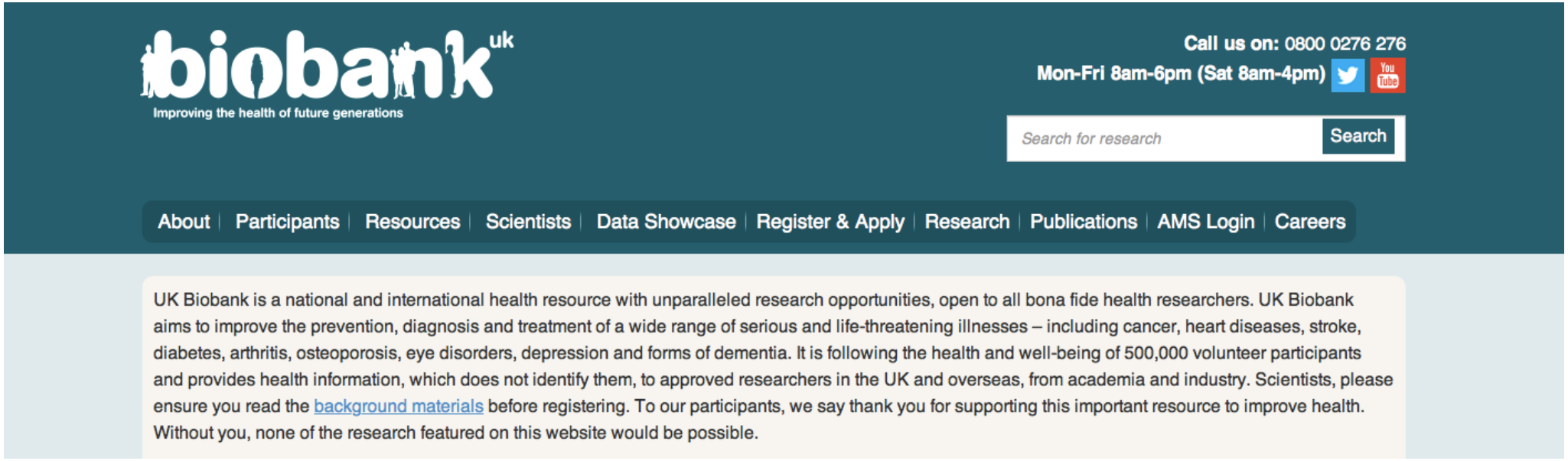
UK Biobank



http://www.ukbiobank.ac.uk

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

TECHNOLOGY FEATURE

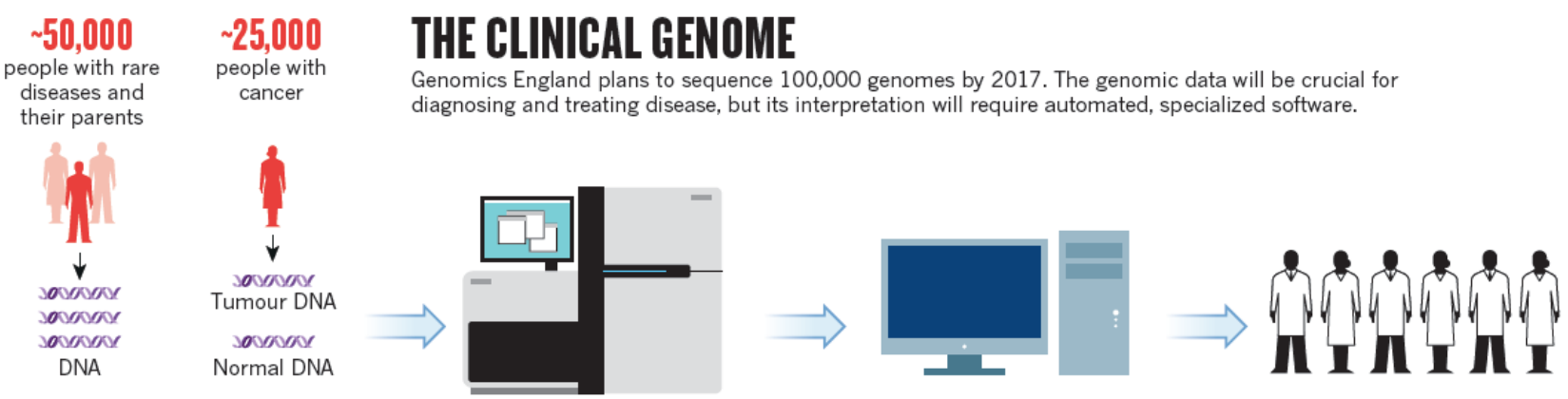
THE DNA OF A NATION

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

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.



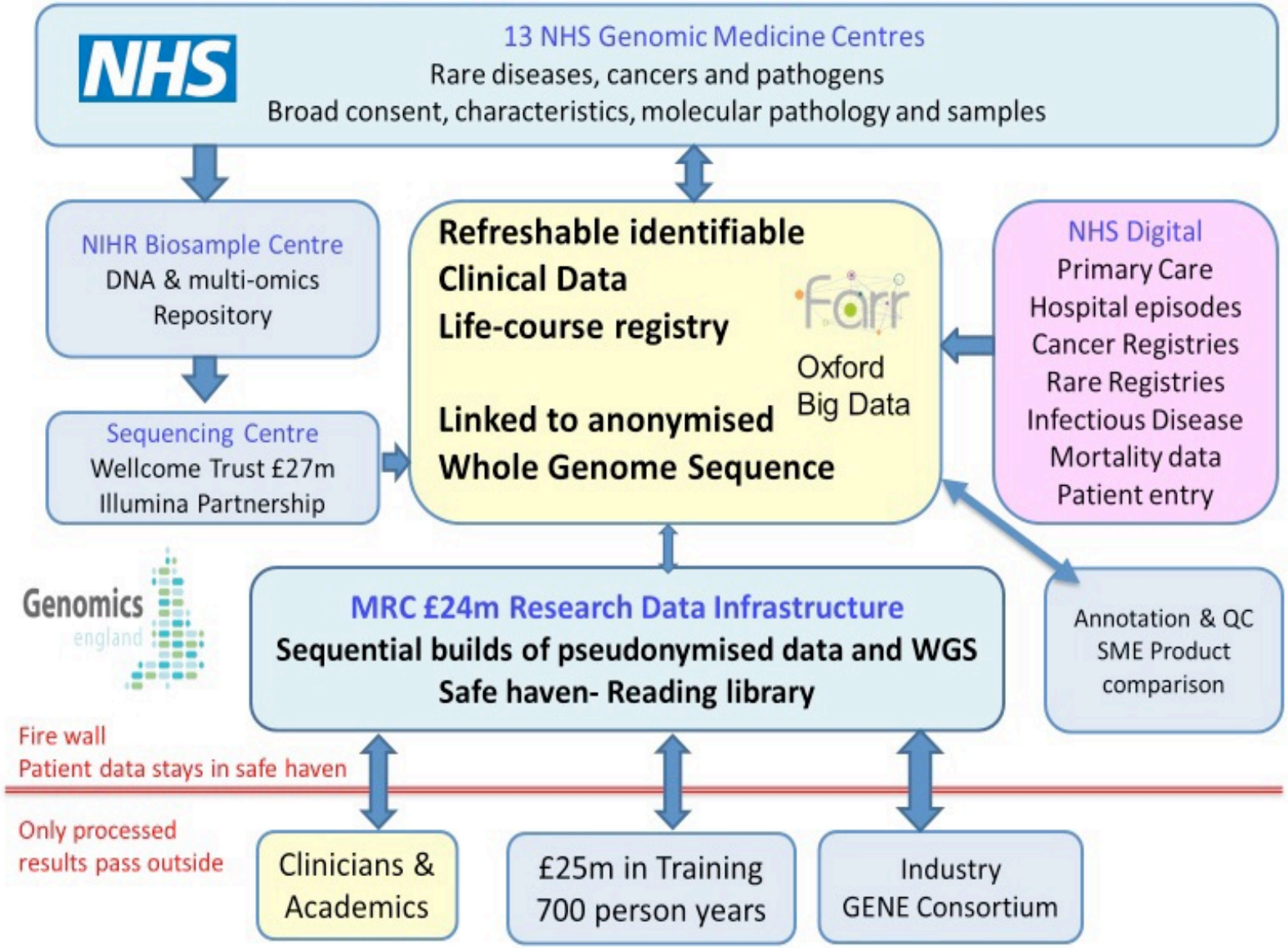
RECRUITMENT OF 75,000 PEOPLE
The 100,000 Genomes Project is recruiting people with cancer and rare diseases. The genomes of both normal and tumour cells will be sequenced in people with cancer.

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.

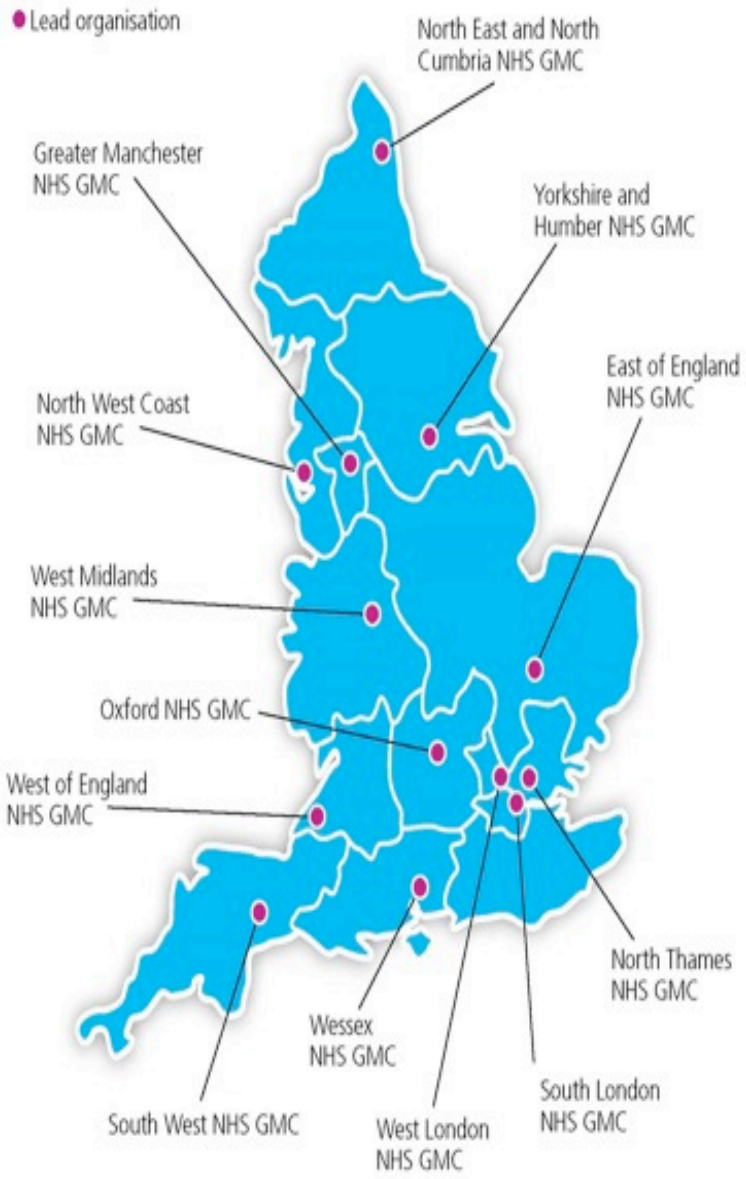
Genomics England – The Big Data Potential



GeCIP Health Education EnglandGenomics 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

Prof. Mark Caulfield, FMedSci

The 100,000 Genomes Project in numbers

Over **100,000** genomes

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

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)

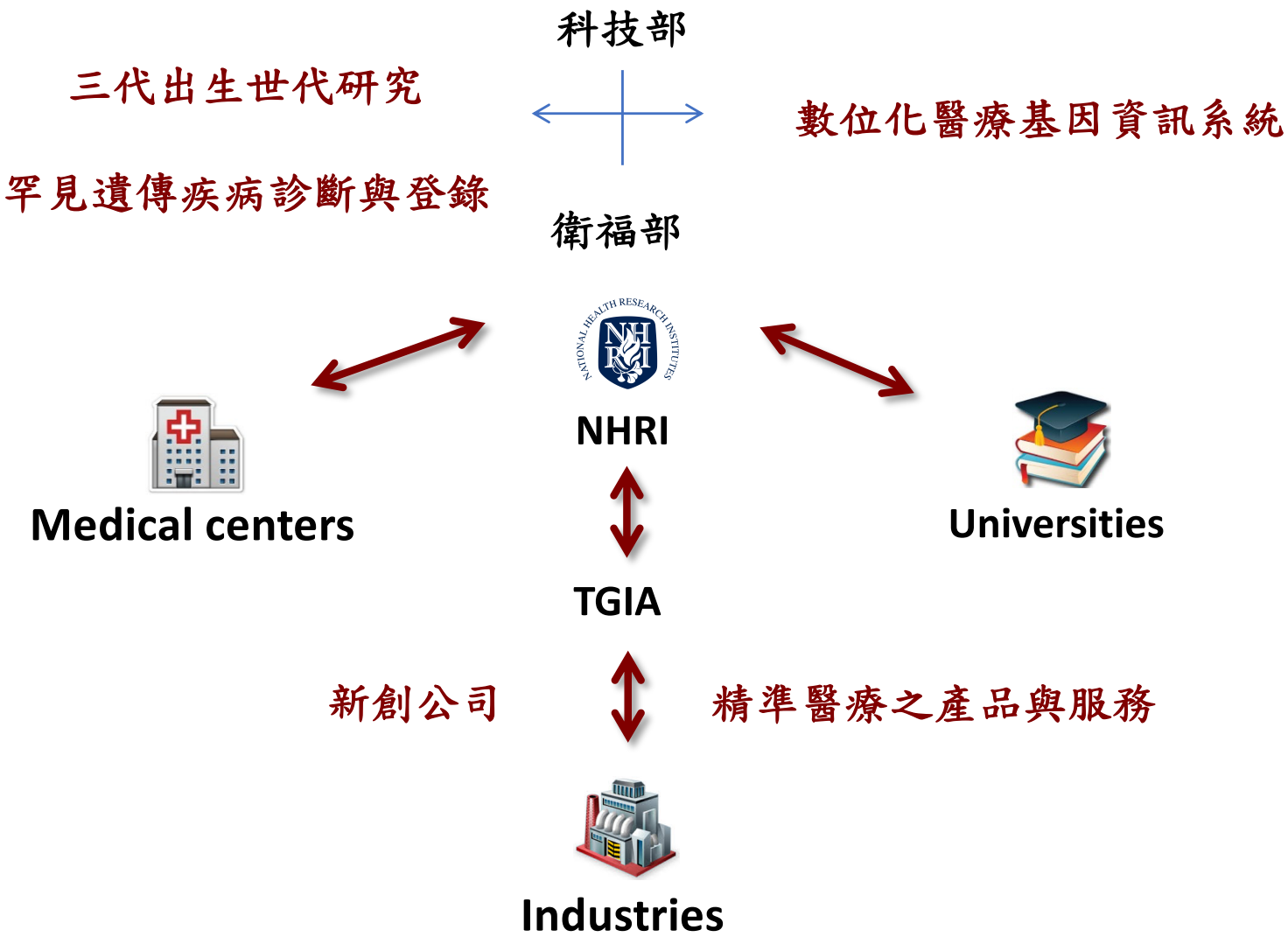
Over **3,000** researchers and trainees

Introducing

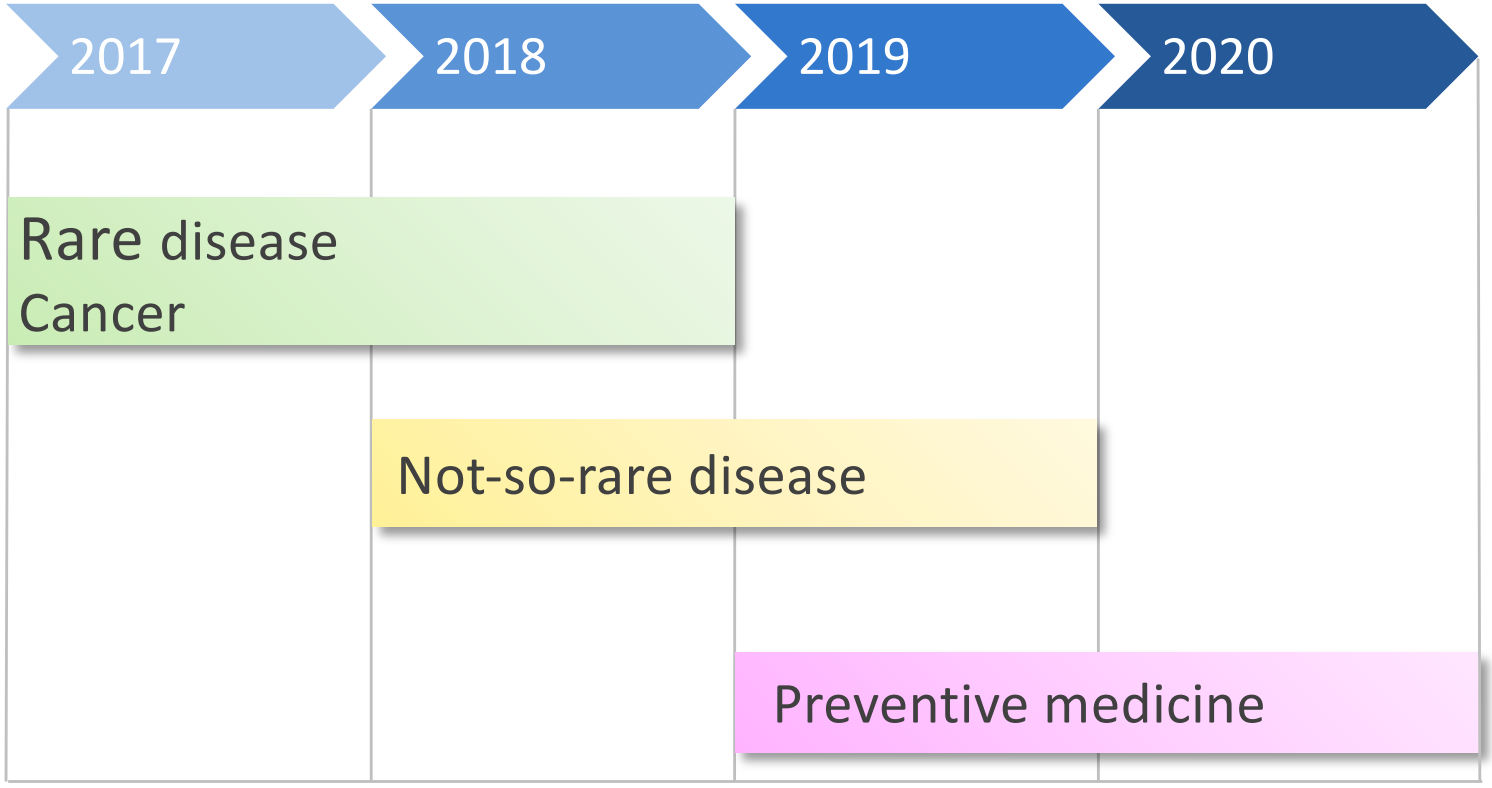
“亞太生醫矽谷精準醫療旗艦計畫”

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

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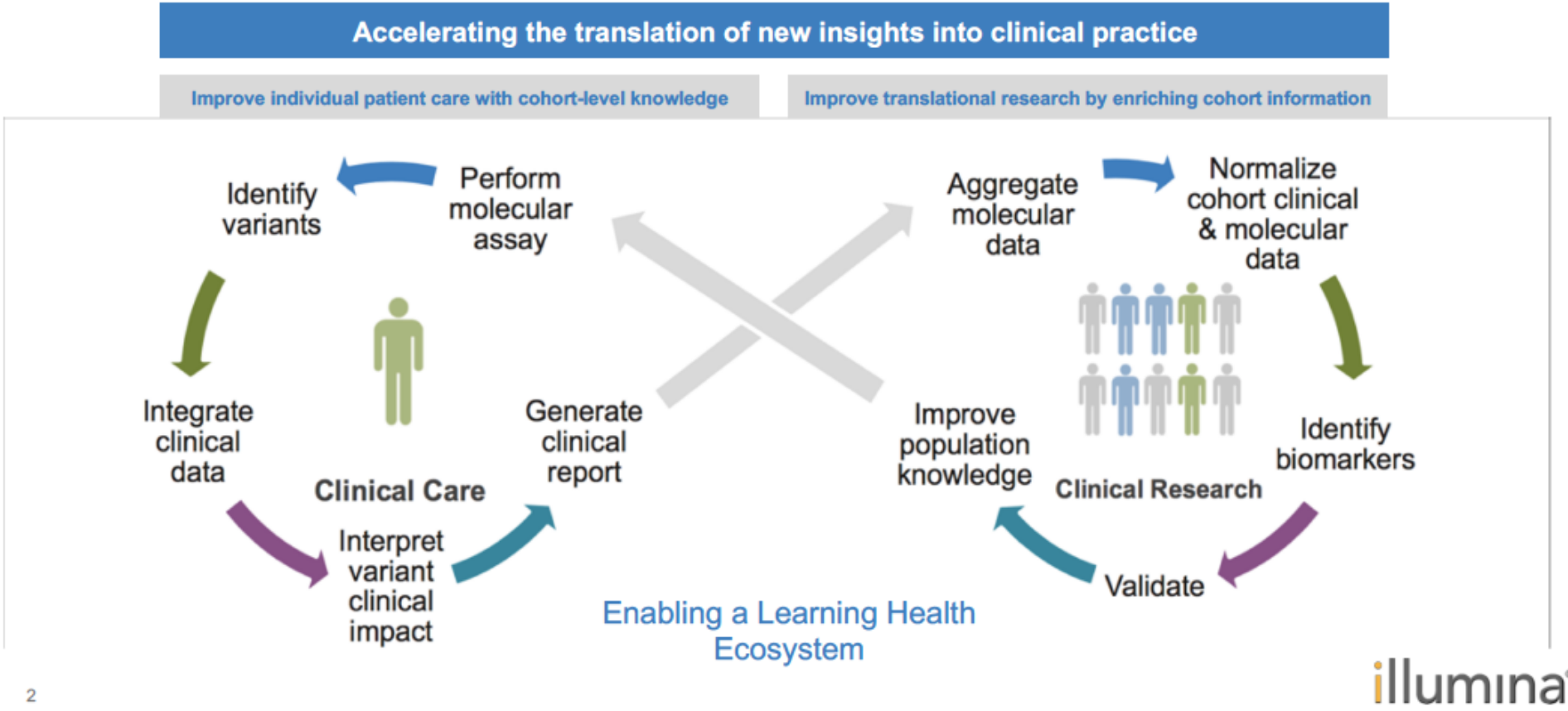
GenomeAsia100K Consortium*

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



illumina

你的基因 我的基因 大家的健康