

# Biobanking for the 100,000 genomes project

BBMRI-UKCTC satellite meeting  
29<sup>th</sup> September



# The challenge

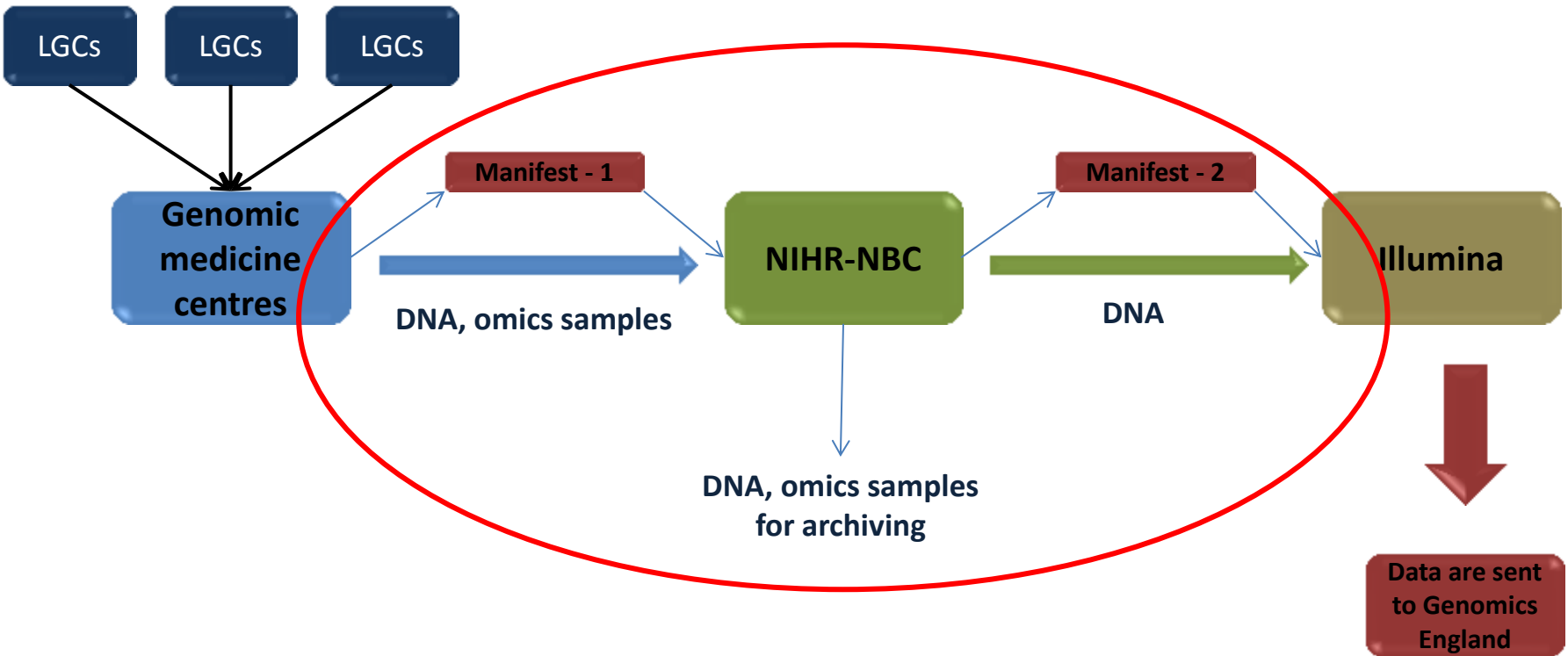
- Scale
- Quality
- Patient to sample/data to clinician
- Impact of pre-analytical variables vs cost of process
- Distributed collaborator network
- Data management
- Time

# The approach

Recruitment, consent,  
collection, processing,  
shipping

QC, formatting,  
normalisation,  
archiving

Sequencing,  
data management



# The solution – NIHR National Biosample Centre

- Support studies involving human volunteers/patients.
- Support NIHR-funded and other academic/NHS research on a cost-efficient basis.
- Researchers can access the full range of biobanking and sample assay services;
  - High throughput, high quality
- Charges are on a fee-for-service basis
- Control of the samples is maintained by the study.
- The range of processes and assays will evolve to the needs of the researcher.



# The service concept

Pre-requisites:  
Quality and consistency in processing and analysis  
Robustness and security of the data trail

Study design	<u>Study operations:</u>		Data analysis
Clinical trials, Cohort studies, Diagnostics, Basic research studies, Public Health initiatives	Process/technology/IT design and testing  Sample collection and logistics Sample processing Sample distribution  Data management Regulatory management Project management H&S Reporting	Sample archiving Sample retrieval Sample assay  Quality management Operations management Financial control	

# Implementing an industrialised approach

## Infrastructure

- supply chain
- logistics
- data

## Organization

- availability of manpower
  - skill levels
- structure of the organization

## Technology

- enabling technology
- technology in parallel activities
- technology equilibrium

## Scientific production methods

- planning and process description
- technical specification and testing
  - tolerances
- standardisation

Fit for Quality/focus on purpose

# The core resource

- High throughput, flexible sample processing.
- Very large ultra-low temperature automated sample archiving capacity – impact on quality and accuracy
- Fully redundant back-up capacity.
- Downstream processing and quality control.
- Fully automated sample curation and distribution.
- Strategic partnerships forming

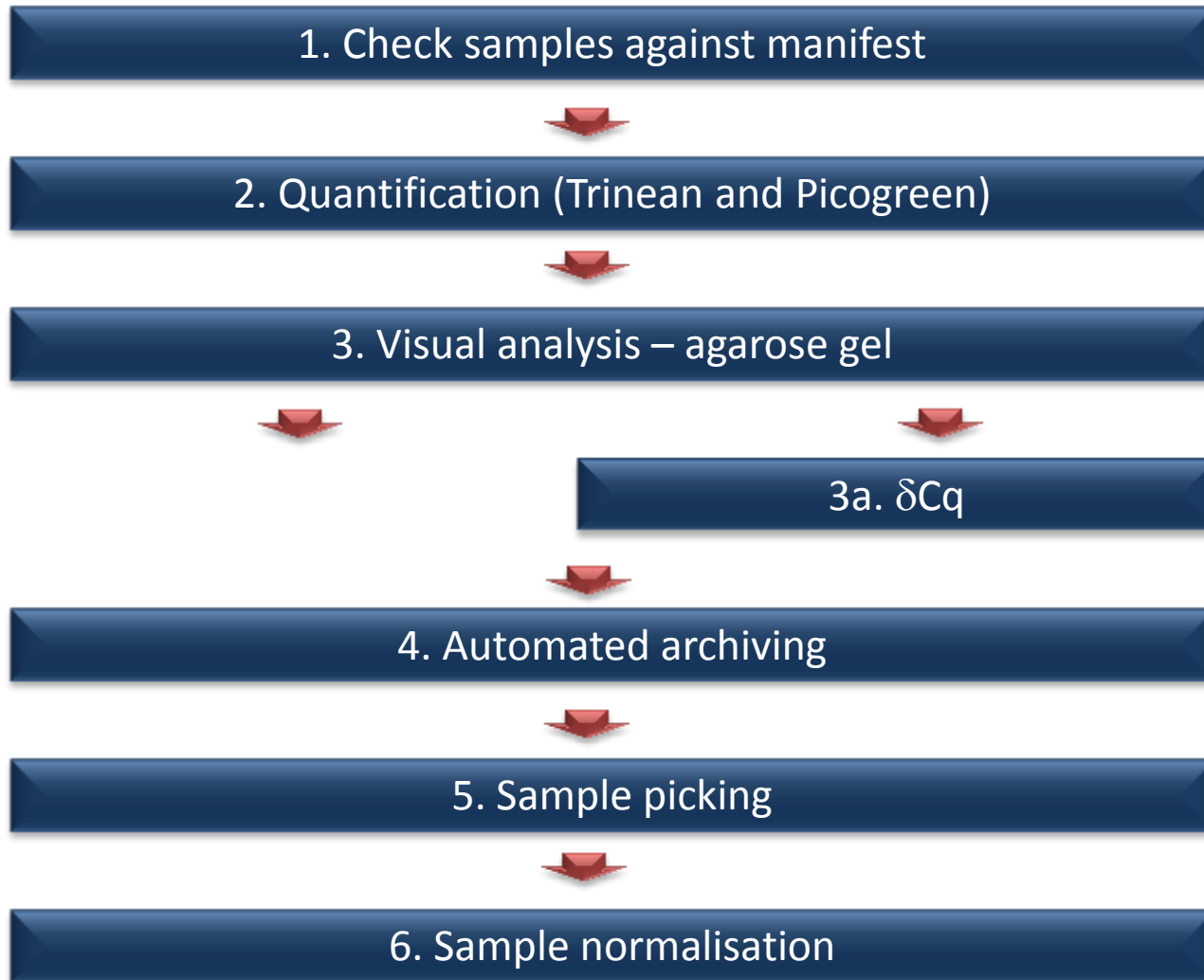
# The challenge of scale

- At peak recruitment, NIHR-NBC will be receiving samples from 500 patients per day
- 500 DNA samples
- 50 plates “omics samples” per fortnight

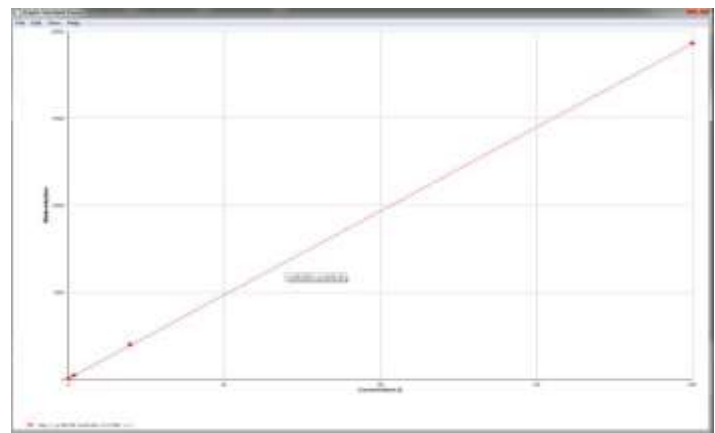
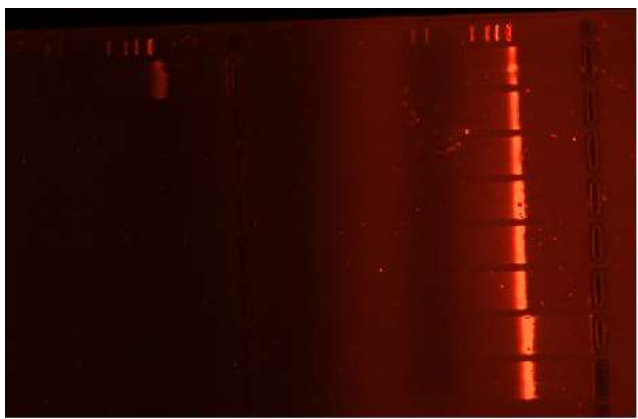
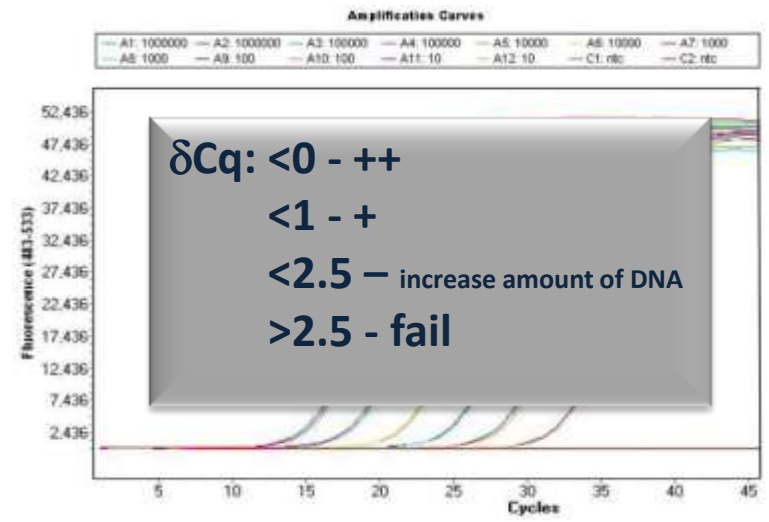
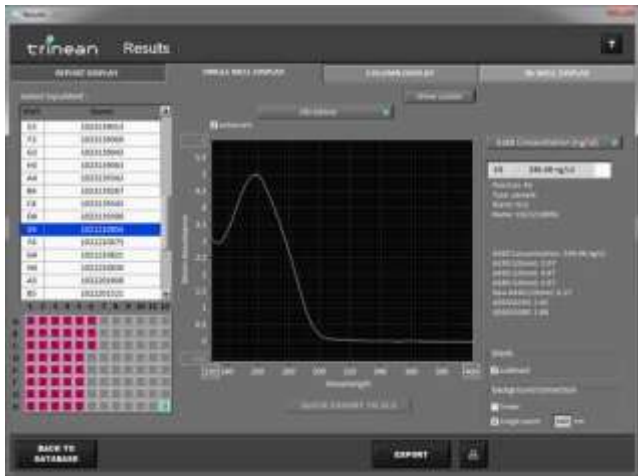




# The challenge of quality



# The challenge of quality



# The challenge of data attribution

## Manifest - 1

### GMC – Genomics England - NIHR-NBC

- Participant ID
- Group ID – identification of trios for rare disease
- Disease area
- Location details/sample details
- Laboratory ID
- Plating sequence

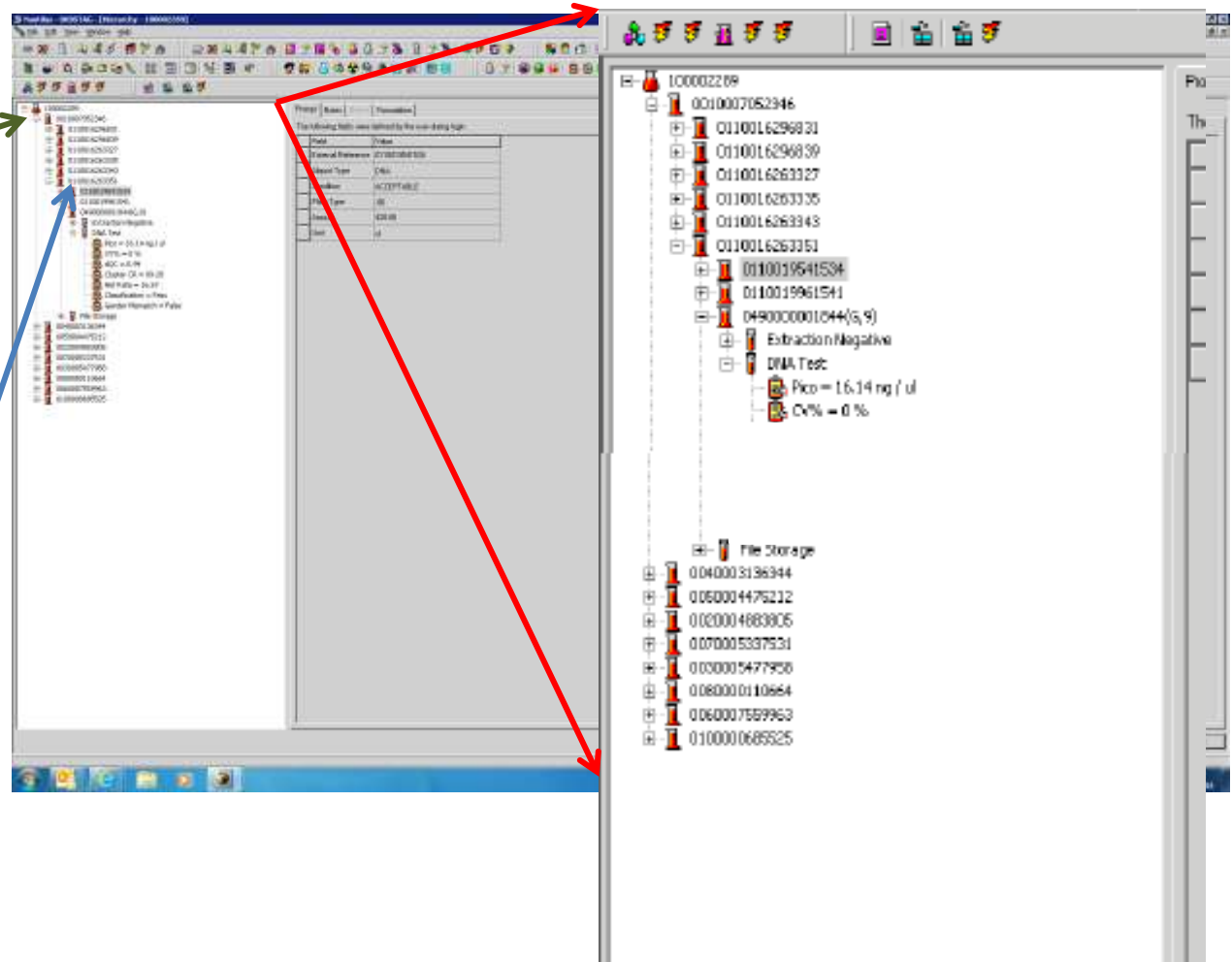
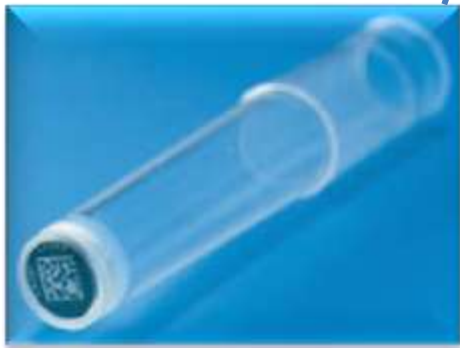
## Manifest - 2

### NIHR-NBC – Genomics England – (Illumina)

- Volume
- ID and Plating sequence
- Genomics England add additional data e.g. gender

# Challenge of patient – data-sample – storage - transfer

Participant ID  
from manifest



The screenshot displays a laboratory information system (LIS) interface. On the left, a patient manifest window shows a list of sample IDs. A green arrow points from the text "Participant ID from manifest" to this list. A blue arrow points from the same text to a specific sample ID in the list. On the right, a sample list window shows a detailed view of a sample, including its ID, extraction method, and DNA test results. A red arrow points from the top of the manifest window to the top of the sample list window.

ID	Value
0010007052346	
0110016296831	
0110016296839	
0110016263327	
0110016263335	
0110016263343	
0110016263351	
0110019541534	
0110019961541	
049000001044(s, 9)	
Extraction Negative	
DNA Test	
Pico = 16.14 ng / ul	
Ct% = 0 %	

File Storage

- 0040003136344
- 0050004476212
- 0020004683805
- 0070005337531
- 0030005477950
- 0080000110664
- 0060007599963
- 0100000685525

# Summary

- Genomics England has established a robust, secure and scalable process.
- Involves multiple partners working to unified standards.
- The processes have been “bedded in” and are approaching rapid scale-up.
- The NIHR-NBC is a critical component in this process.
- It was conceived and implemented for this type of study and has the capacity and infrastructure to support many others.