Precision Medicine Transforming Healthcare

Prof Sir Mark Caulfield

Vice Principal for Health

Disclosures

Seconded from Queen Mary to be Chief Scientist for Genomics England a Department of Health and Social Care company 2013-21 – received usual salary

Helped multiple countries with Genomic Projects (unpaid)

UK-France Genomique Programme – 3 m Euros 2016-2021 (Department of Health)

Advised UAE on the Emirates Genome Project (paid consultancy – 2022)

UK-Qatar Genomic Health Alliance (funded by UK Foreign and Commonwealth Office £750K over 3 years)

MRC Council Member (£6K per year) – Chair of Longitudinal Cohorts Review (unpaid 2025)

Non-Executive Director of Barts Health and Barking Havering and Redbridge NHS Trust (£20K/annum from NHS England (2022 onward)

The origin of the 100,000 Genomes Project

The 100,000 Genomes Project in numbers

Over **100,000** genomes

Over **97,000** patients and family members

40 Petabytes of data.
1 Petabyte of music would take 2,000 years to play on an MP3 player.

13 Genomic Medicine Centres, and **98** NHS Trusts within them were involved in recruiting participants

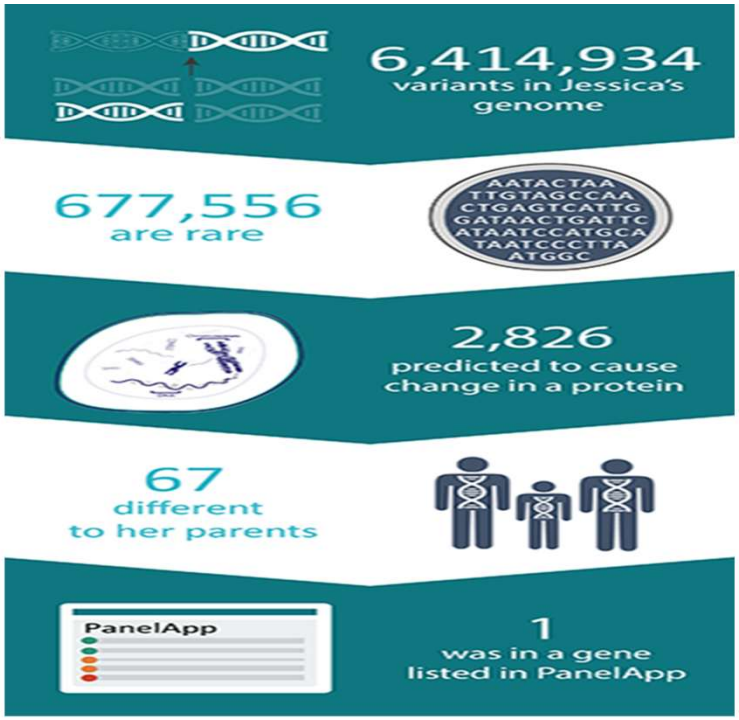
Around **5,000** NHS staff (doctors, nurses, pathologists, laboratory staff, genetic counsellors)

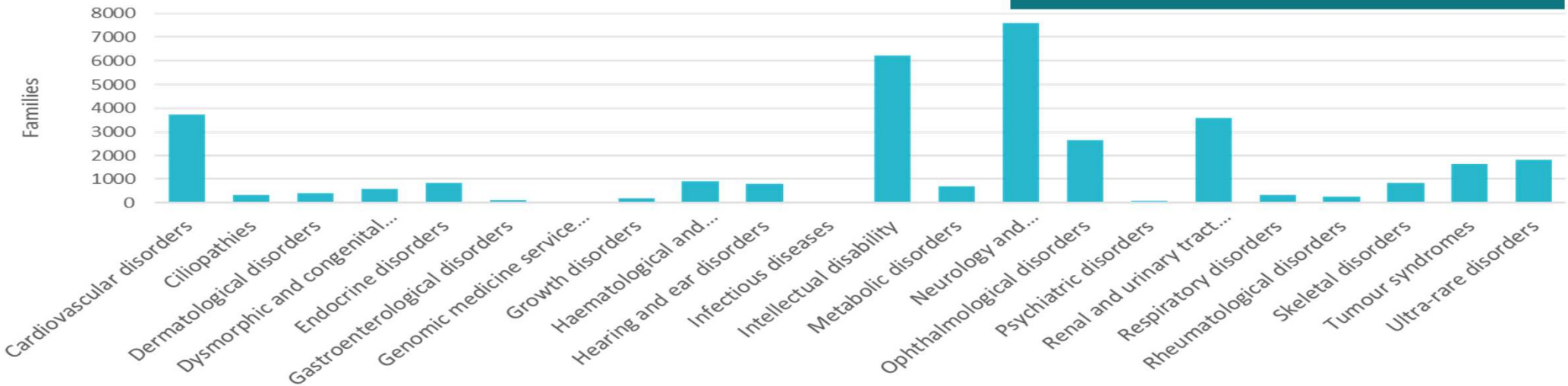
Over **3,000** researchers and trainees

- 20%-25% diagnoses in rare diseases
- 25% influenced cancer care
- Participants heavily engaged
- Trusted Research Environment
  Acts as a reading library
- Analytical pipeline development
  Workforce development
- 400 molecular fresh tissue pathology pipelines/ care pathways
- Available to researchers from 33 countries & industry
  90,178 people
  Multi-billion clinical data-points
  106,000 whole genomes

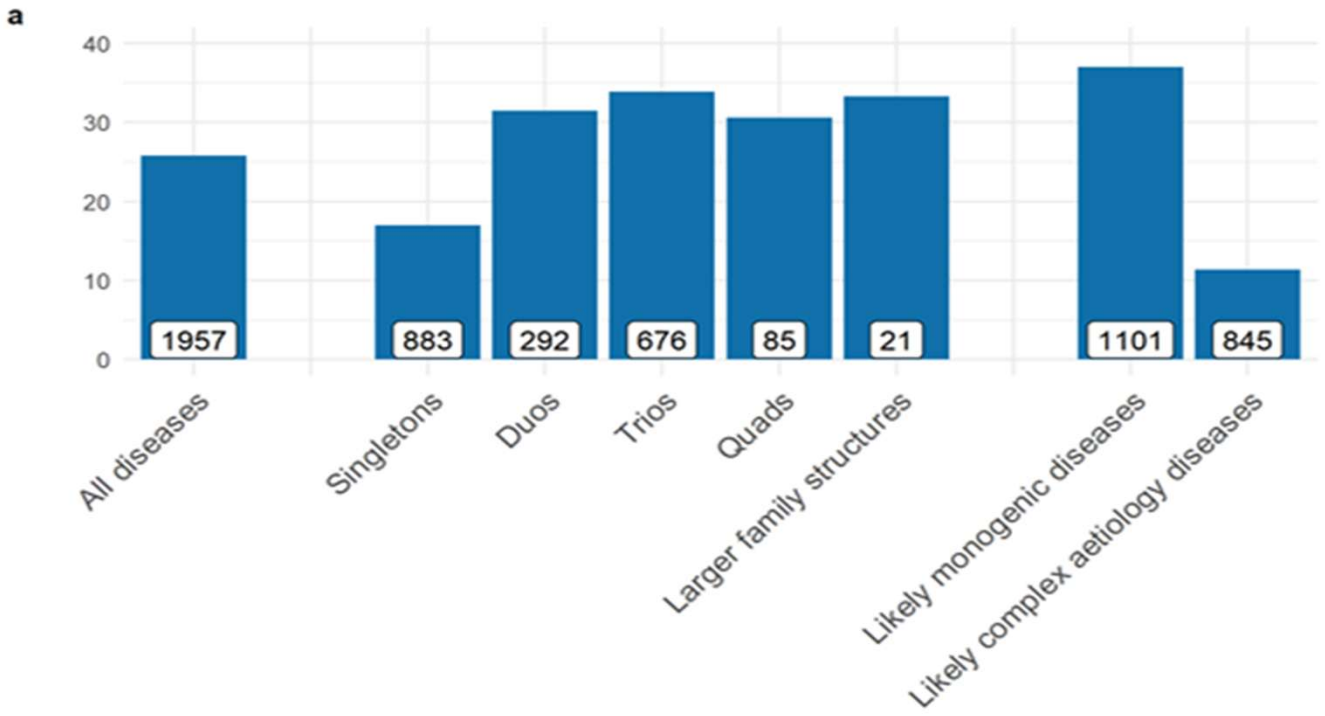
Rare Inherited diseases

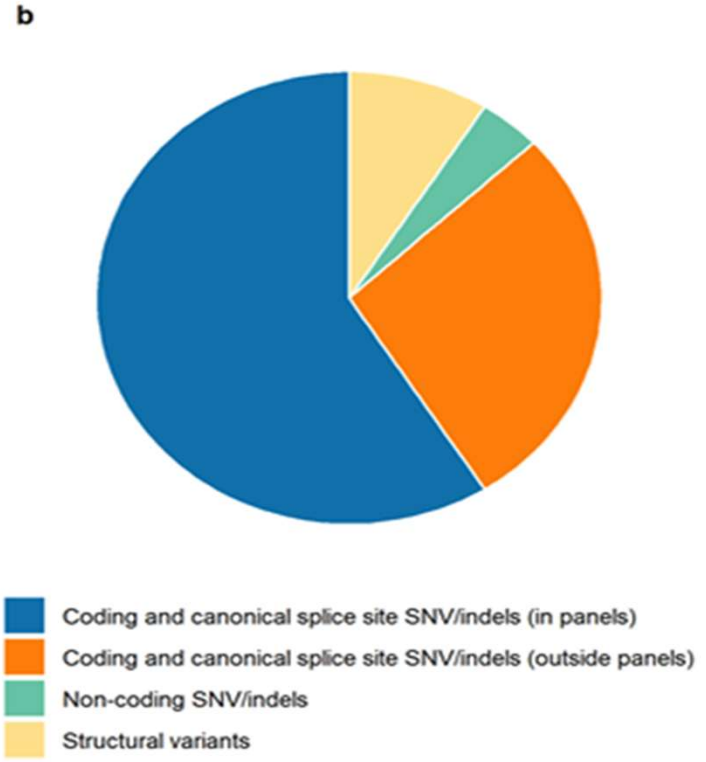
- <6% of the UK population 3 mi
- 3 million people in the UK
- 1200 disorders unmet need
- Standardised eligibility & phenotyping
- Human Phenotyping Ontology
- Automated analytics
- NHS confirm gene panels & close cases





Rare Disease Diagnoses and Family Size
2183 families from 160 rare disease categories in 4660 people





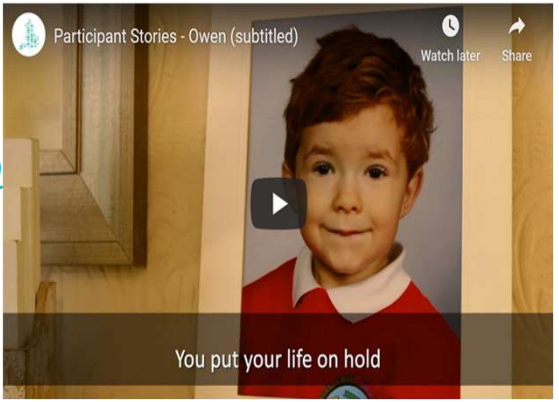
Application in the NHS

10 year old girl admitted to ITU with life threatening chicken pox

- Prior unusual severe infections. Detailed immune testing no diagnosis.
- Mutations in *CTP synthase 1* gene affects B and T lymphocyte responses to infection of both capsulated bacterial infection and viruses
- Curative bone marrow transplant- Siblings tested and not at risk of these infections
- *J Allergy Clin Immunol 2016 Vol: 138: 6*

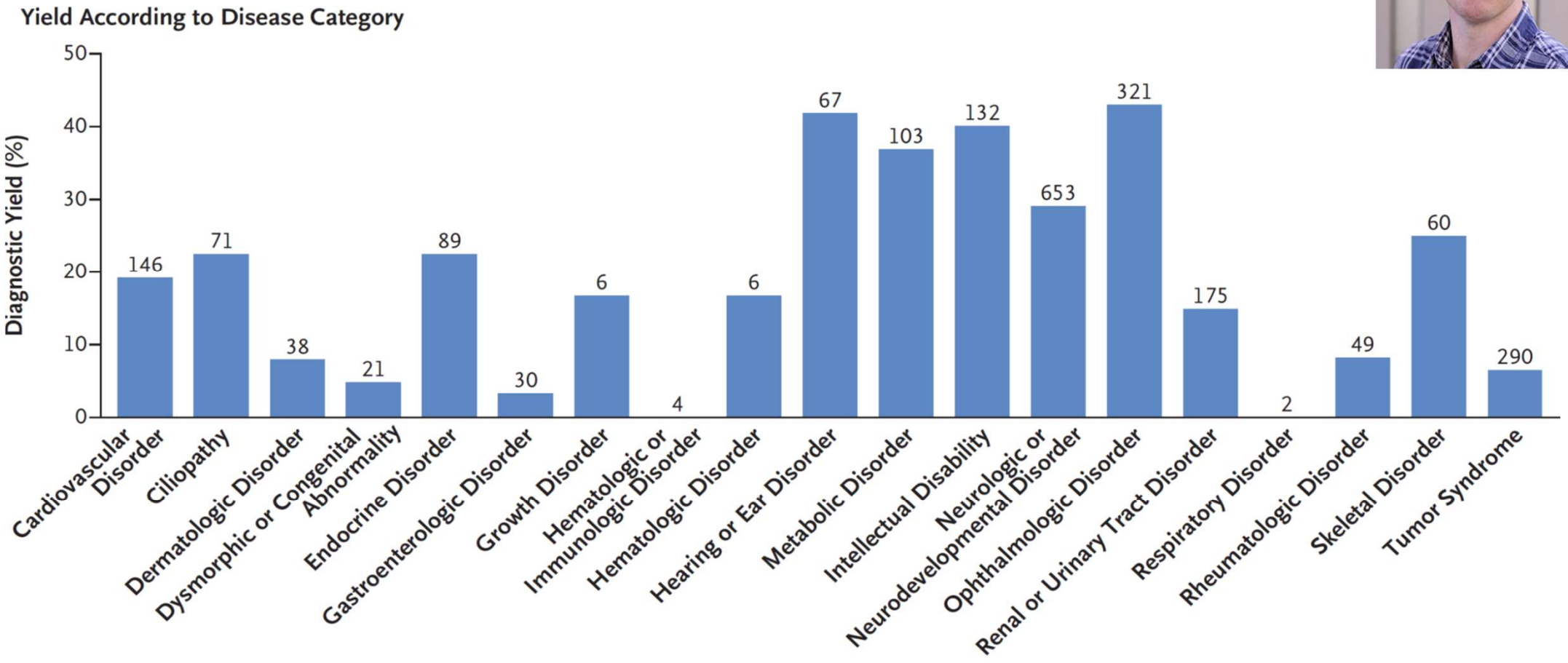
4-year-old with anaemia, developmental delay and short stature

- Initial diagnosis Diamond Blackfan Anaemia
- Trio genome sequencing found a *de novo* mutation in *THRA*, (thyroid hormone receptor alpha)
- Thyroxine dose titrated to metabolic rate, not thyroid function tests;
- growth and general health have improved on treatment
- A further 7 families have now been diagnosed and treated
- https://www.genomicsengland.co.uk/about-genomics-england/participant-sto



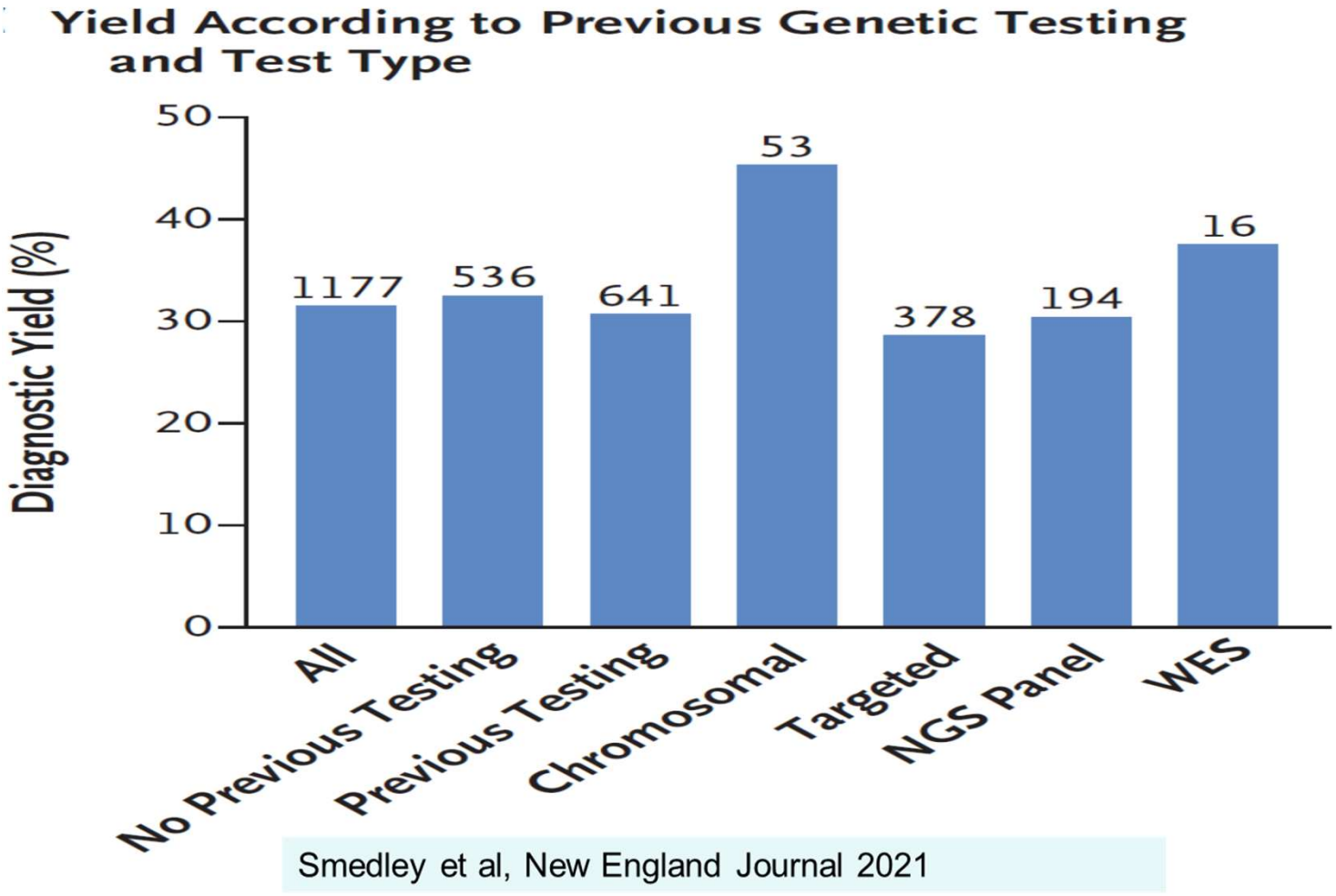
Smedley et al, New England Journal 2021

25% average diagnostic yield



Smedley et al, New England Journal 2021

Diagnostic yield by WGS against no prior testing or prior testing



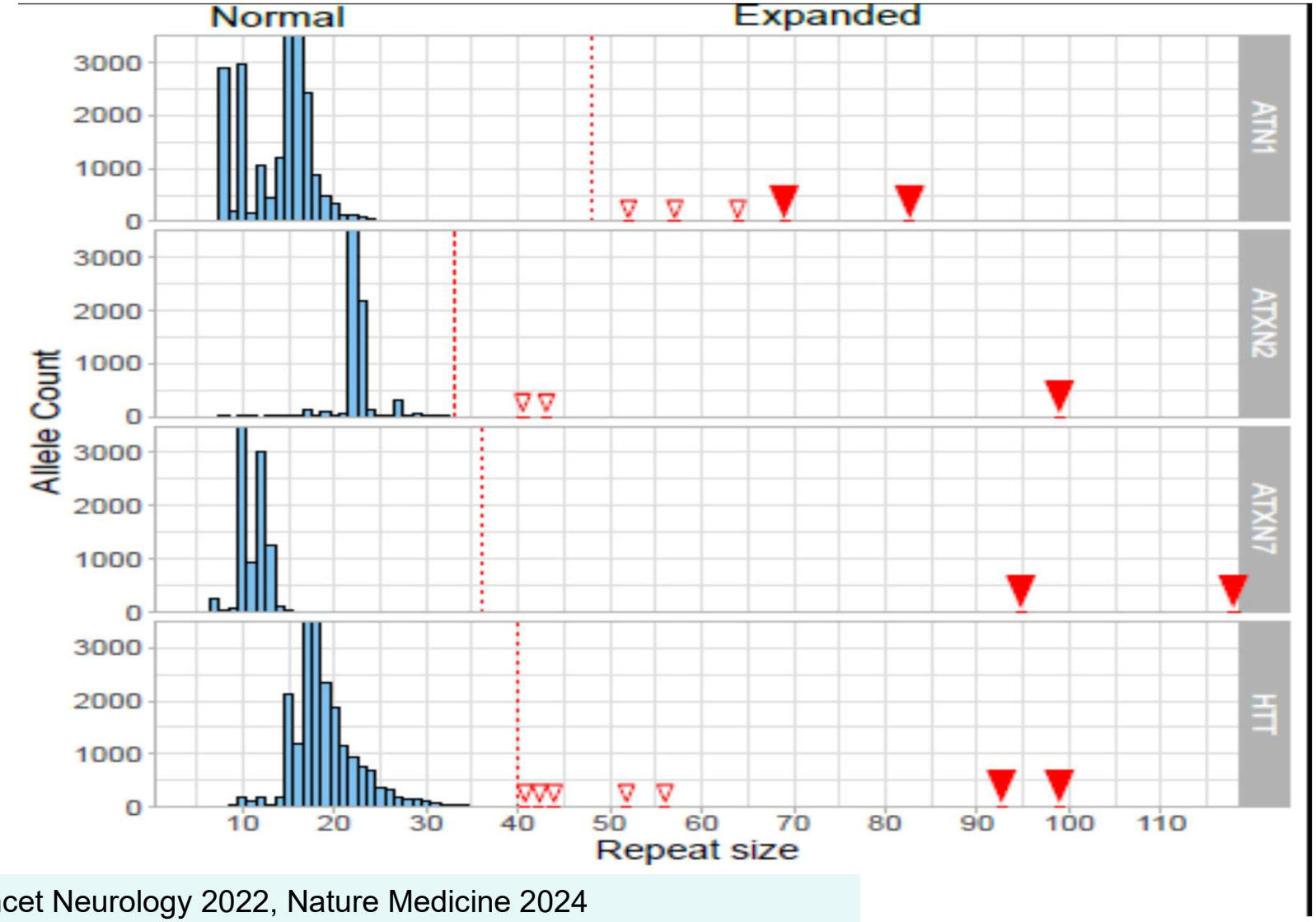
Smedley et al, New England Journal 2021

Diagnostic odyssey of children born 2003 onward

- **Families spent 6 years** (median 75 months) attended a median of 68 hospital appointments prior to diagnosis
- Unaffected relatives attended a median of 18 appointments over 120 months from birth.
- Post-diagnosis over 18 months, fewer focused clinical episodes
- **Affected participants used 183,273** episodes of hospital care via the emergency department, outpatients, inpatients and critical care,
- **Cost £87 million** (median cost of £15,310 per participant)
- **Compared to 53,706 episodes at a cost of £21 million** (median cost of £4,285/participant) for the unaffected participants
- Not including visits to the family physician, or disease treatment costs.

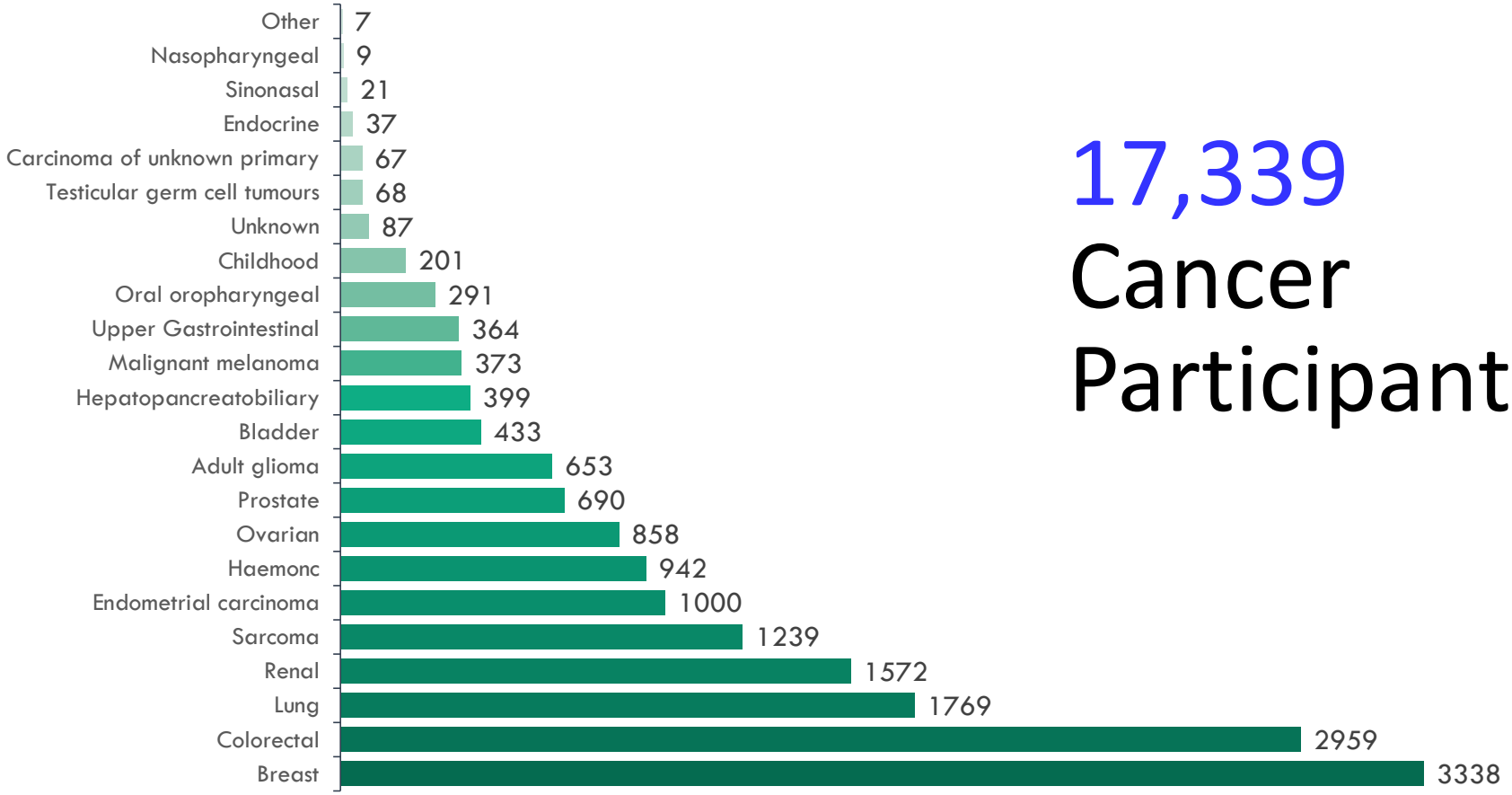
Smedley et al, New England Journal 2021

Expansion Hunter for Repeat Expansion Disorders



Tucci et al Lancet Neurology 2022, Nature Medicine 2024

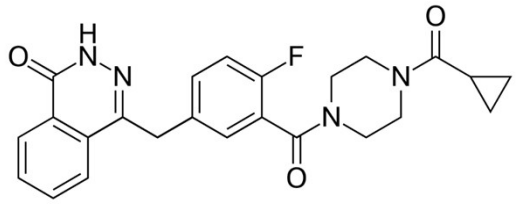
100,000 Genomes Project: Cancer participants
Affects 1 in 2 people



17,339
Cancer
Participants

Cancer Case: Implications of result

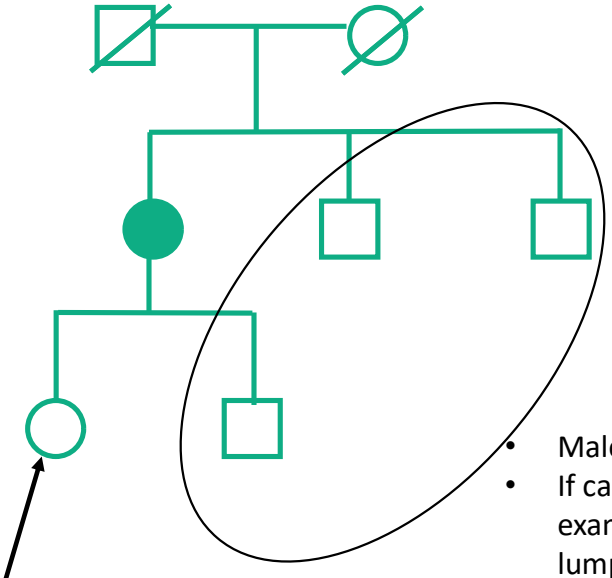
For the patient



- Targeted therapy with Olaparib through clinical trial (OLYMPIA)

- 1-3/10 women develop ovarian cancer
- Offer risk reducing surgery
- 1 in 2 lifetime chance of left sided breast cancer – requires ongoing screening or consideration of risk reducing surgery

For her family



Predictive test
BRCA2 carrier

- Daughter will receive breast screening with scans yearly from age 30
- consider risk reducing surgery
- consider chemoprevention

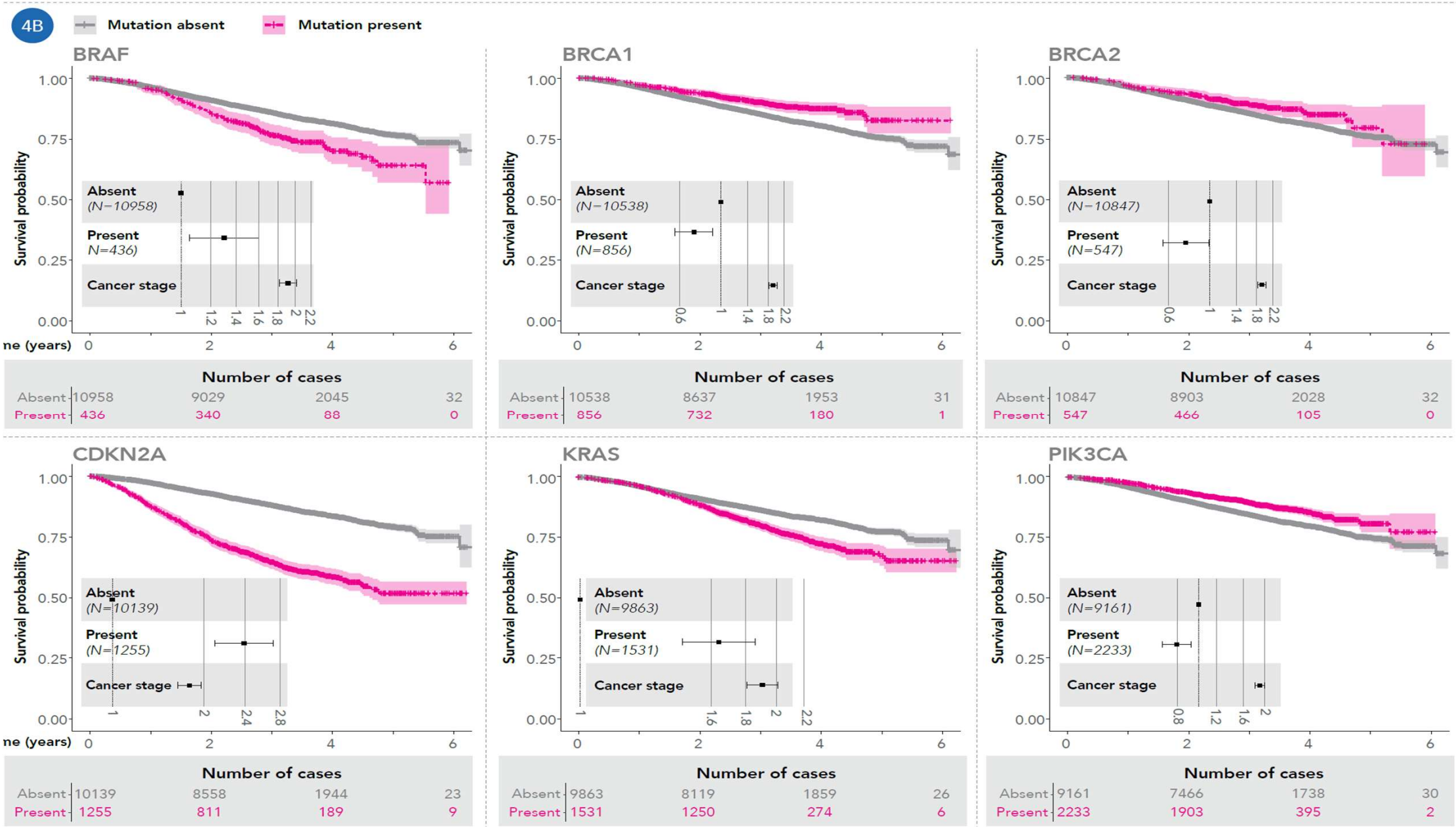
- Males awaiting testing
- If carriers, self examination for breast lumps
- Eligible for prostate cancer screening from age 40

13,380 solid tumours with real-world treatment and outcome data

- UK cancer incidence has increased by approximately 4% over the past 10 years
- Glioblastoma multiforme – Somatic SNVs in 94% and CNVs in at least one gene in 58%
- Mutations in 20-49% of breast invasive carcinoma, ovarian high grade serous carcinoma, uterine endometrial, sarcoma, mesothelioma, bladder urothelial carcinoma and lung squamous cell carcinoma cases
- Sarcoma has the highest occurrence of actionable SVs (13%).
- Homologous recombination deficiency 40% of high-grade serous ovarian cancer cases
- <20% had mutations in pancreatic, prostate, oesophageal and stomach adenocarcinoma
- *PIK3CA* 2nd highest mutated gene in 19.8% of patients with uterine corpus endometrial carcinoma (53.5%), ovarian endometrioid adenocarcinoma (49.0%), breast invasive carcinoma (42.2%), uterine corpus endometrial serous carcinoma (38.1%) and colon adenocarcinoma (26.5%). ? clinical trials needed.
- 30% linked to pathogenic germline variants, highlighting need for combined analysis

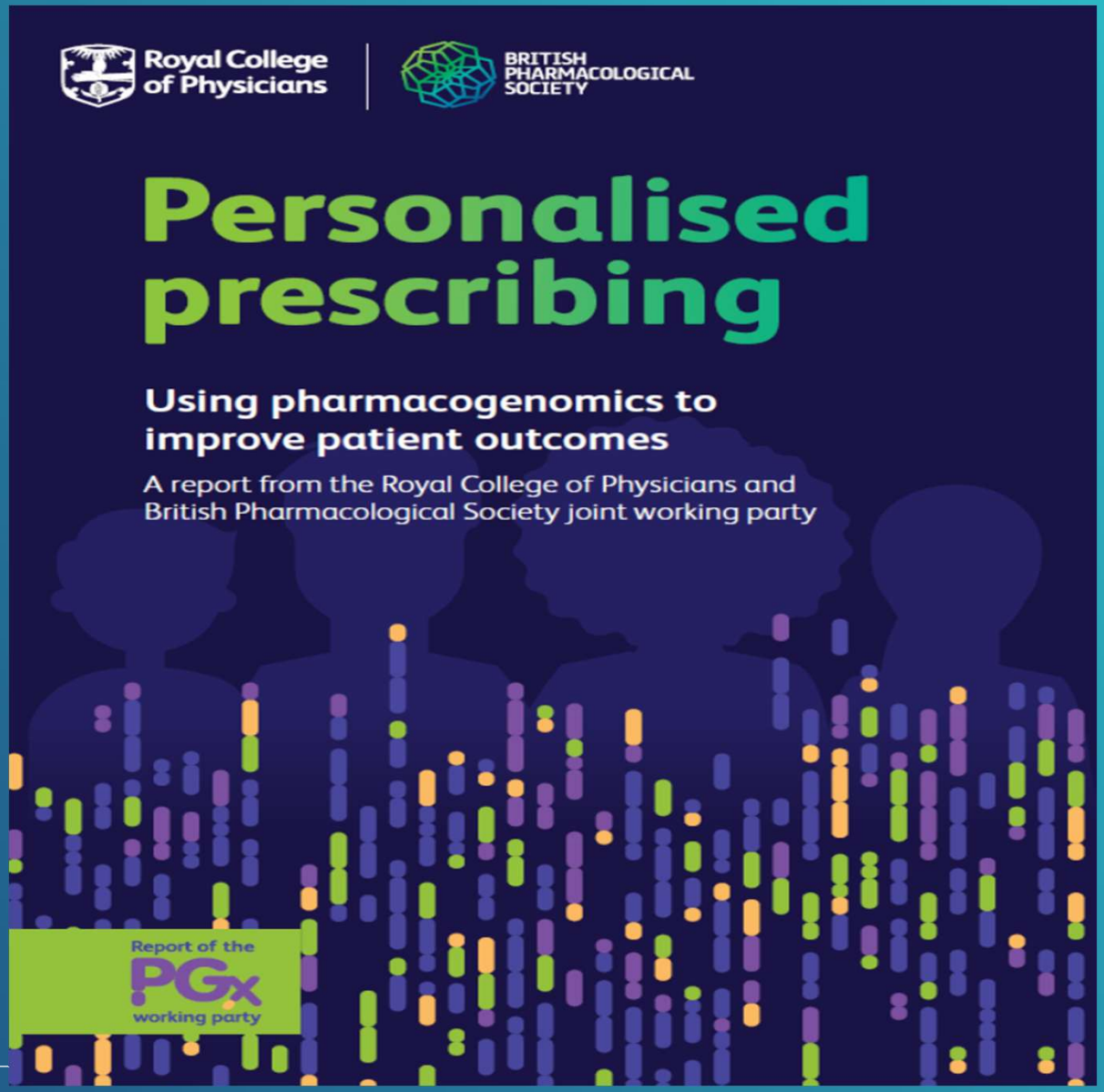
Sosinsky et al, Nature Medicine January 2024

4B

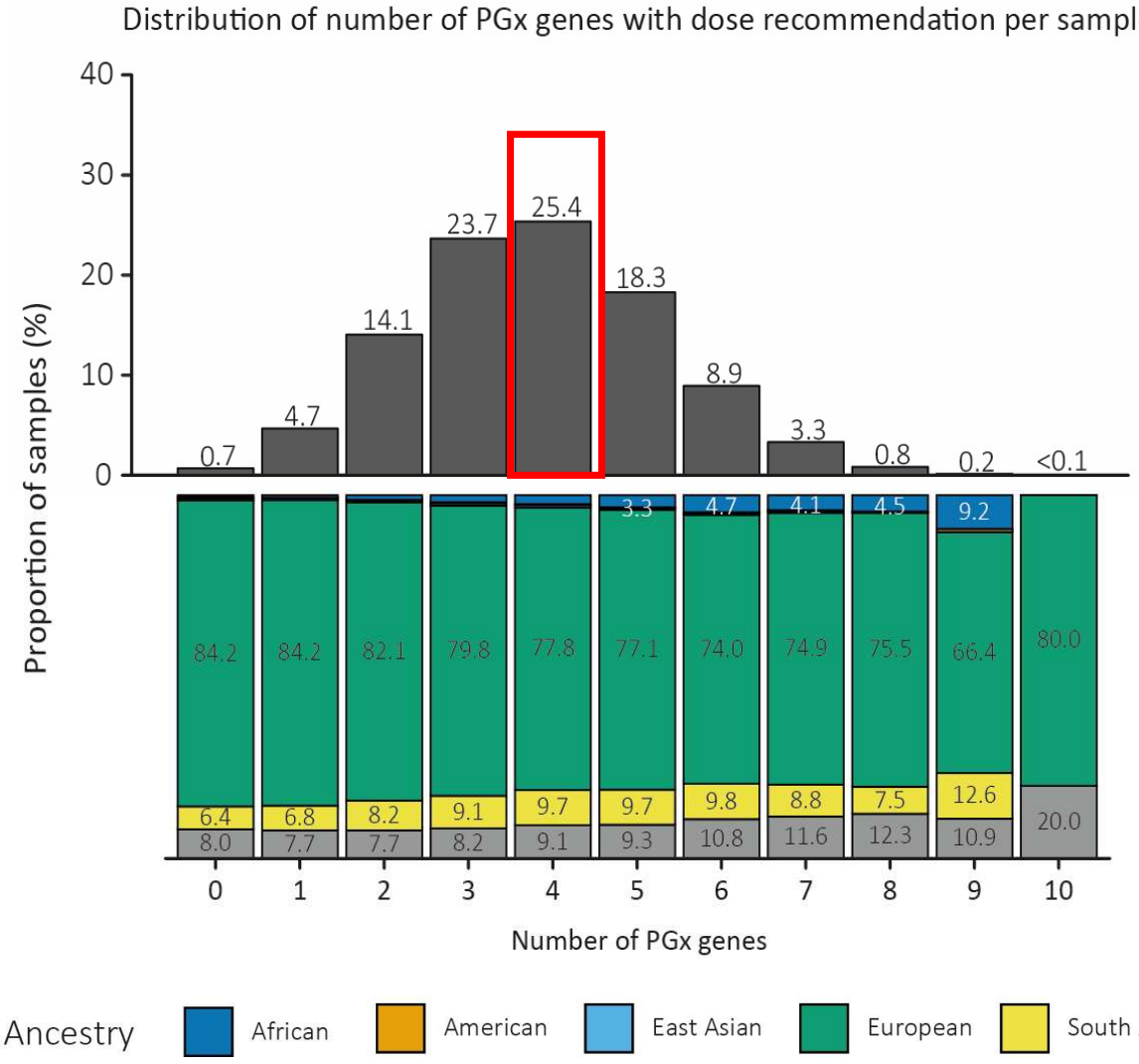

Mutation absent
Mutation present
BRAF
Survival probability
1.00
0.75
0.50
0.25
0.00
Absent
(N=10958)
Present
N=436)
Cancer stage
1
1.2
1.4
1.6
1.8
2
2.2
ne (years)
0
2
4
6
Number of cases
Absent 10958 9029 2045 32
Present 436 340 88 0
BRCA1
Absent
(N=10538)
Present
(N=856)
Cancer stage
0.6
1
1.4
1.8
2.2
Number of cases
Absent 10538 8637 1953 31
Present 856 732 180 1
BRCA2
Absent
(N=10847)
Present
(N=547)
Cancer stage
0.6
1
1.4
1.8
2.2
Number of cases
Absent 10847 8903 2028 32
Present 547 466 105 0
CDKN2A
Absent
(N=10139)
Present
(N=1255)
Cancer stage
1
2
2.4
2.8
ne (years)
Number of cases
Absent 10139 8558 1944 23
Present 1255 811 189 9
KRAS
Absent
(N=9863)
Present
(N=1531)
Cancer stage
1
1.6
1.8
2
2.2
Number of cases
Absent 9863 8119 1859 26
Present 1531 1250 274 6
PIK3CA
Absent
(N=9161)
Present
(N=2233)
Cancer stage
0.8
1.2
1.6
2
Number of cases
Absent 9161 7466 1738 30
Present 2233 1903 395 2

Queen Mary
University of London

Pharmacogenomics



Frequency of PGX gene variants in whole genome sequences



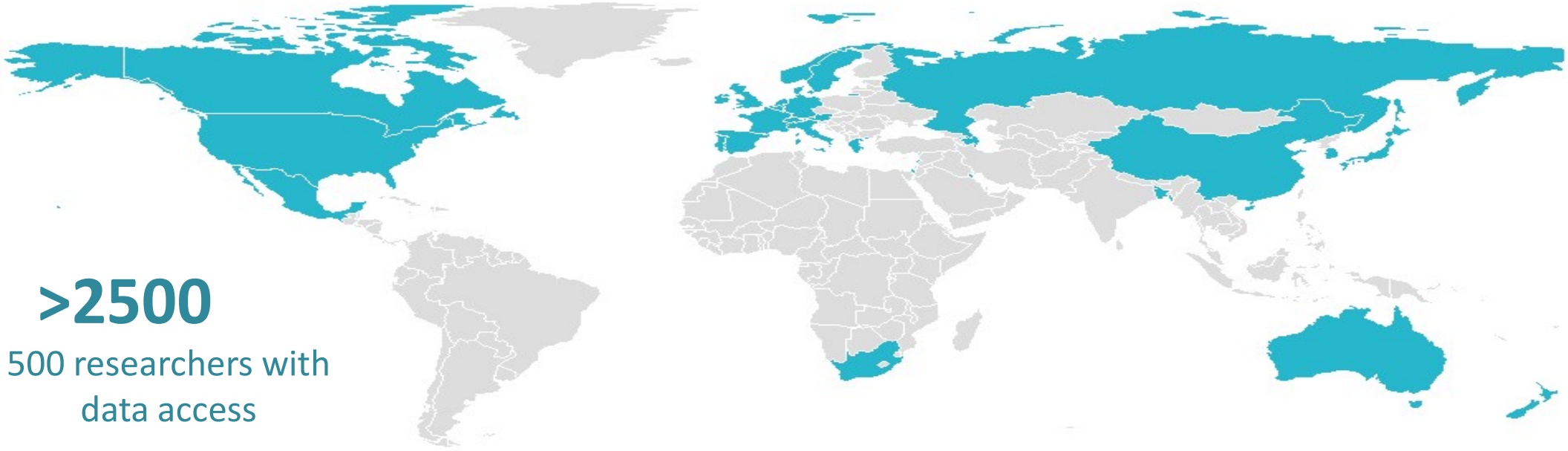
Genomics England Cohort

Participants n = 76,805

- African = 1,916
- American = 173
- East Asian = 450
- European = 60,388
- South Asian = 7,019
- Mixed ancestry = 6,859

- ~99.5% of participants have haplotypes in at least 1 PGX gene.
- 25.4% participants have haplotypes in 4 PGX genes.
- Yellow Card Biobank – pancreatitis with GLP1 drugs

Genomics England Clinical Interpretation Partnership
4 billion clinical data points and 140,000 whole genomes



>2500

500 researchers with data access

165 papers

>300 new diagnoses made

£55 million in grants won

NHS infrastructure

NHS Genomic Medicine Service

Clinical genetics services
Informed patients & clinical teams

Test ordering & Genomic MDT clinicians

Patients & families

National Genomic Testing service

7 NHS Genomic Laboratory Hubs (GLHs)

National Genomic Test Directory

National Whole Genome Sequencing provision

Whole Genome Sequencing interpretation & decision support

Ongoing research & discovery

National Genomic Research Library

Elements delivered in partnership with Genomics England

Genomics england

7 Genomic Medicine Service Alliances

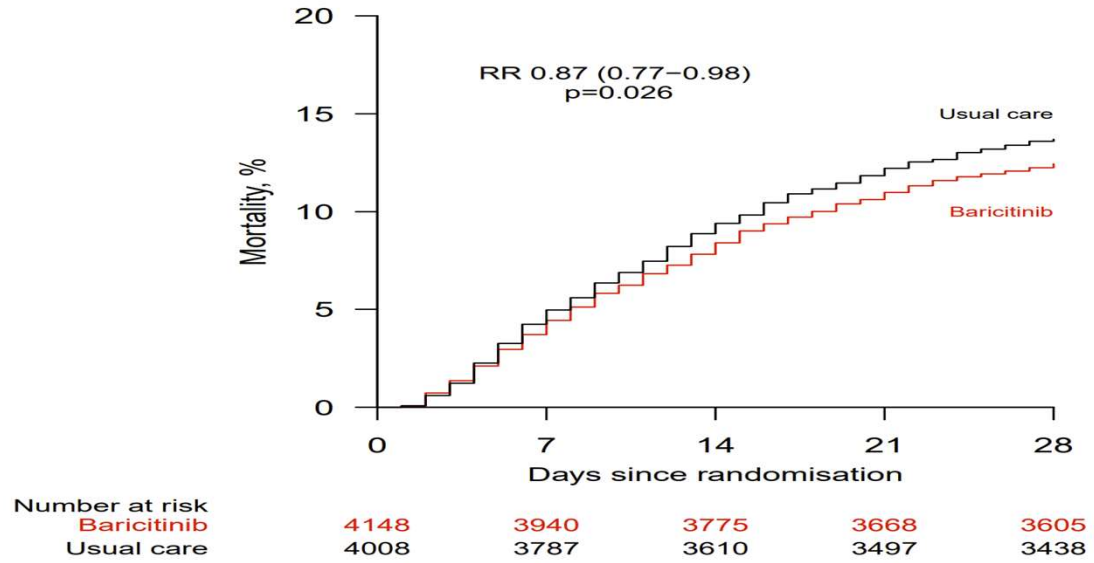
Multidisciplinary leadership to embed genomic medicine via patient pathways through networks across defined geographies

Workforce development and education

Integrated & co-ordinated workforce Genomics Education Programme and appointed workforce development leads

Genetics of host response to COVID-19 in critical illness

- Host-mediated lung inflammation drives critical illness caused by COVID-19.
- Genome-wide association study in 2,244 critically ill patients with COVID-19 from 208 UK ITUs.
- Validated associations on Chr 12q24.13 OAS1, OAS2 and OAS3; 21q22.1 in the interferon receptor gene IFNAR2: Chr 19p13.3 in the gene that encodes dipeptidyl peptidase 9 (DPP9)
- Chr 19p13.2 near tyrosine kinase 2 (TYK2) targeted by Baracitinib;
- Helped the case for baracitinib in the RECOVERY Trial which reported 13% reduction of mortality and length of ITU stay when baracitinib is added to dexamethasone and tocilucimab in 2022



Nature 2021

Baillie et al, Nature 2020

The GenOMICC study in Severe COVID

The Study Design

7,491 severely ill COVID-19 from 224 UK intensive care units

compared with

48,400 unrelated controls from the 100,000 Genomes Project

validation in

The Host Genetics Initiative minus GenOMICC plus 23andme

TWAS and Mendelian Randomisation

Gene Burden Testing for rare variants cases v 100K

The Results

- Genomewide significance adjusted for 2.26 M tests to a P value of 2.2×10^{-8}
- 23 signals were GWS
- Validation by same direction of effect and P Value <0.002
- 16 novel loci
- Transethnic GWAS – 3 signals in S. Asians
- *TRIM46, BC11A, LINCO1276*
- TWAS – multiple signals with significant fold changes in expression
- Gene burden – nothing v 6000 controls
- Two putative therapeutic targets

Baillie K et al Nature 2022

Genome Analysis in Children

NHS South West NHS Genomic Laboratory Hub

ITU Rapid Genome Service Yield

- Neonates and children – extended up to 25 years of age
- Unexplained admission to Intensive Care or suspected rare disease
- Started as rapid exomes
- Now rapid whole genomes

October 2019 - 2023

- >1000 cases plus a year
- 41% diagnostic yield

The Generation Study

Focus on fully penetrant disorders in 100,000 Newborns in the NHS
Identify treatable rare diseases in 500-1000 babies early

Database identified 639 disorders present in childhood with a treatment circa 1:190 births. Start with 208 conditions caused by 468 genes

702,680 live births in the UK in 2022
3,698 children born/ year with a treatable rare disease (10 babies born every day)

Where treatment has impacts on outcomes within five years of birth, the estimated funds released are between £360,323 and £1,441,292,

Quality adjusted life year gains of 16.8 and 40.3 QALYs per child diagnosed.

2021 – Department of Health funded whole genome sequencing of 100,000 Newborns with £100 million

https://www.genomicsengland.co.uk/news/genomics-england-announces-list-of-rare-conditions-to-be-included-in-world-leading-research-study

Public Dialogue on Newborn screening

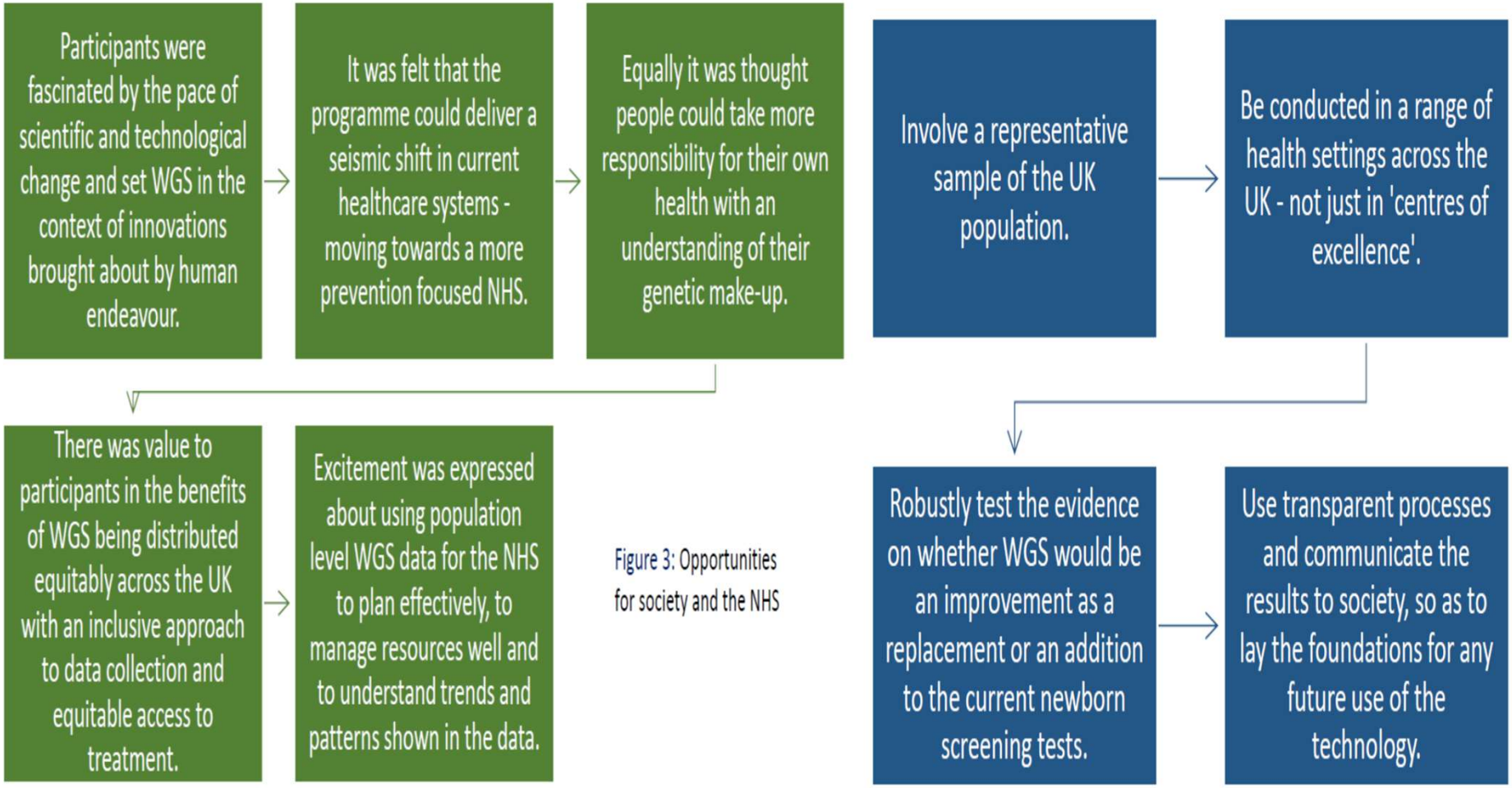


Figure 3: Opportunities for society and the NHS

The Generation Study

12,840 Newborns
35 Diagnoses returned
Confirmatory testing
1 in 250 cases
Revio do 2 week sequencing
Recall for research
Testing for adult conditions

BabySeq2, Harvard

BeginNGS, Rady

GUARDIAN study, Columbia
ScreenPlus, Albert Einstein
EarlyCheck2, North Carolina

GENE	CONDITION
DUOX2 – 2 cases	Thyroid dyshormonogenesis
POMC	Obesity, adrenal insufficiency, and red hair due to POMC deficiency
NR3C2 – 2 cases	Pseudohypoaldosteronism type I, autosomal dominant, 1 confirmed
RB1	Retinoblastoma
GH1	Isolated growth hormone deficiency type II, autosomal dominant
OTC	Ornithine transcarbamylase deficiency
INS	INS related neonatal diabetes

The Department of Health and Social Announced £650 million over 10 years to extend the Newborn Study and to include genomic prevention strategies for children's, adults & Pharmacogenomics

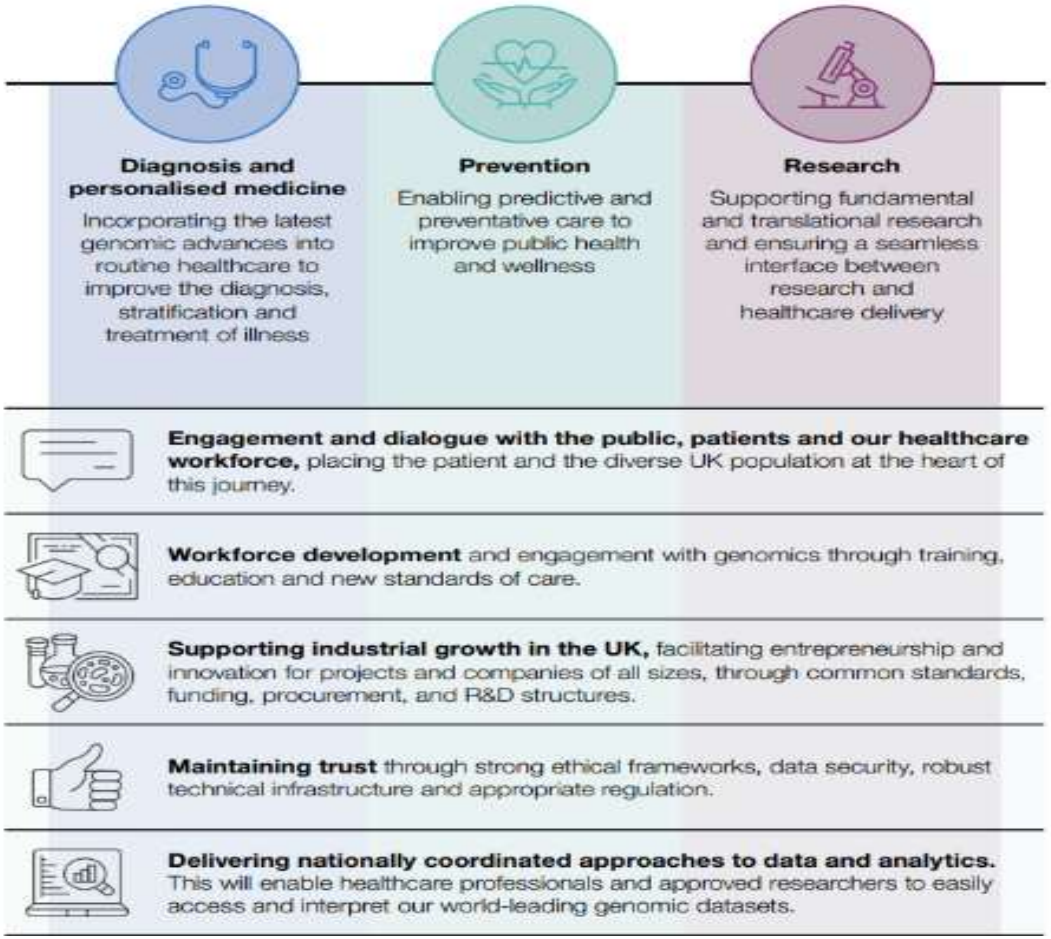
Public survey showed significant interest in receiving personalised genomic results as Genomic Volunteers

Source: Ipsos-Mori survey of the public perspectives of genomic volunteers. Electronic survey of 1,866 people (selected to be a representative sample of adults aged 16 – 75 across England)

GENOME UK – The future of healthcare (2020)

- Sets out how the UK genomics community – from **researchers through to the NHS** – will harness the latest advances in genomic science, research and technology for the benefit of patients, **to create the most advanced genomic healthcare system in the world**

- It will drive improvements in healthcare for patients, **reducing boundaries between clinical care and research**, and deliver innovation in the UK. Unites the genomics community behind a **shared vision** for the future of the system



Transforming the future genomic medicine service

The National Health Service is creating:

- A National Genomic Medicine Service potential for consistent & equitable care for 56 million population
- Operating to common national standards, specifications & protocols
- Standardised genomic consent for NHS care and Research
- An annually reviewed National Test Directory - single gene to WGS
- Building a single UK Genomic Research Library to which 96% consent
- De-identified data for academic, NHS & industry research
- Newborn study extended with £650 million for childrens, adults and Pharmacogenomics
- The future is a global coalition of intellects driving genomics into healthcare and our goal is for the UK to be at the heart of that

Genomics england

Thank you to everyone who has taken part in the 100,000 Genomes Project

NHS

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