

# UK Biobank



Call us on: 0800 0276 276

Mon-Fri 8am-6pm (Sat 8am-4pm)



Search for research

Search

[About](#) | [Participants](#) | [Resources](#) | [Scientists](#) | [Data Showcase](#) | [Register & Apply](#) | [Research](#) | [Publications](#) | [AMS Login](#) | [Careers](#)

UK Biobank is a national and international health resource with unparalleled research opportunities, open to all bona fide health researchers. UK Biobank aims to improve the prevention, diagnosis and treatment of a wide range of serious and life-threatening illnesses – including cancer, heart diseases, stroke, diabetes, arthritis, osteoporosis, eye disorders, depression and forms of dementia. It is following the health and well-being of 500,000 volunteer participants and provides health information, which does not identify them, to approved researchers in the UK and overseas, from academia and industry. Scientists, please ensure you read the [background materials](#) before registering. To our participants, we say thank you for supporting this important resource to improve health. Without you, none of the research featured on this website would be possible.

<http://www.ukbiobank.ac.uk>

<http://www.ukbiobank.ac.uk/2018/07/watch-again-uk-biobank-scientific-conference-2018-2/#>

## TECHNOLOGY FEATURE

# THE DNA OF A NATION

*The United Kingdom aims to sequence 100,000 human genomes by 2017. But screening them for disease-causing variants will require innovative software.*

GENOMICS TECHNOLOGY

**~50,000**  
people with rare  
diseases and  
their parents



**RECRUITMENT OF 75,000 PEOPLE**  
The 100,000 Genomes Project is recruiting people with cancer and rare diseases. The genomes of both normal and tumour cells will be sequenced in people with cancer.

**~25,000**  
people with  
cancer



## THE CLINICAL GENOME

Genomics England plans to sequence 100,000 genomes by 2017. The genomic data will be crucial for diagnosing and treating disease, but its interpretation will require automated, specialized software.



**NEXT-GENERATION SEQUENCING**  
The Californian company Illumina will use UK-based high-throughput sequencing machines to produce whole-genome sequences and identify genetic variants.

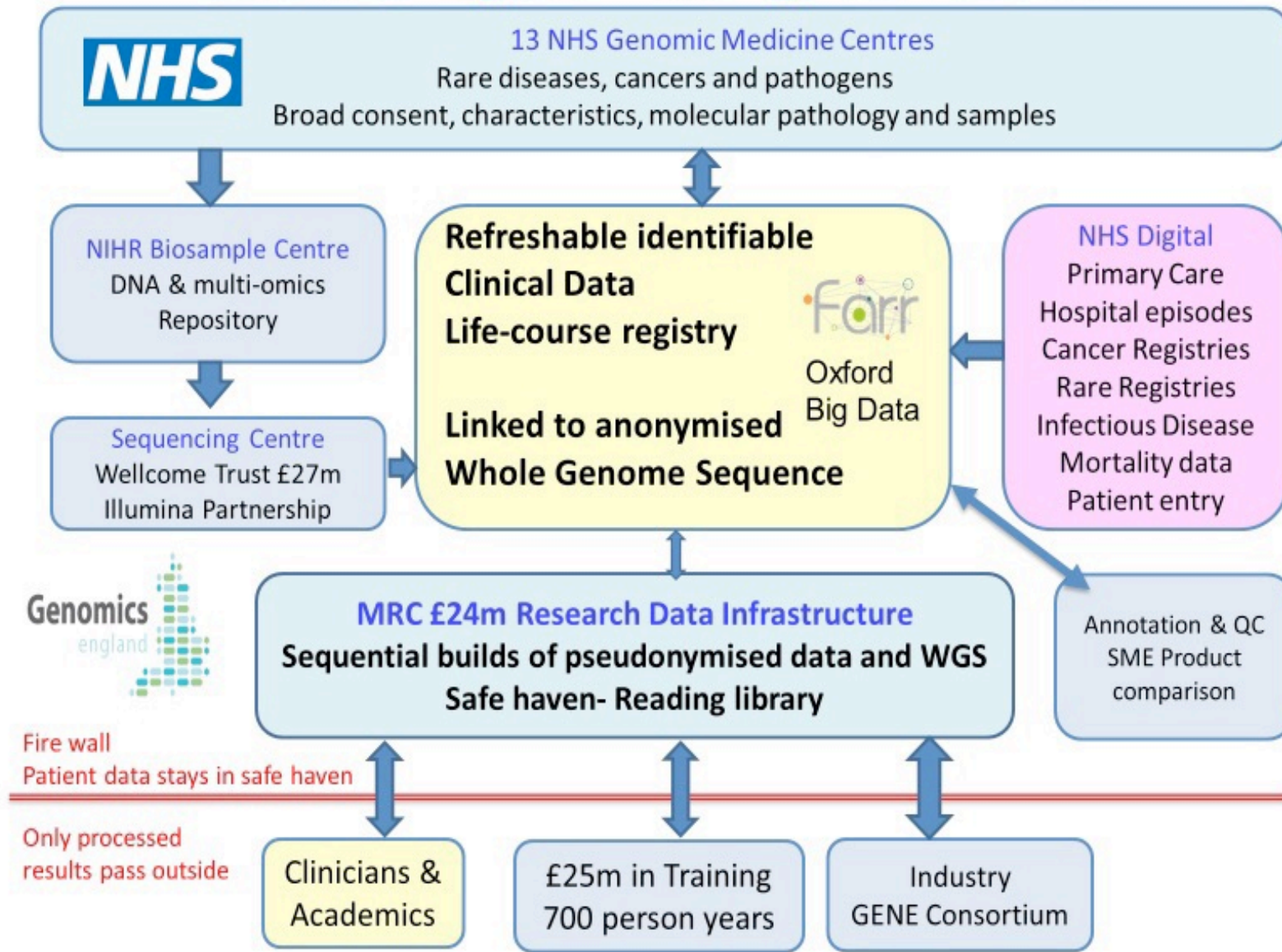


**AUTOMATED INTERPRETATION**  
Four UK and US companies will use specialized software to automatically analyse the genetic variants that may be linked to disease.



**CLINICAL INTERPRETATION**  
Around 2,000 UK scientists and clinicians will pore over the data to validate or better understand how the variants may cause disease before the information is fed back to patients.

# Genomics England – The Big Data Potential

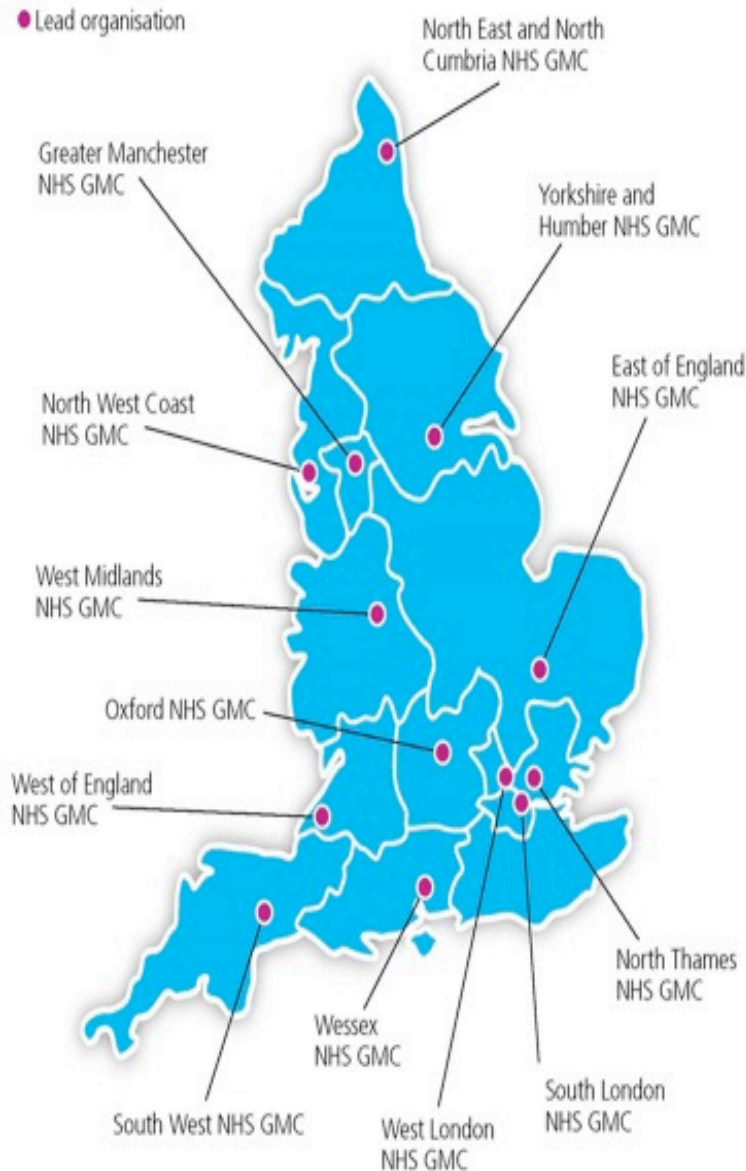


GeCIP

Health Education England Genomics Expert Network for Enterprises (GENE) Consortium

Prof. Mark Caulfield, FMedSci

# NHS clinical interface via 13 “GMCs”, Scotland, Northern Ireland and Wales



- Standardisation is key
- Genomic Medicine Centres
  - Networks of NHS hospitals including genomics labs
  - 13 “Lead organisation” plus 71 “Local Delivery Partners”
  - Contracted by NHS England
  - Cover recruitment, data and return of results
- Scotland
- Northern Ireland
- Wales

## The 100,000 Genomes Project in numbers



Over **100,000** genomes



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

```
110001010101001010100101010000101
110110111010101010001011101000101
110101010001001101010001010100010
001001001110010001000010101010100
1001111011001010101110101111001101
```

**21+** 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)



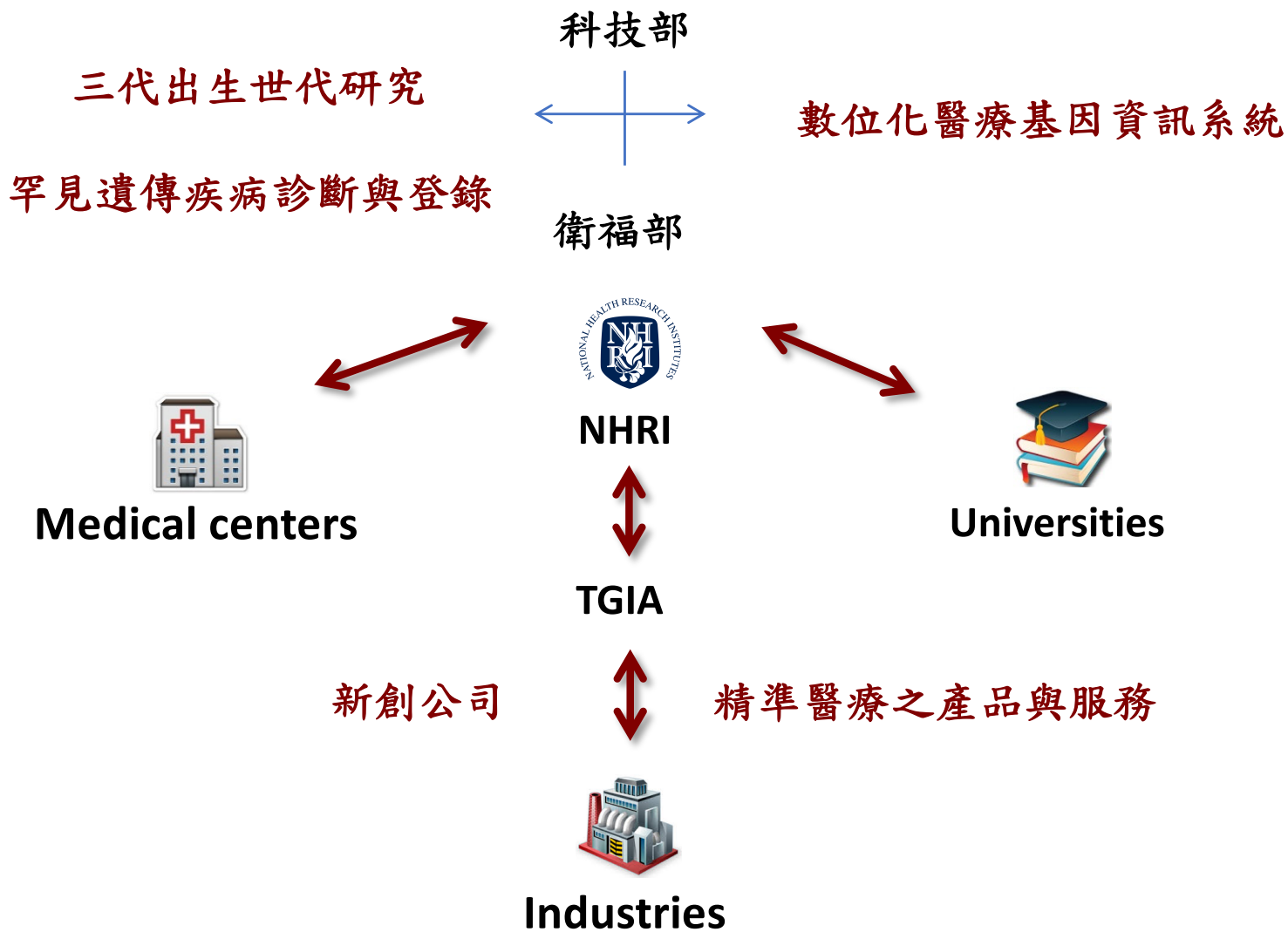
Over **3,000** researchers and trainees

Introducing

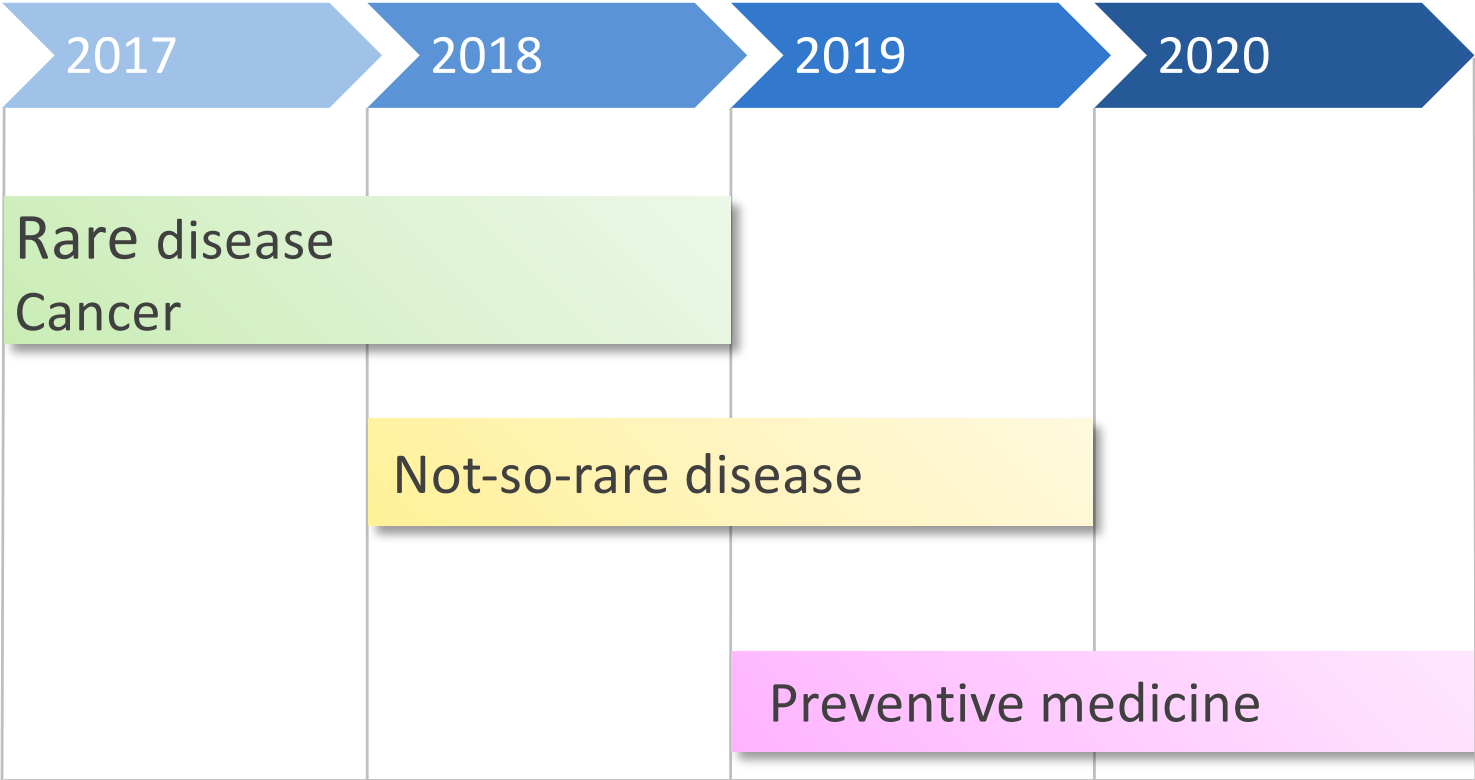
“亞太生醫矽谷精準醫療旗艦計畫”

Flagship Program of Precision Medicine for  
AsiaPacific Biomedical Silicon Valley”

# 亞太生醫矽谷精準醫療旗艦計畫 整合運作



# WGS: Driver of Precision Medicine Lifetime Healthcare Management





## Article

---

# The GenomeAsia 100K Project enables genetic discoveries across Asia

---

<https://doi.org/10.1038/s41586-019-1793-z>

GenomeAsia100K Consortium\*

---

Received: 29 January 2018

---

Accepted: 11 October 2019

---

Published online: 4 December 2019

---

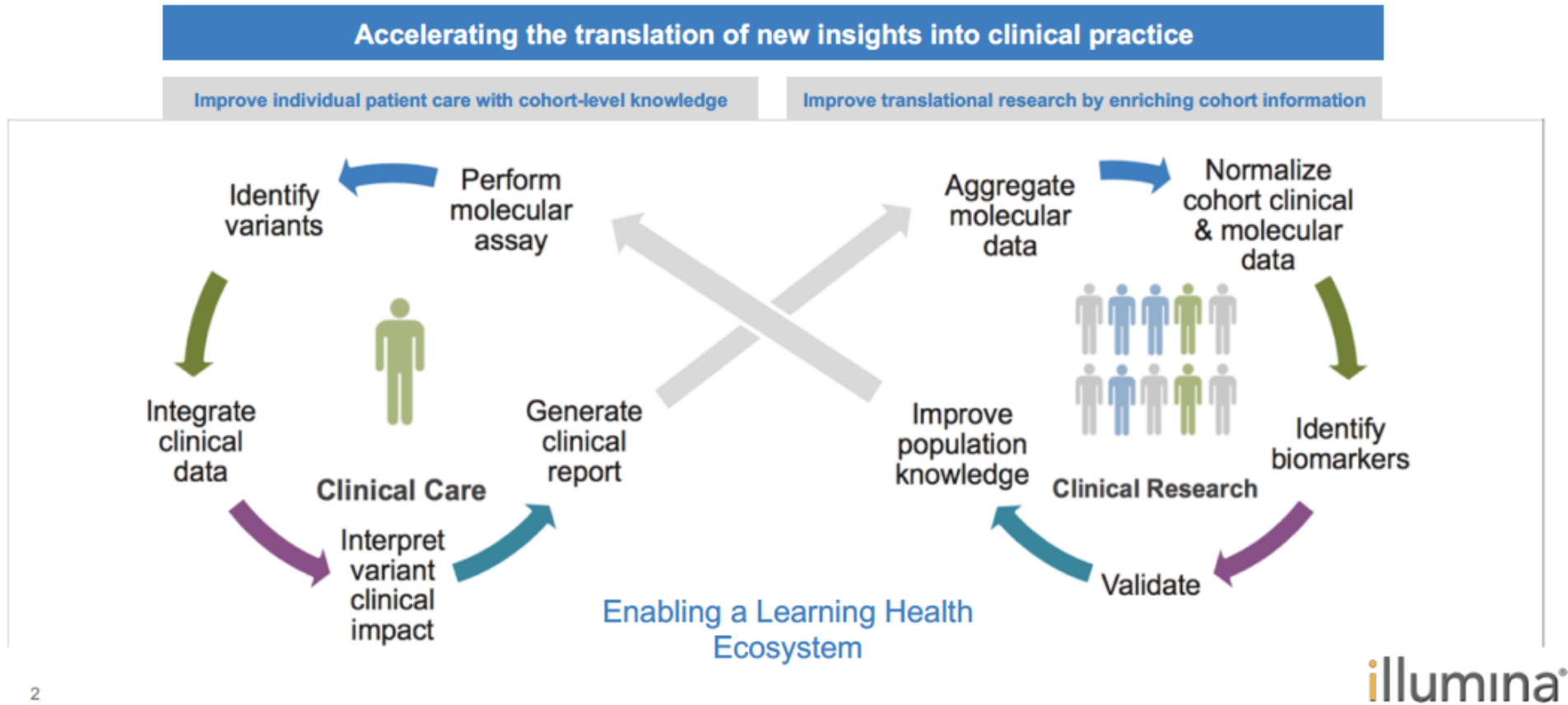
Open access

---

The underrepresentation of non-Europeans in human genetic studies so far has limited the diversity of individuals in genomic datasets and led to reduced medical relevance for a large proportion of the world's population. Population-specific reference genome datasets as well as genome-wide association studies in diverse populations are needed to address this issue. Here we describe the pilot phase of the GenomeAsia 100K Project. This includes a whole-genome sequencing reference dataset from 1,739 individuals of 219 population groups and 64 countries across Asia. We catalogue genetic variation, population structure, disease associations and founder effects. We also explore the use of this dataset in imputation, to facilitate genetic studies in populations across Asia and worldwide.

# Population Genomics Links Research with Clinical at Scale

*Fully Reap the Benefits of Big Data in the Context of Clinical Care*



2

你的基因 我的基因 大家的健康