Genomics England Experience

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Our history

December 2012

Announced by former Prime Minister David Cameron

July 2013

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

November 2016

Former Prime Minister Theresa May opens a new Sequencing Centre

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”

Our vision is a world where everyone benefits from genomic healthcare

We do so by enabling others to deliver genomic healthcare and accelerate genomic research.

These means we work across the ecosystem:

biobank uk
Improving the health of future generations

NHS England

NHS Digital

NIHR | National Institute for Health Research

CANCER RESEARCH UK

Genomics England

HDRUK Health Data Research UK

+ Our Future Health

Oxford NANOPORE Technologies

lifebit

LifeArc

NICE National Institute for Health and Care Excellence

Congenica

UKRI

aws

illumina

Academia ~4,000 researchers

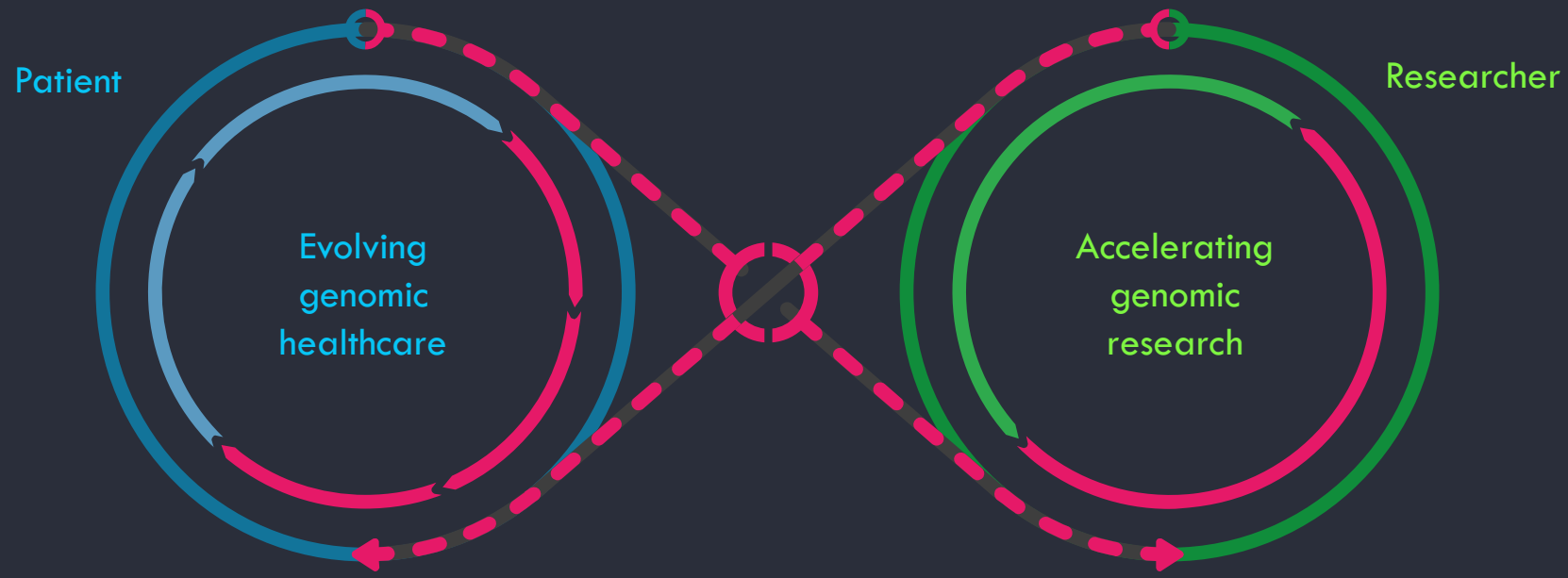
BioPharma & Start-ups
(over 100 Discovery Forum members, 8/10 Tier 1 Pharma)

Tech
(e.g., AWS, Nvidia, Lifebit, Congenica)

Sequencing Tech
(e.g., Illumina, Oxford Nanopore)

Funders
(e.g., UKRI & MRC & InnovateUK, CRUK, Wellcome, BHF, Lifearc)

GEL Strategy: An Infinity Loop



Patients

Healthcare teams

Researchers

100,000 Genomes Project Data

Release v.15

Genomics

	Cancer	Rare Disease	Total
Participants	17K	73K	90K + 30K COVID
Genomes	15K Germline + Tumour 30x 100x	72K Germline <20% Singleton	120K 150K 30X genomes equivalents

100,000 Genomes Project Data

Genomics

Clinical Data

- Tumour staging
- Tumour location
- Histological subtype
- Treatment regimen
- Pathology full-text
- Radiology full-text

- Hospital Episode Statistics
- Mental Health Services Data Set

- Mortality data ONS

- COVID-19 status

- Diagnostic outcomes

- Primary Care Data (coming soon)

100,000 Genomes Project Data

Genomics

Clinical Data

Consent

Clinically accredited pipelines

for diagnostics

Lifetime follow-up

+ full retrospective data

Re-engagement

re-phenotyping

re-sampling

re-recruiting

Diagnostic odyssey of children born 2003 onward Rare Diseases Pilot

68 hospital appointments and **6** years to obtain a diagnosis

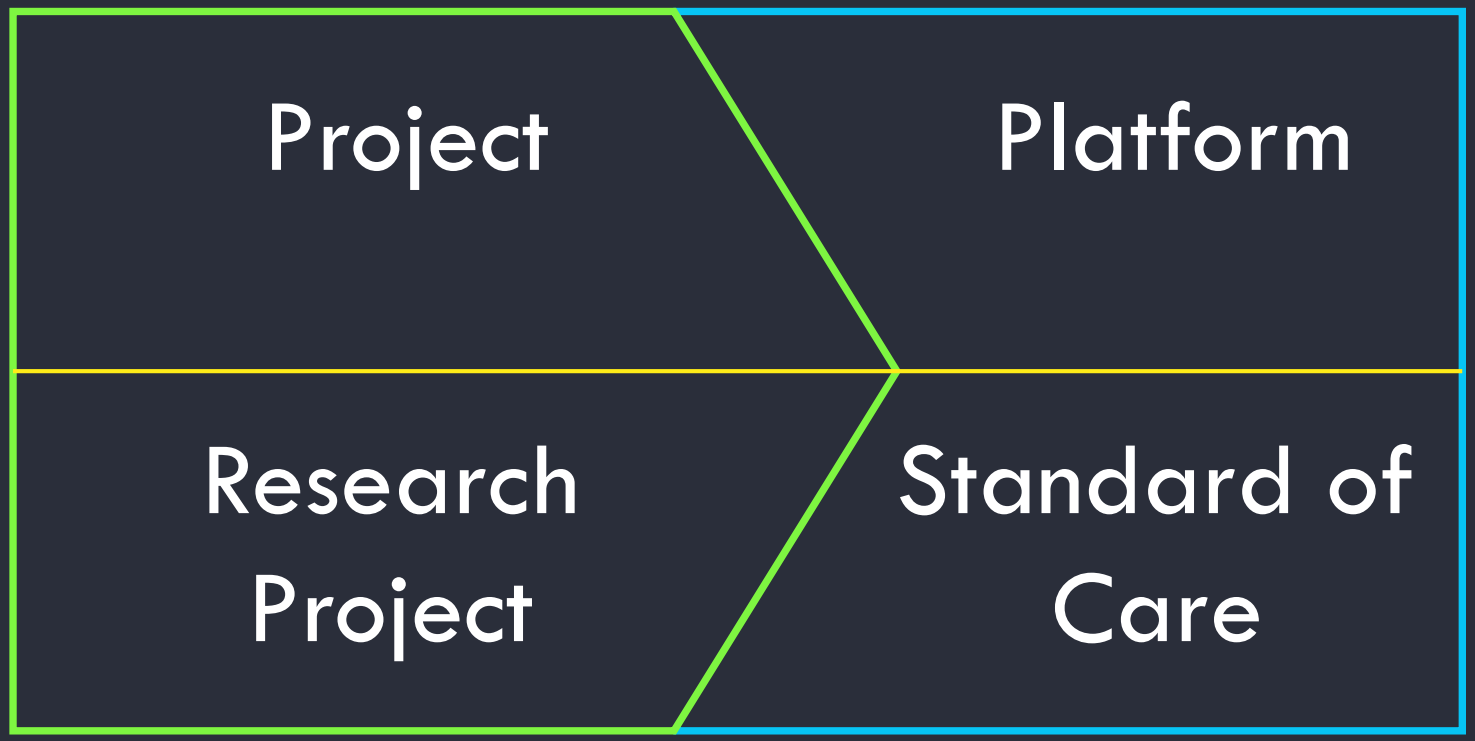
18 hospital appointments over **10** years from birth, for unaffected relatives

183,273 episodes of hospital care and cost of **£87m**, for participants

53,706 episodes of hospital care and cost of **£21m**, for unaffected participants

25% of cases with diagnosis had actionable findings

N Engl J Med 2021; 385:1868-1880 DOI: 10.1056/NEJMoa2035790



Required continued investment in:

- Stronger engineering
- Products and services mindset
- Operational excellence and accreditations
- Implementation in healthcare

NHS Genomic Medicine Service

National Genomic Test Directory

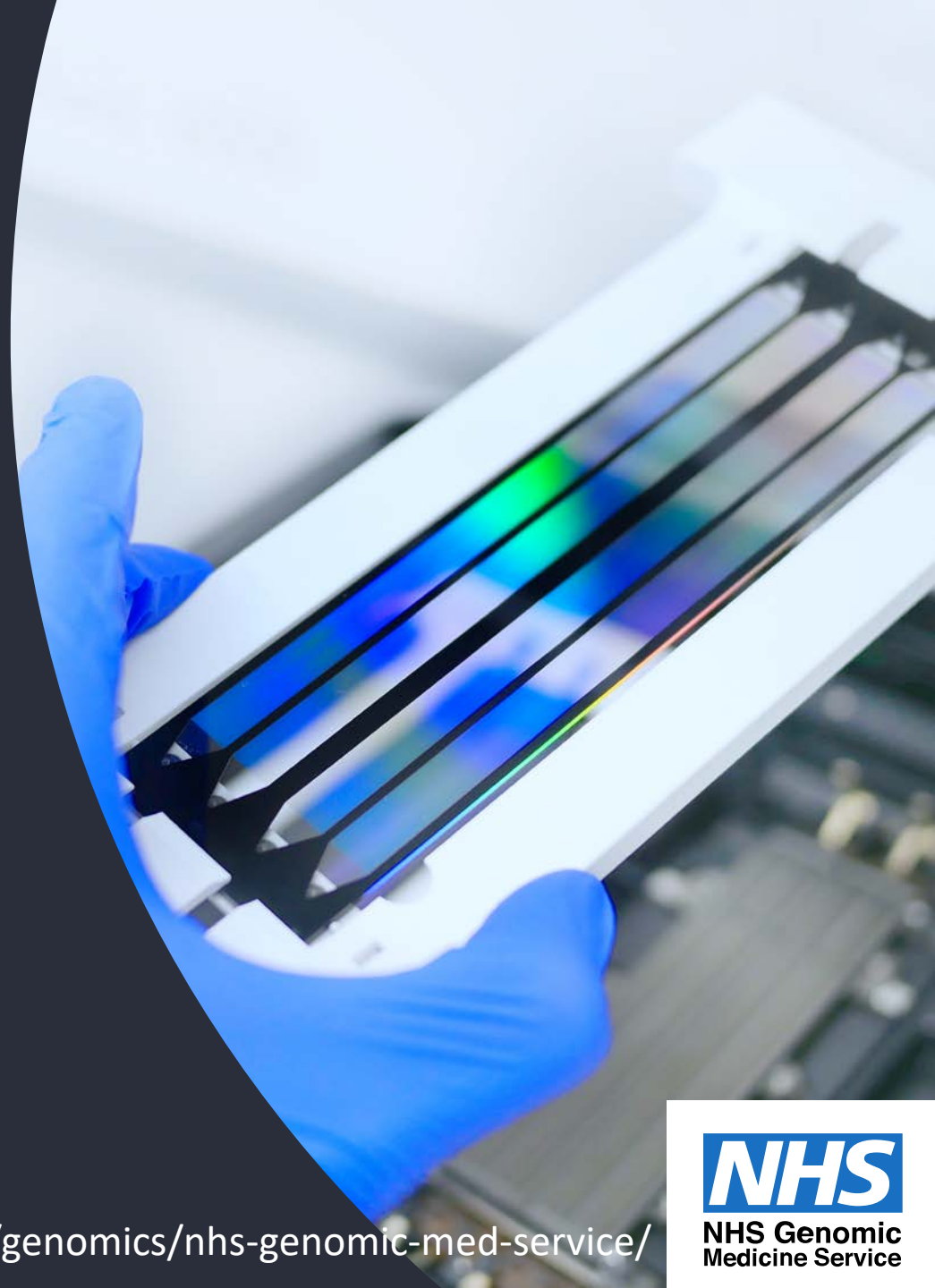
WGS Cancer indications

- Wave 1: Acute Leukemias, Paediatric Tumors, Sarcomas
- Wave 2: Ovarian HGS, Triple Negative Breast, Glioma, Other Heam Onc, Various relapse & refractory

WGS Rare Disease indications

- Wave 1: 20 rare conditions
- Wave 2: +10 rare conditions

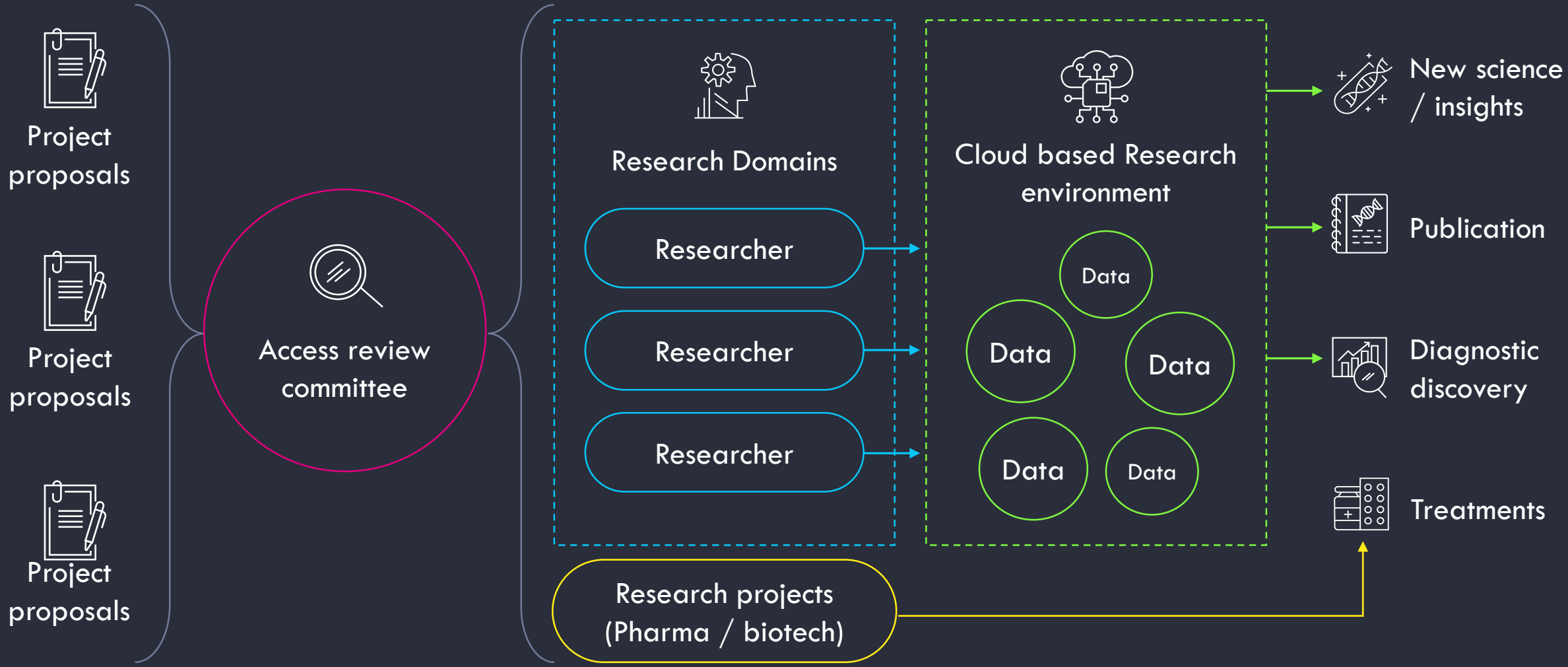
>20,000 genomes sequenced in the last 18 months.



https://www.england.nhs.uk/genomics/nhs-genomic-med-service/

NHS Genomic Medicine Service

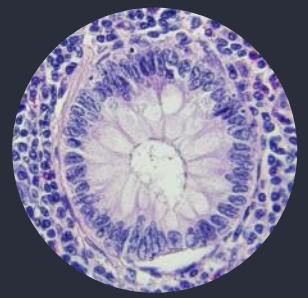
How do academics and biotech/pharma work with us? The National Genomics Research Library



Where next?

Newborns screening

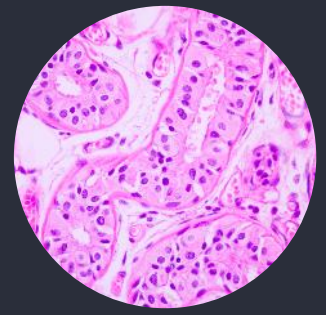
Increasing data diversity



Cancer 2.0

Long-read Sequencing

To support rapid, precision diagnostics and treatment planning;



Multi-modal Data

Enriching our data set for cancer research with the addition of pathology and radiology imaging data alongside whole genome data.

In the long run we aim to enable a lifelong relationship with the genome

Screening; Rare genetic diseases; NICU PICU

e.g., Autoimmune disease, Mental health

e.g., Polygenic risk of common diseases; Pharmaco-genomics

e.g., Cardiovascular; Cancer

e.g., Neuro-degenerative disease


https://www.genomicsengland.co.uk/