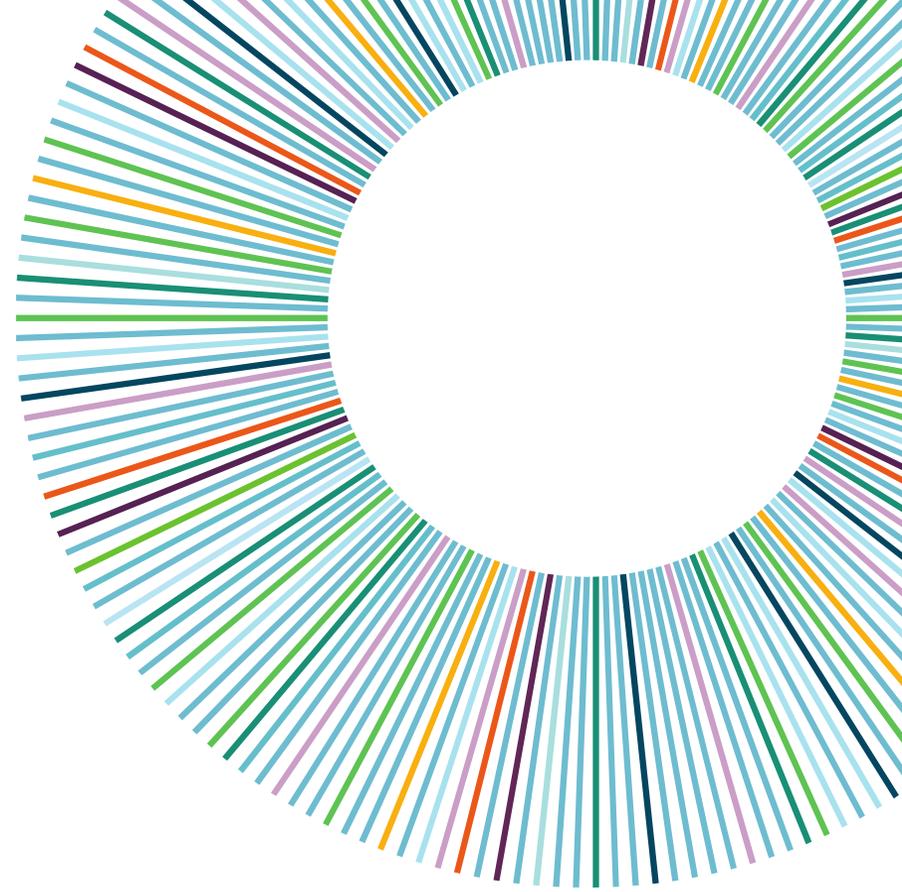


Paul Jones

*Director, Population Genomics
Illumina*



#IMITenYears
#IMICarryTheTorch

Technology convergence - capitalising on the opportunity

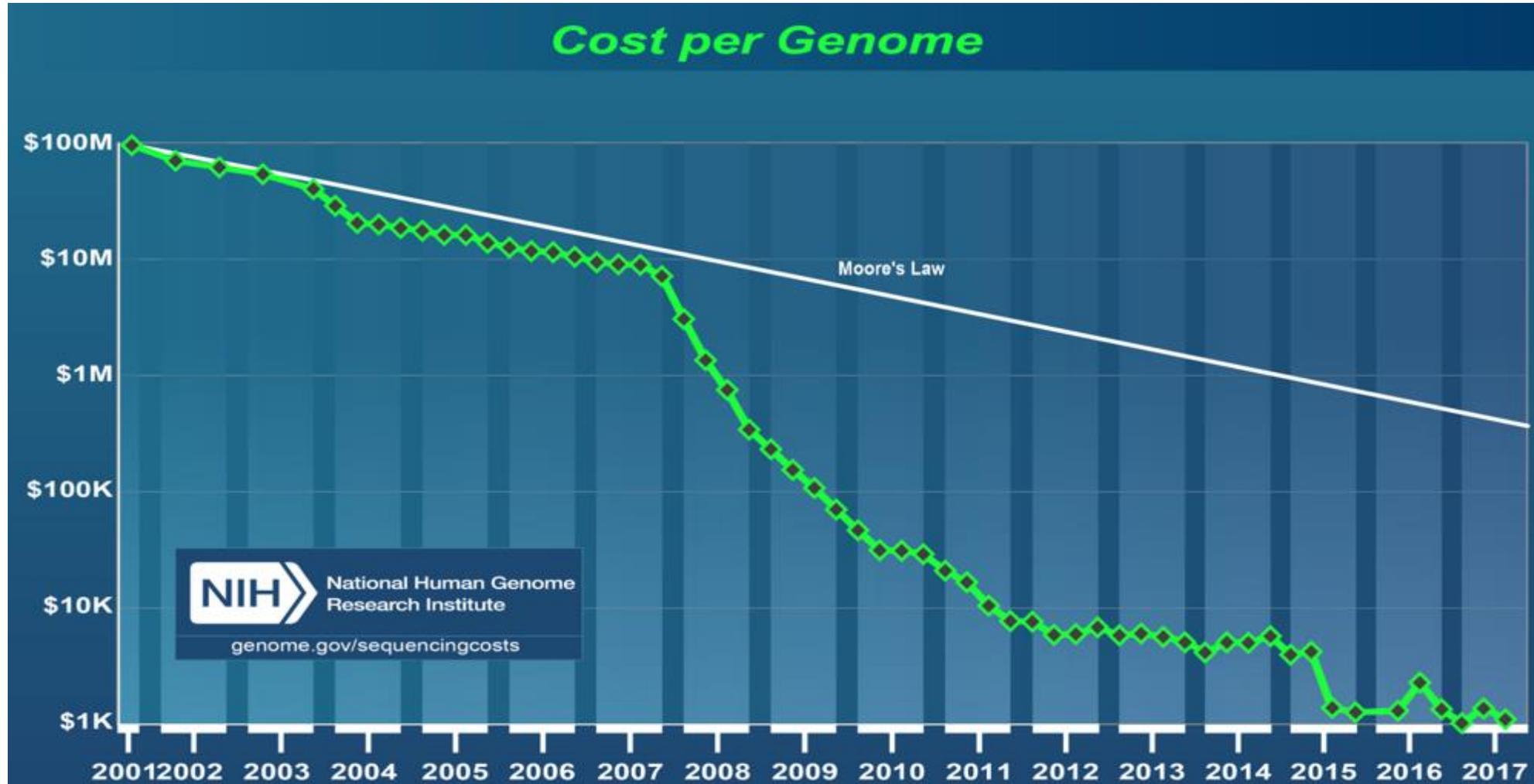
24th October 2018



Technology convergence

- **What does it mean in genomics?**
- **How has it manifested itself?**
- **What strategies are at play to capitalise on it?**
- **Where will it lead in the next 5 years?**

Technology convergence has helped to enable a dramatic fall in the cost of sequencing a genome



Source: National Human Genome Research Institute. www.genome.gov/27541954/dna-sequencing-costs-data/

This in turn has stimulated innovation and growth in a series of adjacent possibles*

Microbiome Research

An enhanced ability to study the human microbiome

Consumer Genomics

The adoption at scale of consumer genomics

Precision Therapeutics

Advances in precision therapeutics

- 132 personalised medicines currently on the market**
- 42% of all drugs [73% in oncology] in development have potential to be personalised medicines**

Gene Editing

The rapid acceleration of gene editing through e.g. CRISPR/Cas9

Liquid Biopsy

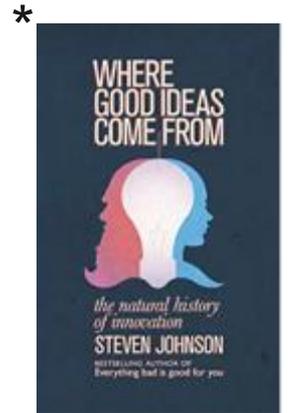
Ongoing research into the potential for liquid biopsy

- Potential to revolutionise the early detection and ongoing monitoring of cancer

Population Genomics

National-scale endeavours to integrate genomics into healthcare systems

- e.g. 100,000 Genomes Project in England



Population Genomics characteristics

Embedded in the health system with full and ongoing *patient engagement*

- The programme links research and clinical care with the opportunity (through appropriate consent) to re-engage patients on an ongoing basis – thus creating a **learning health & care system**

Involves an *accessible integrated data platform* that grows in value over time

- Genomic, phenotype, medical record and outcomes data are linked and made available for research (academic and industry) and clinical purposes
- Critical to be able to **connect, exchange, analyse and act** on the data

Has active *translational research* involvement

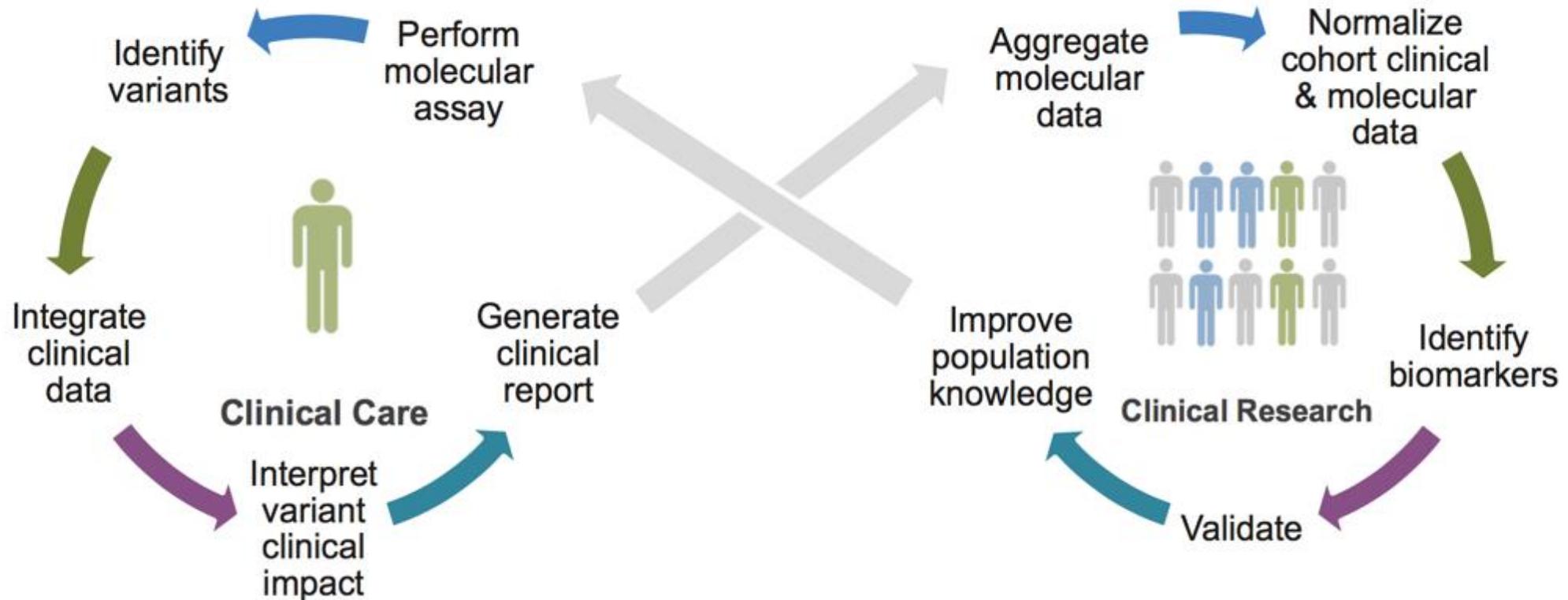
- Academic and industry players are integrally involved in the programme with the goal of **translating research into clinical practice**

Population Genomics programmes aim to link research with clinical care in a never-ending cycle

Accelerating the translation of new insights into clinical practice

Improve individual patient care with cohort-level knowledge

Improve translational research by enriching cohort information



A great example of Population Genomics @ scale

The 100,000 Genomes Project

The combination of a large UK heterogeneous population, world class genomics science and an NHS can deliver ...

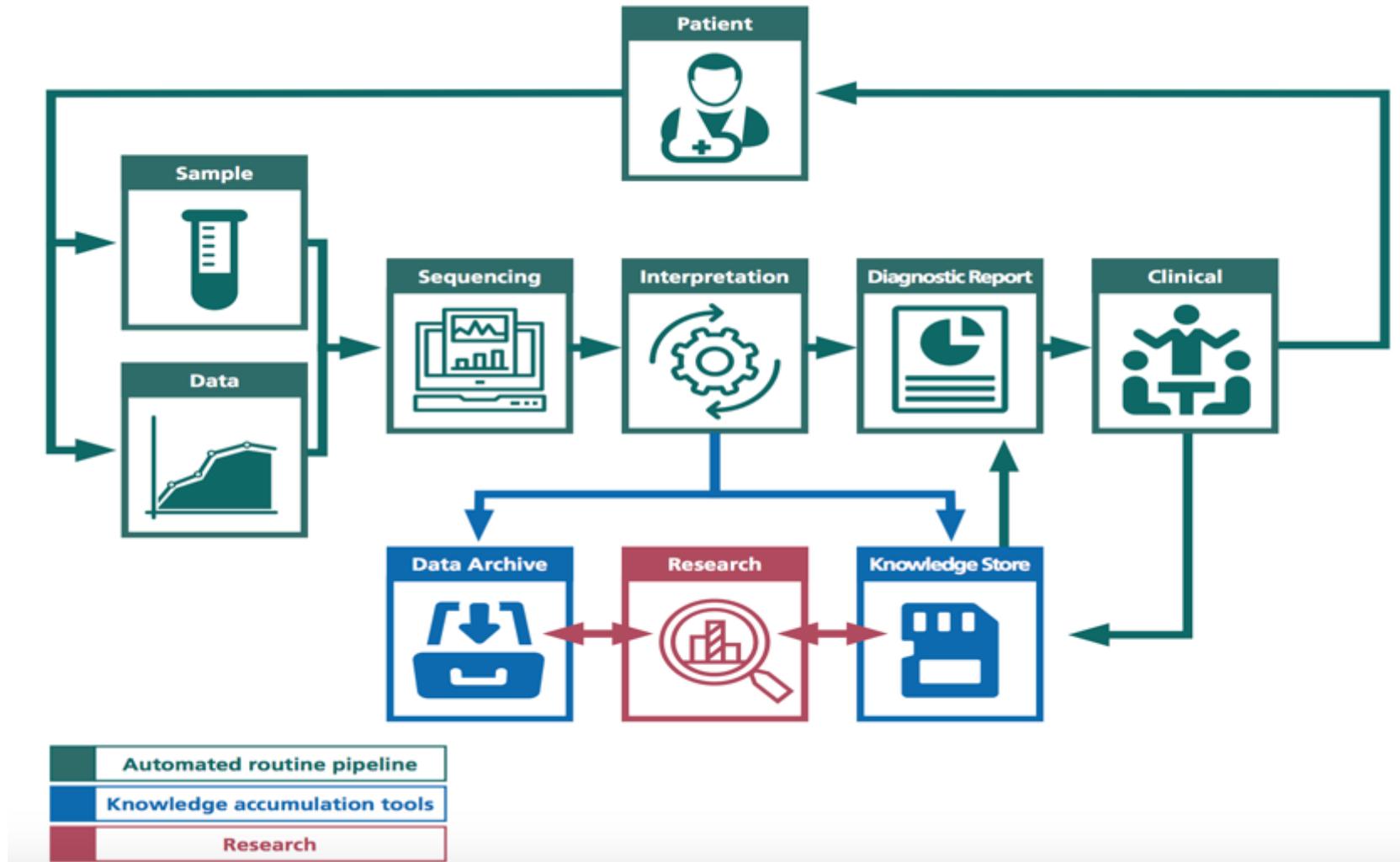
Increased insight and understanding leading to **new treatments, devices and diagnostics**

Advanced genomic medicine practice **integrated into the NHS**

Thriving private sector investment and commercial activity in genomics

All of which require **positive public support**

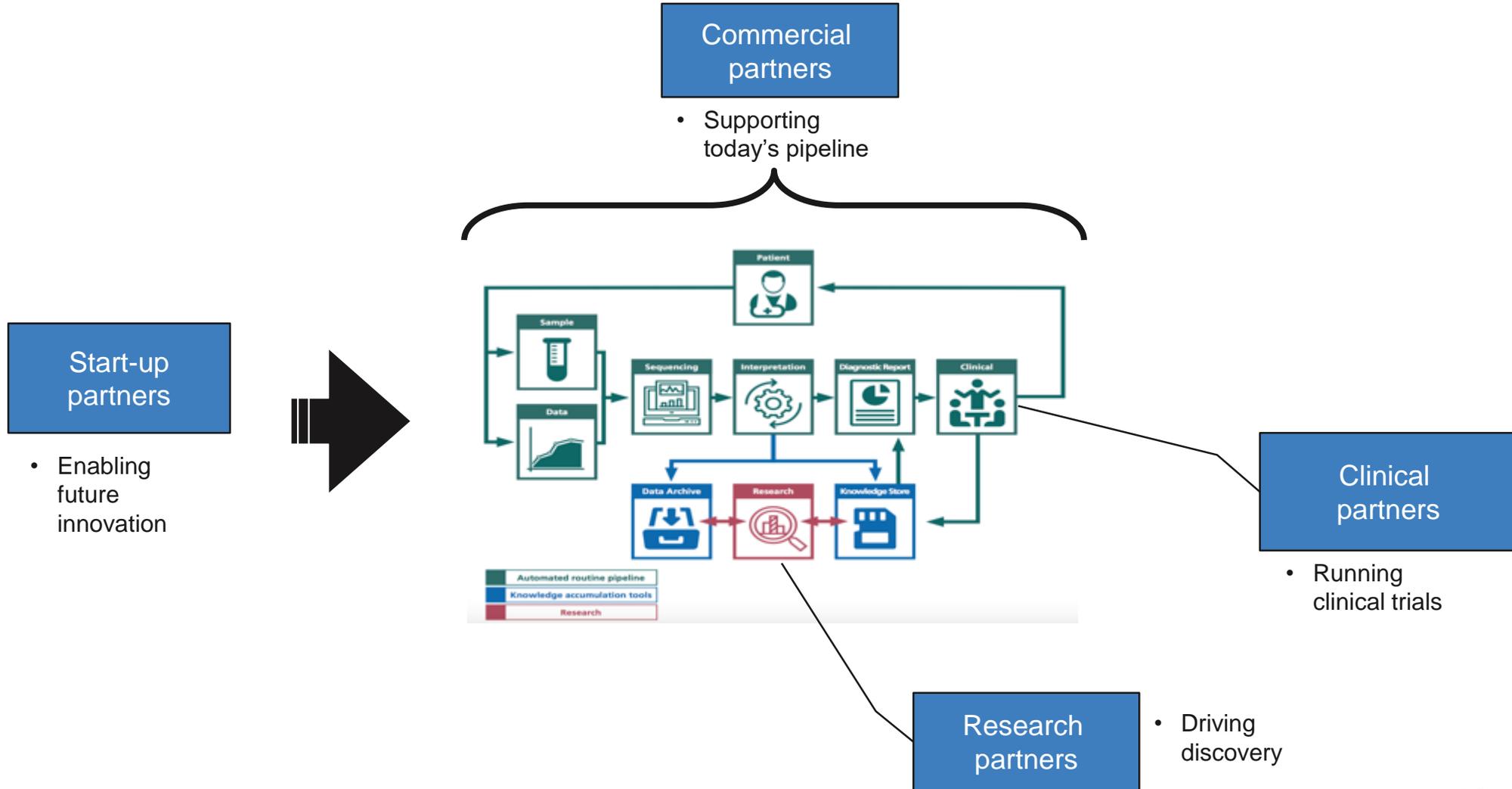
A simplified view on the pipeline for whole genome sequencing at Genomics England



But implementing Population Genomics at scale is challenging

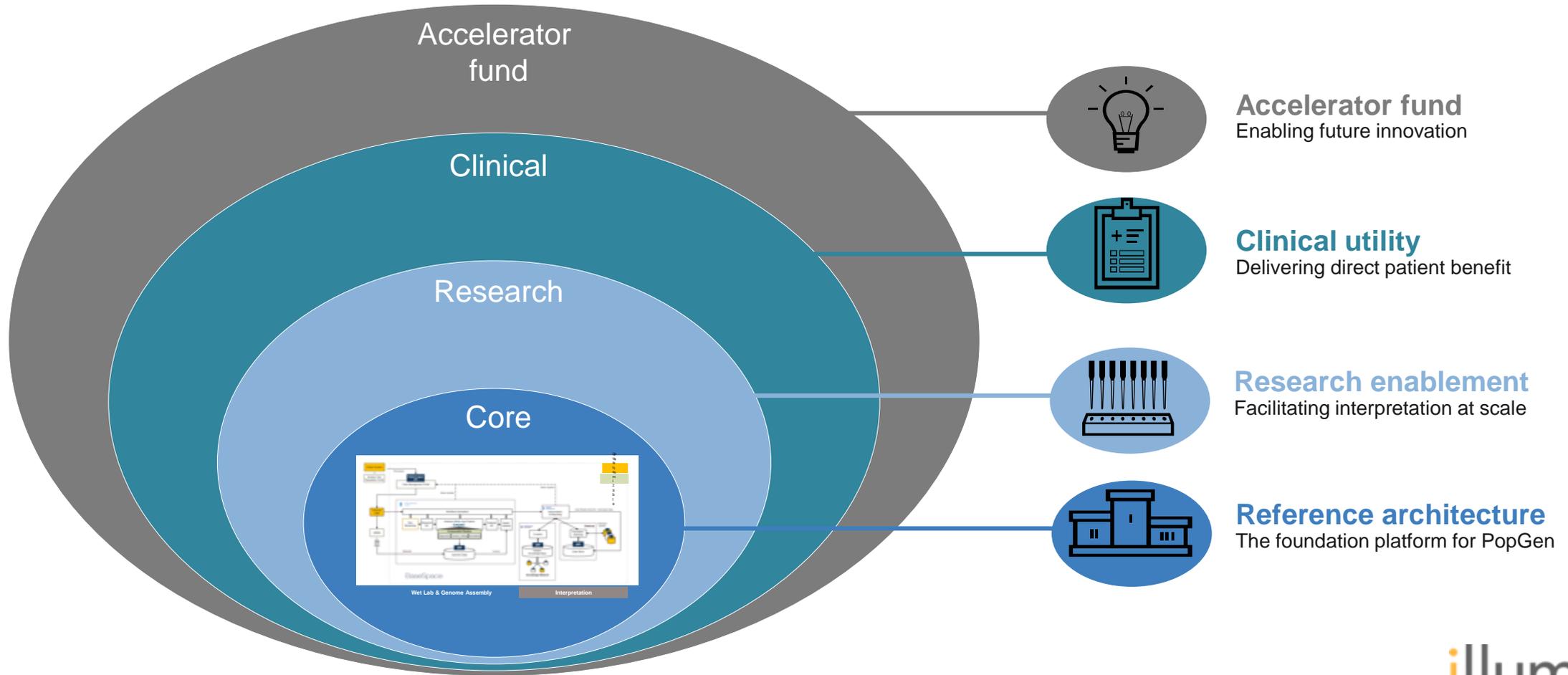
Scientific, Clinical & Societal Challenges	Patient consent, privacy, and ethics	Clinical utility, approach and measurement of outcomes
	Funding, costs, return on investment	Scope and focus
	Confluence of scientific and political leadership	Patience and endurance
Operational & Technical Challenges	Integrating multi-vendor systems at scale	Accreditation and regulatory
	Standardisation across pipelines, disease models and phenotypic ontologies	Interpretation and reporting at scale
	Managed access to data	Translational analytics across 10Ks, 100K, 1000Ks of patients

And requires an open platform approach to continue to encourage innovation



Convergence around a core but open platform helps to unlock value across multiple domains

A phased approach to creating and capturing value



Where next? The Million European Genomes Alliance (MEGA)

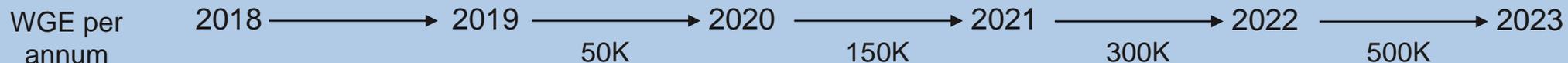
Initiative & funding

MEGA is an EU initiative (funding commitment tbc) with engagement from 17 Member States linking their national-scale genomics endeavours to position Europe as a global leader in the delivery of personalised medicine

Strategy

MEGA will encourage initiation of national-scale PopGen activity for countries in the EU and drive collaboration across countries through supporting an open architecture that involves accredited infrastructure, interpretation and reporting

Size and timing



Objectives

- Prepare healthcare systems for a future of genomic medicine
- Deliver better and cheaper diagnostics and therapeutics for the benefit of patients
- Stimulate investment and commercial activity related to genomics

Key deliverables

- Reference architecture allowing for managed data access across EU countries
- Accelerated interpretation agenda to deliver real patient value
- Aligned regulatory and legislative frameworks

Phased approach

- I. Country specific buy-in with detailed audit of activity to date
- II. Industry alignment and engagement
- III. Country mobilisation and cross-country controlled data access (including outcomes monitoring)

2018 milestones

- EC endorsement of MEGA - signing ceremony in Apr '18
- Industry alignment through EFPIA / IMI
- MEGA launch at EAPM Congress in Nov '18

Industry asks

- Collaborative industry interface through EFPIA / IMI
- Build on existing programmes – don't reinvent the wheel
- New models of partnership with healthcare systems