
actionability in cancer

130
companies in Discovery Forum

4k
academics

Unique research environment

Ending the diagnostic odyssey

Rare disease patients often face a difficult journey to diagnosis, commonly termed a 'diagnostic odyssey' which encompasses the time from initial disease recognition or symptom onset to a final diagnosis as experienced by patients. This can usually involve multiple referrals and encounters with specialists and a string of often unnecessary investigations having the right diagnosis and the appropriate treatment being delayed, potentially causing their condition to deteriorate further.

Genomics England aims to deliver life changing results for patients and end prolonged diagnostic odysseys. Through the NHS GMS, England has become the first in the world to systematically integrate genomic technologies, including WGS, into routine clinical care. Going forward, this will mean patients with suspected rare genetic diseases can be offered WGS under the NHS GMS which aims to dispel uncertainty by reducing the time it takes to gain a diagnosis and develop new and effective personalised treatment plans.

"Many rare diseases, like mine were notoriously difficult to pick up in the past. But now the maturity of genomic technology allows us to diagnose rare conditions much earlier and more accurately. Not only can this give life-changing results and access to treatments, but a diagnosis is also a huge emotional relief for families, who may have waited years."

Baroness Nicola Blackwood
Chair, Genomics England

Impact

Research and partnerships

Personalisation of cancer therapies by developing algorithms which help characterise tumours and expose their weaknesses

Professor Serena Nik-Zainal
NIHR Research Professor
Cambridge University's Department of Medical Genetics

UNIVERSITY OF CAMBRIDGE

NIHR | National Institute for Health Research

> WGS is providing clinicians and researchers with a treasure trove of data and enables the personalisation of treatments according to the specific genetic signature of the tumour in question.

Genomic sequencing is transforming our understanding of cancer's origins and complexity. Unlike previous categorisations based on the location of tumours and basic biomarker tests, WGS is providing detailed characterisations of the genetic changes that cause cancer and the molecular pathways that tumours are exploiting to evade our natural defence mechanisms. Thanks to WGS, clinicians and researchers have access to a treasure trove of data that enables the personalisation of treatments according to the specific genetic signature of the tumour in question. Until recently, we could only test for individual genes or gene panels such as mutations in tumour suppressor genes like BRCA1 and BRCA2, which increase the risk for breast, ovarian and prostate cancer. Now with WGS, we can identify not only the genes in question but also other tumour types where these genomic signatures report similar dysfunction of tumour suppressing pathways.

For example, in a recent study, academic partners in Cambridge used Genomics England data to develop an algorithm (MMRDetect) to identify tumours with 'mismatch repair deficiency' which potentially makes them sensitive to checkpoint inhibitors (immunotherapies). The algorithm uses the mutational signatures that were identified in knock-out experiments and was trained on WGS data from NHS cancer patients in the 100,000 Genomes Project. Having developed the algorithm on tumours in this study, the plan now is to roll it out across all cancers picked up by Genomics England.

The MMRDetect algorithm could be potentially used as soon as a patient has received a cancer diagnosis and their tumour signature has been identified through genome sequencing. We can then use those tumour signatures to figure out which repair pathways have stopped working in each person's tumour, and what treatments should be used specifically to treat their cancer. Our team believes that this tool could help to transform the way a wide range of cancers are treated and save many lives by providing the best diagnosis for patients.

> The outcomes from Dr Nik-Zainal and her team's work demonstrate perfectly how quickly and effectively we can return value to patient care by bringing together a community of leading researchers through Genomics England's platform.

Parker Moss
Chief Ecosystems and Partnership Officer
Genomics England

MMRdetect algorithm

MMRDetect is a mutational signature-based classifier for identifying tumours with mismatch repair deficiency using whole-genome sequencing data. Similar algorithms have been developed to identify other features of genomic instability like homologous recombination deficiency that can identify responders to parp-inhibitors, and there is a pipeline of many more algorithms to come.

For the full scientific publication:

nature cancer

A systematic CRISPR screen defines mutational mechanisms underpinning signatures caused by replication errors and endogenous DNA damage

www.nature.com/articles/s43018-021-00200-0

Commercial partners case studies

BIOMARIN

BioMarin is an American biotechnology company founded in 1997 and headquartered in San Rafael, California. BioMarin's core business and research is in enzyme replacement therapies and it was the first company to provide therapeutics for mucopolysaccharidosis type I (MPS I) and for phenylketonuria (PKU). BioMarin is actively developing promising first-in-class or best-in-class therapies to treat rare genetic diseases, which mostly affect children. BioMarin have a full pipeline of drugs in development for a number of rare diseases which is highly relevant to Genomics England dataset, given that Genomics England holds data of participants representing several of these indications, including those recruited for lysosomal storage diseases (e.g. MPS I) and participants with secondary diagnoses for PKU, MPS and Batten disease.

BioMarin started their collaboration with Genomics England in 2018 and were the first adopters of Genomics England services and have been with us right from the start, focusing on helping rare disease patients get better treatment options. Genomics England is closely engaged with the Bioinformatics group based out of the San Francisco Bay area.

During their collaboration with Genomics England, BioMarin identified one patient recruited into the 100,000 Genomes Project who carried two pathogenic mutations in a gene not related to a condition or which they were referred for, and this was fed back to the patient's NHS clinical team. In addition, BioMarin published a paper where they included few participants from 100,000 Genomes Project for which intellectual disabilities could be attributed to mothers with pathogenic mutations and leading to maternal PKU diagnosis. BioMarin continues to work within Genomics England research environment looking further into genes and variants contributing to neurological phenotypes and they will be using our new, cloud based research environment to accelerate their research in rare disease space even further.

COMPANY PROFILE

> More than twenty years of making a big difference in rare patient populations.

BIOMARIN AT A GLANCE

Headquarters
San Rafael, California

Employees
3,000

Founded
1997

Focus
Rare genetic diseases

Chairman and CEO
Jean-Jacques Bienaimé

Real world evidence (RWE) datasets are datasets generated from data obtained from clinical routine care. NHS' ability to track and record patient interactions with the healthcare system makes UK one of the global leaders in RWE data generation. Genomics England has demonstrated that WGS can provide diagnostic value-add as a genetic test introduced into mainstream healthcare as evidenced by the pioneering 100,000 Genomes Project. However, more translational research is needed to understand the genetic mechanisms of disease to design better drugs, clinical trials and eventually, precisely administer right treatments for the right patient populations.

This project aimed to leverage the unique data asset generated by Genomics England as well as to draw on the collaborative framework of clinical – academic experts, known as Genomics England Clinical Interpretation Partnership (GeCIP). It also had a strategic aim to introduce Genomics England across MSD, with the potential of expanding collaborations in the future. Melanoma patient cohort was used as the case study example.

To date, MSD, working in collaboration with the melanoma GeCIP and Genomics England, successfully characterised the genomic profile of the 300+ melanoma patients enrolled into the Project. Additionally, we described the completeness of clinical data for outcomes data available at the time of analysis. We presented our research collaboration and initial findings at EU ISPOR 2019 and ESMO 2020. Final results of this work have also been submitted for publication and are under review in the journal, Cancer Medicine.

Although this initial work has identified gaps in the clinical data available for melanoma, today we are in advanced stages of a collaboration with the GeCIPs to remedy this. The clinical academics are using their clinical network to reach out to the hospitals who recruited the original melanoma patients and can access clinical records to improve the corresponding clinical data. This industry-funded endeavour will ensure that Genomics England melanoma cohort will have a complete clinical record alongside the genomic information, enabling all research community to conduct studies which in time can lead to identification of new biomarkers for the disease and better design of clinical trials.

Project title:
Methodology of linking and analysing real world genomics and clinical outcomes data

Area of research:
Cancer/melanoma

COMPANY PROFILE

"We're following the science to tackle some of the world's greatest health threats.

MSD AT A GLANCE

Headquarters
Kenilworth, NJ

Employees
34,996

Founded
1891

Focus
Therapeutic discovery and development

Chairman and CEO
Robert M. Davis

Valo

Valo Health is a technology company applying human and machine intelligence to accelerate the creation of life-changing medical treatments by developing a next-generation, end-to-end drug development technology platform with the potential to deliver breakthrough medicines at less than 50% of the cost and in 50% of the time.

Within the Genomics England Research Environment, Valo have applied their proprietary high-dimensional machine learning approaches and combined calculation of genetic risk metrics (measuring each participant's predisposition or risk for a certain phenotype or set of phenotypic traits) and proprietary stratification approaches to derive deeper insight into both genetic and clinical aspects of neurodegenerative patient journeys. They will leverage this high-dimensional combination of genetic and clinical information about each patient in conjunction with more than 125 million patient-years of data and proprietary computational platform to discover subtypes of disease, and the mechanisms, outcomes, and clinical progression associated with each.

COMPANY PROFILE

> Valo envisions a world where every disease has a treatment through the transformation of the drug development process.

VALO AT A GLANCE

Headquarters
Boston, MA

Founded
2019

CEO
Marc Josefsson

Employees
195

Focus
Integrated drug discovery and development platform

New horizons

Scaling up whole genome sequencing

The NHS Long Term Plan committed to sequencing 500,000 whole genomes, as part of a new focus on preventative care:

As part of the NHS' contribution to the UK government's broader aims to reach five million genomic tests and analyses over the same time frame, the new NHS GMS will sequence 500,000 whole genomes. This builds on the legacy of the ground-breaking 100,000 genomes programme, that was made possible because of the unique partnership between Genomics England and the NHS"

As well as referencing:
"Linking clinical, genomic, and other data to support the development of new treatments to improve the NHS and making data captured for care available for clinical research."

The NHS is committed to working with Genomics England in order to deliver this ambition. Given we provide the end-to-end informatics system, as well as the advanced technical analysis to support clinicians to utilise the NHS WGS service.

Genome UK

The Government's Genomics Healthcare Strategy, Genome UK[1], was published in September 2020. It describes a ten-year ambition to "Create the most advanced genomic healthcare system in the world underpinned by the latest scientific advances, to deliver better health outcomes at lower cost." Genomic England's role as a contributor to genomic research and healthcare, means it will be central to the successful delivery of this strategy. Over the course of 2021/22 Genomics England will be delivering programmes to establish the foundations of advanced genomic healthcare which focuses on diagnosis, personalised medicine, prevention and early detection and research (source[2]):

Diagnosis and personalised medicine

"We will help people live longer, healthier lives by using new genomic technologies to routinely identify the genetic determinants of rare diseases, infectious diseases and cancer. We will detect cancers earlier, and we will provide personalised treatments to illness."

2021/22 commitments:

- A proof-of-concept programme to explore next-generation approach for the diagnosis and treatment of cancer, such as trialling Oxford Nanopore's sequencing technology.
- Roll out of WGS in the NHS in England, enabled by Genomics England's end-to-end informatics system.

Prevention and Early Detection

"We will use genomics to accurately predict the risk of chronic diseases and our national screening programmes will use genomics to identify at-risk populations."

2021/22 commitments:

- Significant public engagement on a potential newborn sequencing initiative focusing in particular on ethics, clinician attitudes and public expectations.
- The GenOMICC consortium, in partnership with the University of Edinburgh, will look at why some people develop severe illness after being infected with Coronavirus while others only develop mild symptoms, or even remain asymptomatic.

Research

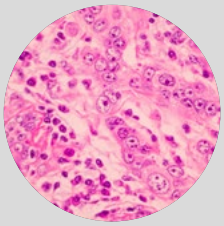
"We will extend the UK's world-leading position at the forefront of discovery-led and translational genomics research, continually expanding our collective genomics knowledge base. We will develop an ecosystem of world-leading secure genomics datasets, powering international research and supporting a seamless transition of impactful research findings into the healthcare setting backed up by robust implementation research programmes.

2021/22 commitments:

- A major drive led to improve the diversity of genomic data, addressing the historic under-representation of data from minority ethnic groups in genomic datasets, which results in health inequalities.
- Develop a new, next-generation Trusted Research Environment to provide improved, authorised access to genomic data and other linked data to researchers from across the sector.

Source
1: www.gov.uk/government/publications/genome-uk-the-future-of-healthcare/genome-uk-the-future-of-healthcare
2: www.gov.uk/government/publications/genome-uk-2021-to-2022-implementation-plan/genome-uk-2021-to-2022-implementation-plan

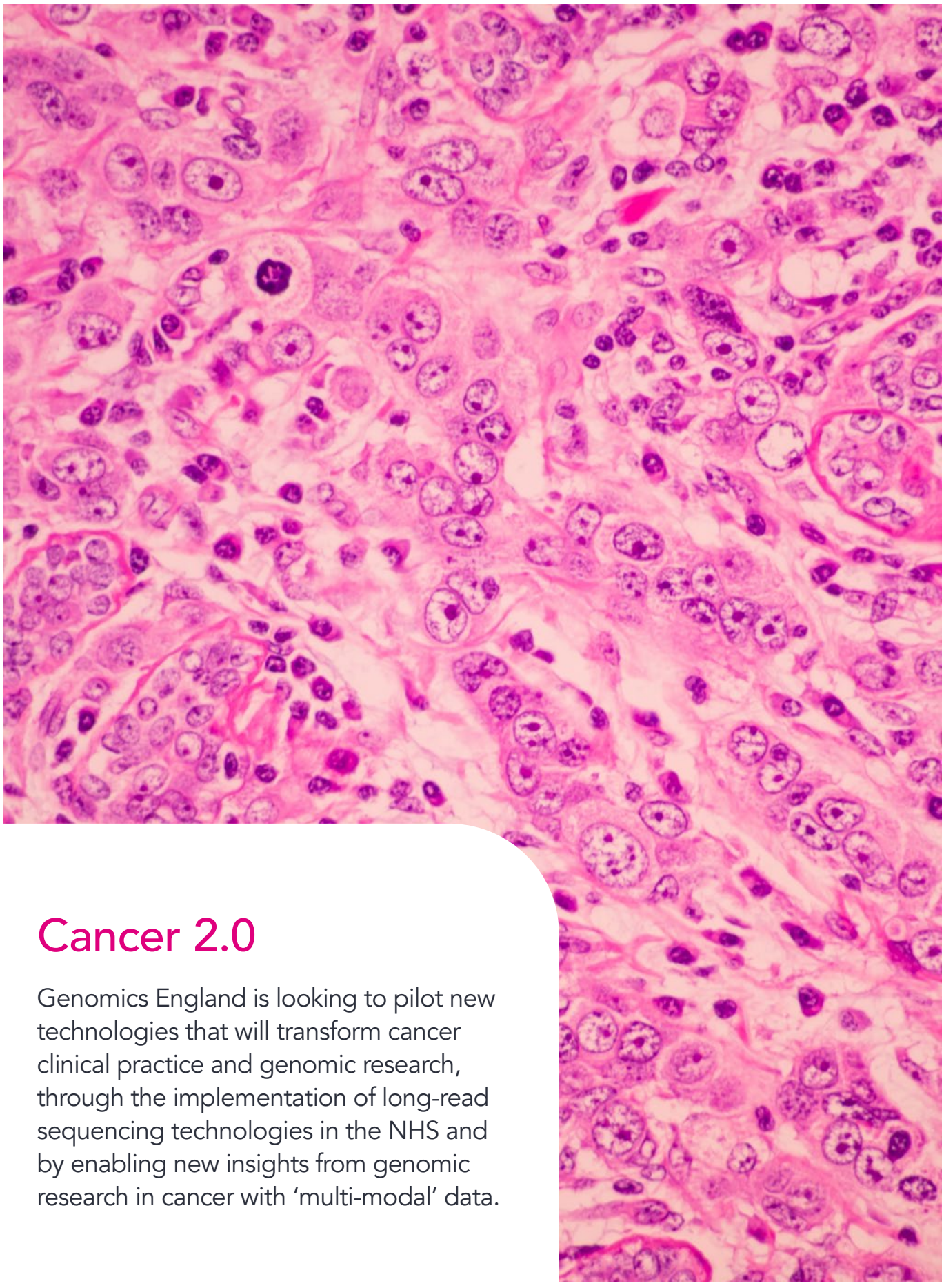
New horizons:
Four key innovations started in 2021



Cancer 2.0
- Multi-modal programme
- Long read programme

Diverse data

Newborns



Cancer 2.0

Genomics England is looking to pilot new technologies that will transform cancer clinical practice and genomic research, through the implementation of long-read sequencing technologies in the NHS and by enabling new insights from genomic research in cancer with 'multi-modal' data.

Multi-Modal Programme

Cancer patients often have vast amounts of health data held within an array of different formats and systems. This data is often unstructured information held within clinical notes, pathological images, laboratory reports and more. Gathering data, sorting it into meaningful consistent formats and marrying it with genomic data will support healthcare professionals to make better and quicker treatment decisions for cancer patients in the future. Organising, indexing and structuring the information also has the potential to allow for researchers to assess vast amounts of deidentified patient data unlocking new insights in the understanding of cancer. In collaboration with the NHS GMS, Genomics England is currently gathering de-identified health data donated by cancer patients to create a structured catalogued library for researchers to access, in a controlled environment, to study the impact of disease within the genome and improve the understanding of cancer.

> We are leading the world with [the Multi-modal project] Genomics england are the only organisation with the vision, the infrastructure, and the data to take this huge step forward. And the effects could be world-changing.

Prabhu Arumugam
Healthcare data analytics and technology focused academic clinician, Genomics England

> The Multi-modal project is incredibly exciting. I think it's really powerful. It will help us realise the potential of precision medicine going forward.

Professor Louise Jones
Lead for Molecular Pathology, Genomics England,
Group leader, CRUK, Barts Cancer Institute

Our aims

1

Legacy
Provide the world's largest library of linked genomic, clinical and imaging data

2

Expedite
Understand the utility of morphological changes in cancer and its relationship to genomic sequencing

3

Foundations
Accelerate the way for the NHS to become an entirely digital pathology service

Long-Read Programme

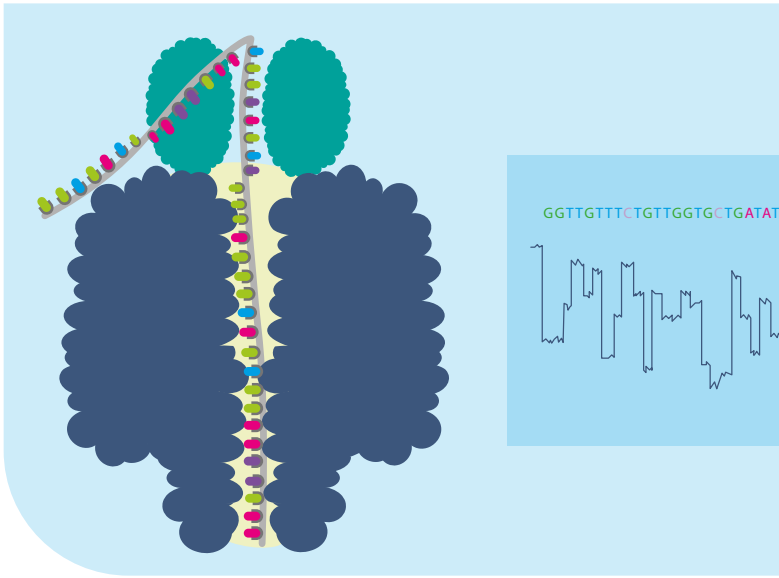
Cancer is a group of genetic diseases that result from changes in the genome of cells in the body, leading them to grow uncontrollably. These changes involve changes in the base pairs – the building blocks – of the genome. Currently these mutations can be analysed using short-read sequencing which cuts up the DNA samples into small segments, amplifies and 'reads' them before piecing them together, like a complex jigsaw. Long-read sequencing is a new technique that involves reading sequences of between 10,000 and 100,000 base pairs in one go, up to 1000 times longer than short-read sequencing. This novel technique ensures the genome sequence is assembled from much larger pieces at a faster rate reducing the opportunities for error, uncertainty, reading major rearrangements of the DNA better and resulting in quicker results potentially allowing the clinician to better treat the patient. Genomics England in partnership with the NHS is undergoing a complex initiative to explore and assess the potential of long-read sequencing with the aim of accelerating turnaround times to benefit patients in the future.

> "I think the really exciting thing about long-read sequencing is we're starting to bring in a new technology, a British technology, that can very quickly get very detailed information about a patient's cancer.

Professor James Brenton
Senior Group Leader and Honorary Consultant in Medical Oncology,
Cancer Research UK
Cambridge Institute

> "The dream for us is to have [long-read technology] embedded in the NHS, running smoothly, accessible to clinicians, and to have ultimately a rapid diagnosis for patients with cancer.

Emma McCargow
Programme Lead - Cancer 2.0,
Genomics England



Our aims

1. The programme will explore and develop the capability to analyse whole genome long-read sequencing generated within the NHS
2. Build a robust high quality long-read & methylation dataset within our research environment
3. Increase the potential opportunities to identify new clinical insights into cancer for use in commissioning the technology within the NHS

Diverse data

To date, studies of human genetics have largely focused on populations from European ancestries, which has contributed to a world where the benefits of genomic healthcare risk are not being shared equally. The overrepresentation of populations from 'WEIRD' societies (Western, Educated, Industrialised, Rich and Democratic) in genomic databases has resulted in the misdiagnosis of gene-disease relationships, limited the generalisability of findings from genomic research, and reduced the evidence base for translating these findings into clinical care for all populations.

To address this historical gap, we must work across the whole pipeline of genomic research and health care delivery, from the populations we work with and the data we collect, to the analyses we carry out and the availability of genetic testing. We expect this to increase our understanding of human history, biology, and health disparities, as well as discoveries relevant for health care delivery.

Genomics England recognises the unjust treatment of many minority groups and their exclusion from the genomic community and is committed to addressing this through the 'Diverse Data' programme. This initiative aims to make genomic medicine and its insights work for everyone, in both a national and international context. Addressing wider healthcare disparities is not something Genomics England alone can solve. We will work with NHS England, grassroots communities, training institutions and other partners, to overcome the challenges caused by the lack of genetic data diversity. This will include developing tools and approaches to enable researchers and clinicians to better interpret genomic variation to support clinical decisions in all communities, and build resilient and trusting relationships within traditionally excluded communities. Taking steps to redress this underrepresentation is vital as we move towards achieving Genomics England's vision of a world where everyone benefits from genomic healthcare.

> "There is both an urgent scientific need and a moral imperative to diversify genomics. Not everyone realises, but by focusing on, and working with, minoritised groups, everyone benefits from better genomic medicine.

Dr. Maxine Mackintosh
Programme Lead for Diverse Data,
Genomics England

> "You shouldn't only be exploring these conversations of diversity on a voluntary basis, they should be built into the way you're working…and this programme is aiming to do that.

Dr. Natalie Banner
Understanding Patient Data Lead,
The Wellcome Trust

Our aims

1

Understand the data gap: Define what diversity means, to whom, and with what impact

2

Fill the data gap: Sequence traditionally excluded groups to improve representativeness in genomic data

3

Bridge the data gap: Work with clinicians, researchers, and community groups to develop new tools, processes, and approaches to changing practices

4

Close the data gap together: Bring together everyone addressing this complex problem to streamline, amplify and augment the community's efforts

Newborns

The newborns initiative provides the opportunity to explore and evaluate the feasibility, effectiveness and implications of using WGS to:

- Expand the number of rare genetic conditions looked for in newborns, allowing timely diagnosis and treatment so avoiding the harm and improving health outcomes.
- Enable research and investment in the UK life sciences ecosystem to accelerate discovery and access to new and improved diagnostics, treatments and preventative therapies.
- Have the potential to allow repeated use of genomic information through the lifetime of an individual.

The initial funding for this work has been focused on engaging with public, NHS, industry and academia to develop the design for a programme that would sequence up to 200k newborn genomes in the next 3-4 years to provide the evidence, feasibility and impact for a broader NHS service.

Public support for the initiative is vital and insight into this was gathered through a large public dialogue of 130 participants (link to report), which found people strongly supportive but identified some key principles that are being used to shape the programme design.

> “This programme has the potential to transform the lives of children born with rare genetic diseases by identifying them quickly so they can access the right care earlier. It will also help us understand whether and how genomic data can be used to help diagnose genetic diseases in individuals through their lifetime. We will work closely with the healthcare community and the public to create a research pilot that will help us understand the benefits and challenges to delivering newborn sequencing in the UK.

Alice Tuff-Lacey
Programme Lead, newborns initiative, Genomics England

Our aims

1. Early diagnosis and care for childhood-onset rare genetic conditions
2. Enabling research to identify potential new treatments for NHS patients
3. Exploring the potential of a lifetime genomic record

> “Having worked with children with rare diseases for over a decade, this really is a unique opportunity to add years to life, and life to years, whilst facilitating research to continually enhance care across all disease groups. I am proud to work in a universal health service free at the point of need, that operates at the limits of science, and I truly believe we are in a fantastic place to lead the way globally on a programme of such huge scale and significance.

Sarah-Jane Marsh
Chair, Newborn Genomes Programme NHS Steering Group; CEO, Birmingham Women’s and Children’s NHS

The ecosystem

The ecosystem

Genomics England is just one part of a wider ecosystem in the UK that has been built through years of consistent investment:

NHS Genomic Medicine Service

In the NHS today genomic testing has received significant increases in funding and testing is now expanded to be more comprehensive for both rare and inherited diseases and cancer. The NHS GMS is driving the embedding of genomics into mainstream healthcare at scale to enable personalisation of treatment and care.

Established in 2006, UK Biobank is the world's foremost biomedical research cohort, which has attracted over £200m of industry investment and is used by over 20,000 researchers. Located in the North-West, it is also a key part of the UK genomics infrastructure outside the "golden triangle".

Our Future Health was established in 2018 through £79m of UKRI funding, originally as the "Accelerating Detection of Disease challenge". It will be the UK's largest ever health research programme and will collect and link multiple sources of health and other relevant information, including genetic data, across a diverse cohort of 5 million people.

Complementing these is the NIHR Bioresource, a national network of 13 centres and 150,000 volunteers who have consented to take part in research based on their genomics and/or presented symptoms.

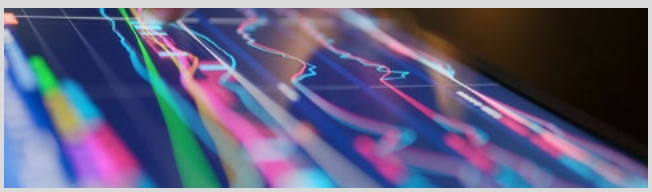
The Wellcome Trust-Sanger Institute is one of the premier centres of genomic discovery and understanding in the world. It leads ambitious collaborations across the globe to provide the foundations for further research and transformative healthcare innovations in genomics.

Competitive landscape

The genomics industry contributed £2.3bn of turnover to the UK economy in 2018/19, up from £1.9bn the year before. We are home to rapidly growing companies such as Oxford Nanopore, a company supported by Genomics England and a strategic partner, is expected to be valued at up to £7bn ahead of its Initial Public Offering. The wider UK genomics sector has approximately 154 companies, over 5000 employees and could have a market cap of over £50bn by 2040, according to a recent BIA report.

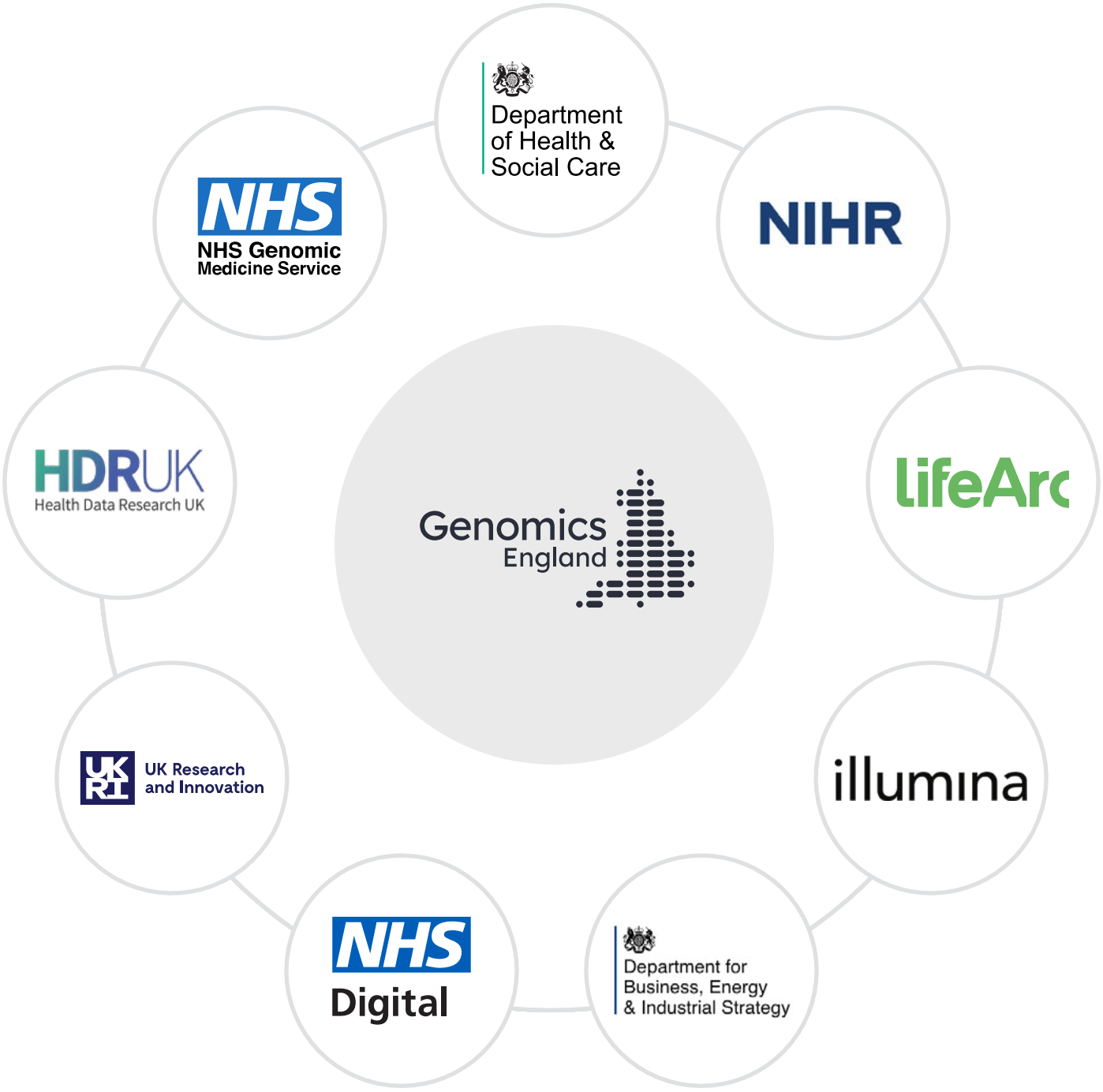
Genomics industry

This is in the context of a set of international competitors who are investing heavily: The US has launched All of US to sequence 1 million genomes, whilst Japan launched its own 100k genome project in 2019. The US Genomics sector was recently estimated to have a total economic impact of $265bn (source), with significant investment from industry to access genomic data and capabilities: GSK invested $300m in a collaboration with 23&me on drug development and share data (source: Bloomberg).



Partners and funders

We work together with our partners and funders to build together an ecosystem which delivers genomic healthcare for patients and accelerates genomic research



Partner profiles

Professor Dame Sue Hill

Chief Scientific Officer and Senior Responsible Officer for Genomics, **NHS England and NHS Improvement**

Professor Dame Sue Hill OBE PhD DSC CBiol FRSB Hon FRCP Hon FRCPath is the Chief Scientific Officer for England – providing expert clinical scientific advice across the health system and head of profession for the healthcare science workforce in the NHS and associated bodies – embracing more than 50 separate scientific specialisms. Sue is the Senior Responsible Officer for the Genomics programme in NHS England and NHS Improvement, leading the transformation of the NHS GMS, having previously established the NHS Genomic Medicine Centres and led the NHS contribution to the 100,000 Genomes Project. She is a respiratory scientist by background with an international academic and clinical research reputation.

Dr. Melanie Lee

CEO, **LifArc**

Dr Melanie Lee, PhD, CBE is the CEO of LifeArc. Prior to joining LifeArc Melanie pursued a 30 year career in healthcare R&D and gained leadership experience both from the biopharmaceutical industry and from the medical research charity sector. Melanie's previous roles have included Chief Scientific Officer at BTG (most recently) as well as senior positions at GSK, Celltech and UCB. She has previously held Trustee appointments at Cancer Research Technology and Cancer Research UK and currently serves on the Board of Directors of Sanofi.

Professor Dame Ottoline Leyser

Executive Chair, Medical Research Council, **UKRI**

UKRI brings together the UK's Research Councils, Innovate UK and Research England, operating with a combined budget of more than £8bn per year.
Ottoline Leyser was Director of the Sainsbury Laboratory, University of Cambridge, an interdisciplinary research institute combining computational modelling with molecular genetics and cell biology to elucidate the dynamical systems underpinning the control of plant growth and development. Ottoline is a Fellow of the Royal Society, a Member of the Leopoldina and EMBO, and an International Member of the US National Academy of Sciences. In 2017 she was appointed DBE for services to plant science, science in society and equality and diversity in science.

Dr. Louise Wood

Director of Science, Research and Evidence, Dept of Health, **NIHR**

Dr Louise Wood CBE is Director of Science, Research and Evidence at the Department of Health and Social Care (DHSC). She has lead the NIHR together with Professor Chris Whitty since 2016. Louise has an honours degree in Physiology from the University of Edinburgh and a PhD in biomedical science from the University of London. Since 2018, Louise has served as a Council Member for the Medical Research Council. She was elected an honorary fellow of the Faculty of Pharmaceutical Medicine in 2011 and awarded a CBE for services to health research in the 2019 Queen's Birthday Honours.

Partner profiles continued

Rosalind Campion

Director, Office for, Life Sciences at Department for **Business, Energy and Industrial Strategy (BEIS)**

Roz started her career as a lawyer working in the City. Her Government career has split between a focus on social policy issues and on international economic and trade issues. This experience includes stints in Washington DC and in Tokyo for the Foreign Office, as well as Director roles in the Department for International Trade (where she was responsible for the UK's global bilateral trade relations), in Cabinet Office (where she was responsible for EU Exit Implementation), and in the Home Office as the Home Secretary's Principal Private Secretary and Strategy Director. Roz was also a member of the ethics committee of Guy's and St Thomas's and is a Fellow at the Centre for Science and Policy at the University of Cambridge.

Francis deSouza

Chief Executive Officer, **illumina**

Francis deSouza was appointed CEO of Illumina in 2016 and is responsible for directing all aspects of company strategy, planning, and operations. He initially joined the company as President in 2013, and led Illumina's business units and core functions responsible for envisioning, developing and producing the company's products. Francis deSouza received a BS and MS in Electrical Engineering and Computer Science from the Massachusetts Institute of Technology.

Professor Andrew Morris

Chief Executive Officer, **HDRUK**

Professor Andrew Morris became the inaugural Director of Health Data Research UK in August 2017. He is seconded from his position as Professor of Medicine, and Vice Principal of Data Science at the University of Edinburgh, having taken up position in August 2014. Prior to this Andrew was Dean of Medicine at the University of Dundee. Andrew was Chief Scientist at the Scottish Government Health Directorate (2012-2017) and has served and chaired numerous national and international grant committees and Governmental bodies.

His research interests span informatics and chronic diseases. He has published over 330 original papers, attracted over £50 million in grant funding.

Laura Wade-Gery

Chair of the NHS Digital Board, as well as Chair of the Talent, Remuneration and Management Committee, **NHSD**

Laura is Chair of the NHS Digital Board, as well as Chair of the Talent, Remuneration and Management Committee. Laura is also a Non-Executive Director of NHS England. She joined Marks & Spencer Group in 2011 and was Executive Director, Multi-Channel, responsible for stores and online until 2016. She was CEO of Tesco.com from 2003 to 2011, and previously held several senior roles at Tesco, having joined them in 1997. Laura is currently a Non-Executive Director of British Land PLC (and chair of their Remuneration Committee). She is a trustee of Britten Pears Arts.

Collaboration with startups

Congenica
Enabling genomic medicine

Digital heath company

Congenica has created their Clinical Decision Support platform to allow the rapid analysis and interpretation of genomic data in genomic medicine and research. They work towards shortening diagnostic odysseys by providing appropriate genomic information for more people, faster, and with higher laboratory analytical yields and reduced costs. Some of their partners include the NHS, New York State and Digital China Health; their platform has been selected for research use by the NHS GMS.

lifebit

Precision medical technology

Lifebit provides an operating system platform to analyse distributed and previously inaccessible genomics data, through federated analysis and automation. They use both open-source and custom tools to work with clients to transform their datasets into the form required for rapid, collaborative and integrated analysis – their platform's solutions are driven by machine learning and AI. Their framework supports a variety of public and private data sources, including EHR records, multi-omics data, clinical studies, among others.

Sano

Genomics platform

Sano provides access to free genome sequencing and analysis for research participants; their participants' data is then anonymised, aggregated and offered to researchers, along with tools for data analysis. Sano helps researchers recruit for their research studies and clinical trials based on the data that Sano's users provide and they work with leading life sciences companies to deliver patient-centric and data-driven matchmaking into clinical research. Their partners include Imperial College London, the University of Cambridge and The University of Manchester.

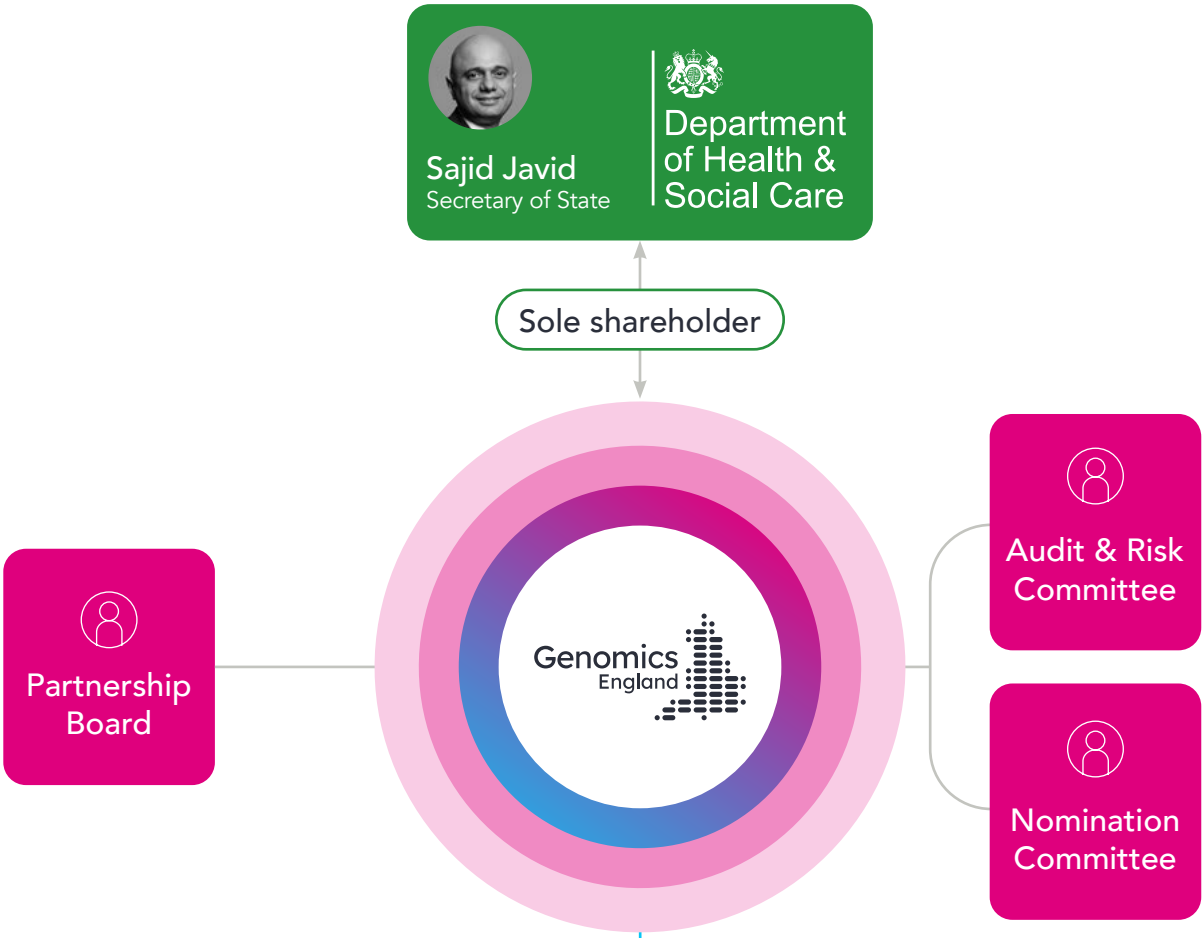
zetta

Genomic data management

Zetta is a leader in large-scale genomic data management. They employ distributed database technology and best practice security techniques to offer clinical laboratories, and research organisations, clinically-validated genome data software architecture. Zetta's OpenCB and Xetabase technologies allow storage, analysis and visualisation of genomic data. Their partners include Microsoft, the University of Cambridge and Mongo DB.

Governance

Company structure



Ethics Advisory Committee

Ensures that ethics are at the heart of everything Genomics England does. Chaired by Professor Mike Parker.

Access Review Committee

Ensures that those who access the data are appropriate and that access is for authorised purposes. Led by an independent chair.

Technology Advisory Group

Focuses on making sure our tech processors are robust and that we resist security threats. Chaired by Andrew Eland.

Participant Panel

Puts the participants at the heart of Genomics England's decision making. Led by an independent chair – Jillian Hastings Ward.

Science Committee

Makes sure that we follow the science. Chaired by Professor Sir John Bell.

GeCIP Board

The GeCIP Board oversees the operation of academic research projects and includes representatives from funders, lead researchers and patients representatives.

Participant panel

The Participant Panel sits at the heart of Genomics England and is made up of participants from the 100,000 Genomes Project, and parents or carers of people involved in this project.

Members of the Panel with the Genomics England and NHS England team.

It is expanding to include patients and relatives from the GenOMICC COVID-19 study and NHS patients who give consent for their whole genome sequences and associated health data to be used for research in the Genomics England data bank.

The Panel acts as an advisory body to the Genomics England Board, working to ensure that the health data held by Genomics England is being looked after with respect and used in the best interests of the participants.

Established in 2016, the Panel meets four times a year and invites senior staff from Genomics England and NHS England to discuss what they are doing with our data.

It advises them how they can continue to ensure that participants' interests are at the heart of their key decisions. Panel members are also often asked for their experience of genomic services and advice on future design. Work regularly continues between these Panel meetings.
The Panel is also actively engaged in decision-making across the committees and boards of Genomics England. Its members sit on the Access Review Committee, the Ethics Advisory Committee and the GeCIP Board.

The Panel Chair is Jillian Hastings Ward and the Vice Chair is Rebecca Middleton.

The Panel currently has vacancies for patients and their relatives involved in the GenOMICC COVID-19 study and for patients (or parents of children) being treated by the NHS GMS. We are especially keen to hear from men, and from people from Black and other minority ethnic communities. If you would be interested in applying, please email info@genomicsengland.co.uk with 'Participant Panel' as the message title.

> The Panel currently has vacancies for patients and their relatives involved in the GenOMICC COVID-19 study and for patients

Access Review Committee

The Access Review Committee (ARC) is an independent advisory body for the 100,000 Genomes Project. It examines requests for access to data in the project.

The ARC approves, declines, or amends requests for access to data in the 100,000 Genomes Project. Decisions take into account many factors including the nature of the research and the risk of identifiability. External sources of expertise may be called to help the decision-making process.

Our Board

Our Board

Baroness Nicola Blackwood
Chair

Nicola became Chair of Genomics England in May 2020. As Minister for Innovation in the Department for Health and Social Care, her responsibilities included research, life sciences, NHS innovation, data, digital and technology, and rare diseases. Today, she is a member of the House of Lords Science and Technology Select Committee and is an Honorary Professor of Science and Public Policy at UCL.

Born in Johannesburg in 1979 to a South African nurse and an English cardiologist, her parents worked for nearly 30 years in the NHS giving her first-hand knowledge of some of the challenges facing the health service.

Key roles
- Elected MP for Oxford West and Abingdon in 2010
- Minister for Innovation in the Department for Health and Social Care
- Chairman Science & Technology Select Committee
- Chair of the Human Tissue Authority
- Board Member of Oxford University Innovations' Advisory Board

Chris Wigley
Chief Executive Officer

Prior to joining Genomics England, Chris was COO at QuantumBlack, a bespoke machine learning and AI technology company, and a Partner at McKinsey working on technology strategy topics.

Chris has also worked for the UK Foreign Office (Counter Terrorism Policy Department) and at the BBC. He has held board roles at the New Entrepreneurs Foundation, Entrepreneur First, and Magic Breakfast.

Key roles
- COO, QuantumBlack
- Partner, McKinsey & Company
- Diplomat, Foreign and Commonwealth Office

Professor Matthew Brown
Chief Scientific Officer (CSO)

Trained as a clinician-scientist and a rheumatologist, Matthew has made contributions to the development of gene-mapping approaches in human diseases and genome-wide association study methodology, leading to the discovery of thousands of genetic variants, including genes responsible for monogenic forms of arthritis, ectopic bone development, and skeletal dysplasias. He has also led efforts in Australia to translate research sequencing capability into precision medicine programmes for cancer patients.

Key roles
- Director, Guy's and St Thomas' NHS Foundation Trust and King's College London NIHR BRC
- Director of Genomics, Queensland University of Technology (QUT)
- Director, University of Queensland, Diamantina Institute.

Our Board

Sir Jonathan Symonds CBE
Non-Executive Director

As Non-Executive Director, Sir Jon provides the Executive Leadership Team with guidance and general counsel on strategic direction, performance monitoring, and communication with both the scientific community and the general public.

Sir Jon has held several leadership and governance positions within financial and life sciences organisations such as HSBC Bank plc, Novartis AG, Goldman Sachs, and AstraZeneca.

Key roles
- Chairman, GSK
- Senior Independent Director, HSBC Holdings plc
- Chairman, Proteus Digital Health
- Non-Executive Director, Rubius Therapeutics
- Non-Executive Director and Chair of the Audit Committee, Diageo plc and QinetiQ Group plc
- Fellow, Institute of Chartered Accountants in England and Wales.

Professor Sir John Bell
Non-Executive Director

Professor Sir John Bell GBE, FRS is Regius Professor of Medicine at Oxford University. His research interests are in the area of autoimmune disease and immunology where he has contributed to the understanding of immune activation in a range of autoimmune diseases. In 2001, he was appointed non-executive director of Roche Holding AG and in 2008 he joined the Gates Foundation Global Health Advisory Board which he has chaired since 2012. He is Chair of the Rhodes Trust. In December 2011, Sir John was appointed one of two UK Life Sciences Champions by the Prime Minister. He sits on the board of Genomics England Limited and chairs its Science Advisory Committee. He was appointed Knight Grand Cross of the Order of the British Empire (GBE) in the 2015 New Year Honours for services to medicine, medical research and the life science industry. Sir John has held prominent roles during the Covid epidemic, enabling the development of the testing platforms for LFTs and helping to initiate the PCR programme nationally as well as helping to manage the relationship with AstraZeneca that produced the Oxford AZ vaccine.

Key roles
- Founder, Wellcome Trust Centre for Human Genetics
- President, Academy of Medical Sciences (2006-2011)
- Chairman, Office for the Strategic Coordination of Health Research (until 2017)

Professor Ewan Birney
Non-Executive Director

Professor Ewan Birney is a Director of the European Bioinformatics Institute (EMBL-EBI) and a Senior Scientist at the European Molecular Biology Laboratory. He completed his PhD at the Wellcome Trust Sanger Institute with Richard Durbin, and worked in the laboratories of leading scientists Adrian Krainer, Toby Gibson and Iain Campbell.

Ewan also led the analysis group for the ENCODE project, and his current areas of research include functional genomics, assembly algorithms, statistical methods to analyse genomic information, and compression of sequence information.

Key roles
- 2003 Francis Crick Award
- 2005 Benjamin Franklin Award
- Fellow, Royal Society
- Fellow, Academy of Medical Sciences
- Honorary Professor of Bioinformatics, Cambridge University School of Clinical Medicine

Our Board

Professor Michael Parker

Non-Executive Director; Chair of the Ethics Advisory Committee

Michael has been a member of the Genomics England Board and Chair of the Ethics Advisory Committee since the beginning of the project in 2013. Prior to this he chaired the Chief Medical Officer's ethics advisory group feeding into its design.

Michael is Professor of Bioethics and Director of the Ethox Centre and the Wellcome Centre for Ethics and Humanities at the University of Oxford. His research focuses on two topics: the ethical aspects of genomics and data-science at the interface between research and clinical practice; and, the ethics of collaborative global health research on infectious disease.

Key roles

- Chair of Ethics Advisory Board at Our Future Health
- Ethics Advisor to UK Biobank
- Member of Moral and Ethical Advisory Group, Department of Health and Social Care
- Participant UK Scientific Advisory Group for Emergencies (SAGE)

Dr. Keith Stewart

Non-Executive Director

Keith has served in several healthcare leadership roles across both research and clinical practice in Toronto and at the Mayo Clinic, where he was most recently Director of the Center for Individualized Medicine. His research and clinical practice is focused on the biology, genomics and treatment of Multiple Myeloma, and more broadly on the application of genomics in health care. He has published over 350 research papers and led numerous clinical trials of novel therapeutics for this disease from first in man to large practice changing studies.

Key roles

- Vice President, Cancer at University Health Network, Toronto
- Director, Princess Margaret Cancer Program Centre, Toronto
- Regional Vice-President, Ontario Health (Cancer Care Ontario)
- Richard H. Clark Chair in Cancer Medicine
- Richard H. Clark Chairin Cancer Medicine

Rosalind Campion

Non-Executive Director

Rosalind Campion became the Director of the Office for Life Sciences in November 2021 and, as such, holds the role of Shareholder Director, representing the interests of the Secretary of State for Health and Social Care, on the GEL board. Roz started her career as a lawyer working in the City. Her Government career has split between a focus on social policy issues and on international economic and trade issues. This experience includes stints in Washington DC and in Tokyo for the Foreign Office, as well as Director roles in the Department for International Trade (where she was responsible for the UK's global bilateral trade relations), in Cabinet Office (where she was responsible for EU Exit Implementation), and in the Home Office as the Home Secretary's Principal Private Secretary and Strategy Director. Roz was also a member of the ethics committee of Guy's and St Thomas's and is a Fellow at the Centre for Science and Policy at the University of Cambridge.

Key roles

- PPS to the Home Secretary and Strategy
- Director, Home Office
- Director for EU Exit Implementation, Cabinet Office
- International Strategy Director, Department for International Trade
- Director of Economic Issues and Strategy, British Embassy Tokyo
- Global Issues Counsellor, British Embassy Washington DC

Our Board

Lord David Prior
Non-Executive Director

Lord David Prior is Chair of the NHS England Board, Board Member of WWG advisory, Chairman of Quality by Randomisation Board and a Deputy Chairman at Lazard.

Key roles
- MP for North Norfolk (elected 1997)
- Chairman, Norfolk and Norwich University Hospitals NHS Foundation Trust (2002-2012)
- Chairman, Care Quality Commission
- Parliamentary Under Secretary of State for Health
- Parliamentary Under Secretary of State, Department of Business, Energy and Industrial Strategy

Dr. Annalisa Jenkins
Non-Executive Director

Annalisa Jenkins, M.B.B.S., F.R.C.P., is a biopharma thought leader with over 25 years of industry experience. She is independent board director of several public and private life science companies, and Chair of Board of Trustees YouBelong programming mental health services in Sub Saharan Africa. She has extensive experience in building and financing biotech companies pursuing cures for rare diseases.

Annalisa is an advocate for diversity and inclusion, particularly for women in science.

Key roles
- President and CEO, Dimension Therapeutics
- Head of global research and development, Merck Serono
- Executive Vice President, Global Development and Medical, Merck Serono
- Senior Vice President, Bristol Myers-Squibb
- Head of Global Medical Affairs, Bristol Myers-Squibb
- Committee Member, Science Board, U.S. Food & Drug Administration
- Board Member, Faster Cures, a Center of The Milken Institute
- Chair of The Court, London School of Hygiene and Tropical Medicine
- Founder and CEO annalisajenkinsllc
- Medical Trustee British Heart Foundation
- Trustee The Kings Fund

Andrew Eland
Non-Executive Director

Andrew's main interest is building software tools to help people understand, and responsibly evolve, complex systems. He's worked on technology products for around 20 years, and currently focuses on the urban environment at Diagonal, a company he founded to address challenges in the domain. Previously, Andrew led the global engineering organisations behind AI pioneer DeepMind's work in healthcare, Google Maps Mobile, and Google's philanthropic efforts.

Key roles
- Founder, Diagonal
- Engineering Director, DeepMind
- Engineering Director, Google
- Software Engineer, BBC
- Advisory Board Member, i-Sense

Our Board

Vikram Bajaj
Non-Executive Director

Vikram Bajaj is a Managing Director at Foresite Capital, a healthcare-focused Venture Capital firm, specifically focusing on the intersection between technology and life sciences, to include personalised and precision healthcare. His research interests are in physical sciences, engineering, and life sciences.

Key roles
- Managing Director, Foresite Capital
- Chief Scientific Officer, GRAIL
- Co-Founder & Chief Scientific Officer, Verily (formerly Google Life Sciences)
- Associate Professor, Stanford School of Medicine
- Affiliate Scientist, Berkley National Laboratory and University of California, Berkley
- Advisor to the U.S. Department of Defense, Defense Science Board's Task Force on Biology

Nick Maltby
Secretariat

Nick joined Genomics England in 2013 following a career in private practice with several large international law firms. He is an experienced and award-winning commercial and procurement lawyer and has received several accolades for his work at Genomics England. He is responsible for all legal activity within Genomics England including negotiating contracts, public procurement, data protection, freedom of information, regulatory compliance, and the Company's Articles and policies.

Key roles
- Partner, Pinsent Masons
- Partner, DLA Piper
- Partner, Bircham Dyson Bell

Executive leadership team

Anna Tomlinson

Chief Communications & Engagement Officer

Anna joined Genomics England in 2019, with more than 20 years in communications at global and national levels. She had led teams across a number of sectors, including healthcare and professional services, and is now responsible for overseeing all internal and external-facing communications, reputation management and engagement strategies at Genomics England

Key roles

- Global Communications Director, Aetna
- Group Marketing and Communications Director, InHealth Group

Dr. Augusto Rendon

Chief Bioinformatician

Augusto leads over 70 developers, analysts, bioinformaticians and curators at Genomics England. He and his team have designed and implemented the clinical interpretation pipelines that return findings to patients, while ensuring that knowledge accumulated through this process is best used to improve patient care and enhance discovery.

Catherine Byers

Chief Transformation Officer

Prior to joining Genomics England, Catherine spent 16 years across the Department of Health and Social Care, the Office of Lifesciences, the Cabinet Office, and with No. 10. Most recently, she was Head of NHS Finance at the Department of Health and Social Care. As Chief Transformation Officer, Catherine leads the finance, estates, quality, and performance chapters at Genomics England.

Key roles

- Deputy Director, COVID-19 National Testing Programme, Dept. of Health and Social Care
- Head of NHS Finance, Dept. of Health and Social Care

Chris Wigley

CEO

Prior to joining Genomics England, Chris was COO at QuantumBlack, a bespoke machine learning and AI technology company, and a Partner at McKinsey working on technology strategy topics.

Chris has also worked for the UK Foreign Office (Counter Terrorism Policy Department) and at the BBC. He has held board roles at the New Entrepreneurs Foundation, Entrepreneur First, and Magic Breakfast.

Executive leadership team

Jackie Kinsey

Chief People Officer

Jackie Kinsey is the Chief People Officer for Genomics England. Jackie has over 30 years' experience in HR across multiple industries and organisations including Thomas Cook, BAA Heathrow and Fiserv. Prior to Genomics England, Jackie was Chief People Officer for ThoughtWorks a global digital transformation technical consultancy. Jackie is a Trustee for PohWer a charity which provides information, advice, support and advocacy to people who experience disability, vulnerability, distress and social exclusion. She is co-founder of a Team Coaching Practise. Jackie lives in Essex with her husband Toby and two children.

Professor Matthew Brown

Chief Scientific Officer (CSO)

Trained as a clinician-scientist and a rheumatologist, Matthew has made contributions to the development of gene-mapping approaches in human diseases and genome-wide association study methodology, leading to the discovery of thousands of genetic variants, including genes responsible for monogenic forms of arthritis, ectopic bone development, and skeletal dysplasias. He has also led efforts in Australia to translate research sequencing capability into precision medicine programmes for cancer patients.

Key roles

- Director, Guy's and St Thomas' NHS Foundation Trust and King's College London NIHR BRC
- Director of Genomics, Queensland University of Technology (QUT)
- Director, University of Queensland, Diamantina Institute.

Nick Maltby

General Counsel and Company Secretary

Nick is Genomics England's General Counsel and Company Secretary, and the Board's Secretariat.

He is responsible for all legal activity within Genomics England, and for signing off all procurement and contracting work. Prior to his role at Genomics England, Nick was a partner in three Top 100 law firms: Pinsent Masons, DLA Piper and Bircham Dyson Bell.

Parker Moss

Chief Ecosystems and Partnership Officer

Parker Moss is Chief Ecosystems and Partnership Officer for Genomics England, a Department of Health organisation focused on driving Genomic Medicine into the NHS, and accelerating Genomic research in industry and academia. Parker is responsible for the organisation's strategic relationships in the Biopharma Sector, the NHS and Academia. Previously Parker served as a member of the executive team at Owkin, an AI/ML Cancer Research company focused on federated learning. Parker was a CTO in the NHS, including technology leadership roles at Great Ormond Street and Virgin Care. He worked at F-Prime and EightRoads, Fidelity-backed VC funds and led the investment in Owkin before joining the company. Parker also holds several non-executive roles, including Membership on the Secretary of State's technology advisory board, and member of Cancer Research UK's commercial board. Parker holds a degree in Physics and Philosophy from Durham University. Outside of work his passion is classical music.

Executive leadership team

Pete Sinden

Chief Information Officer

Pete joined Genomics England in January 2020 with significant experience in building data-driven information systems used across the NHS and other industry sectors.

Key roles

- Chief Digital Officer, NHS Improvement, NHS
- Chief Digital Officer, Care Quality Commission
- Chief Information Officer, Monitor

Rakhi Rajani

Chief Digital Officer

Rakhi has held senior innovation positions at startups and large organisations spanning emerging tech, healthcare, aerospace and defence, hospitality, and automotive. Rakhi joined Genomics England as CDO in May 2020 to drive innovation in healthcare products and services that put the patient and society at the core.

Key roles

- Associate Partner, QuantumBlack
- Associate Partner, McKinsey & Company

Dr. Richard Scott

Chief Medical Officer

Richard trained in medicine at Cambridge University and University College London, and joined Genomics England in 2015. He areas of focus are Paediatrics and Clinical Genetics and his practice focuses on diagnosing children with rare multisystem disorders.

Key roles

- Consultant, Great Ormond Street Hospital for Children
- Honorary Senior Lecturer in Clinical Genetics, UCL Institute of Child Health

Genomics England

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