

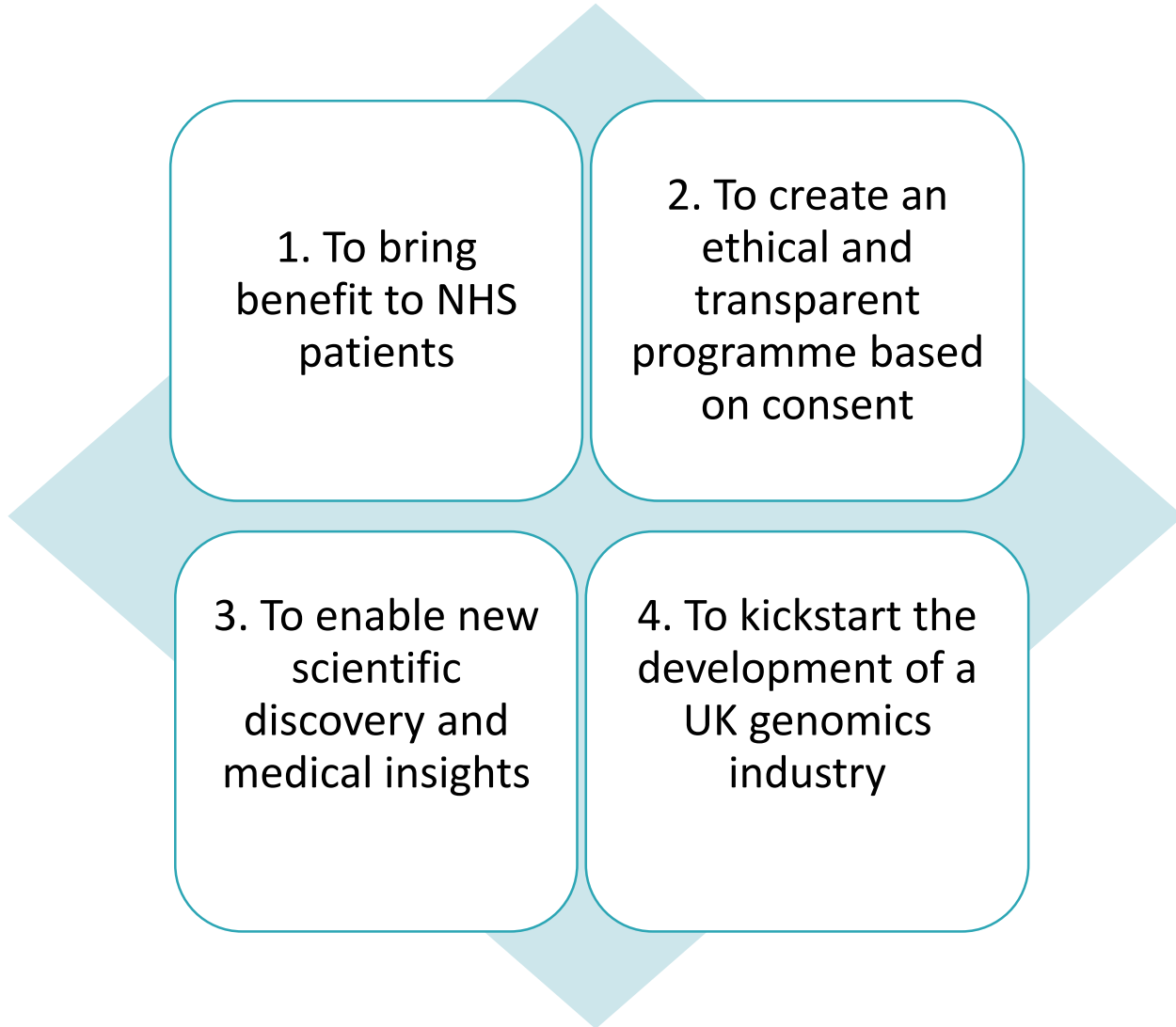
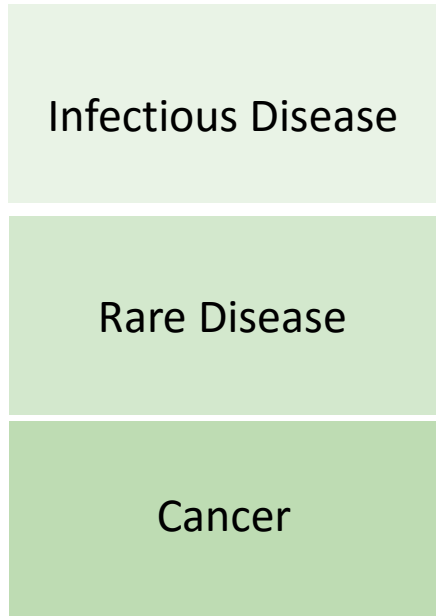
# The 100,000 Genomes Project

Helen Stevens – Sample Handling Manager

16 November 2016

# The 100,000 Genomes Project

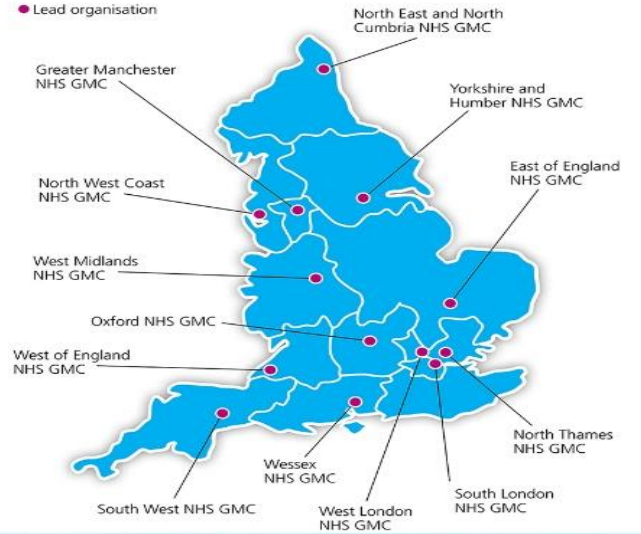
## Four main aims



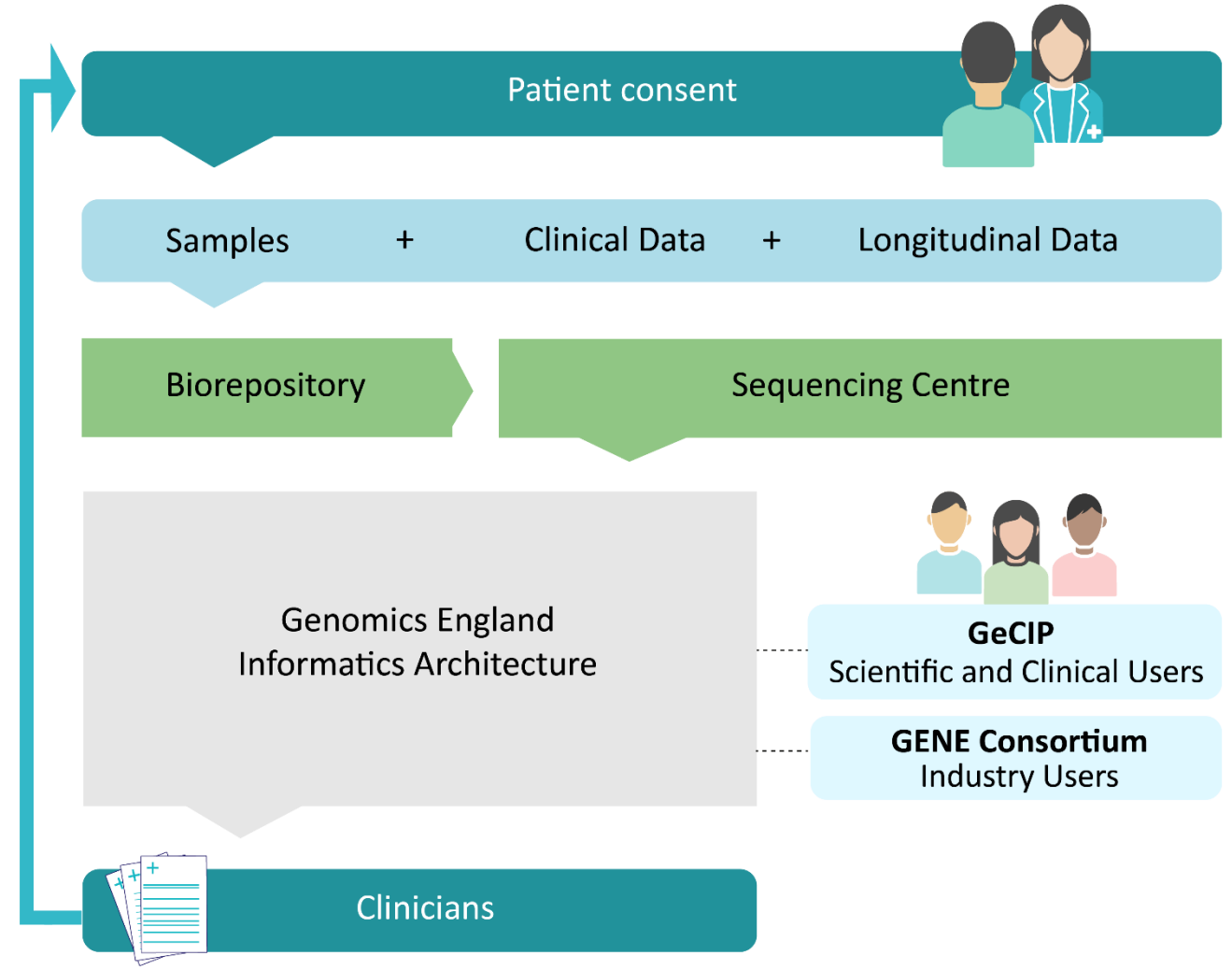
# Genomics England Structure

## NHS Genomic Medicine Centres

Creating a lasting legacy for genomic medicine



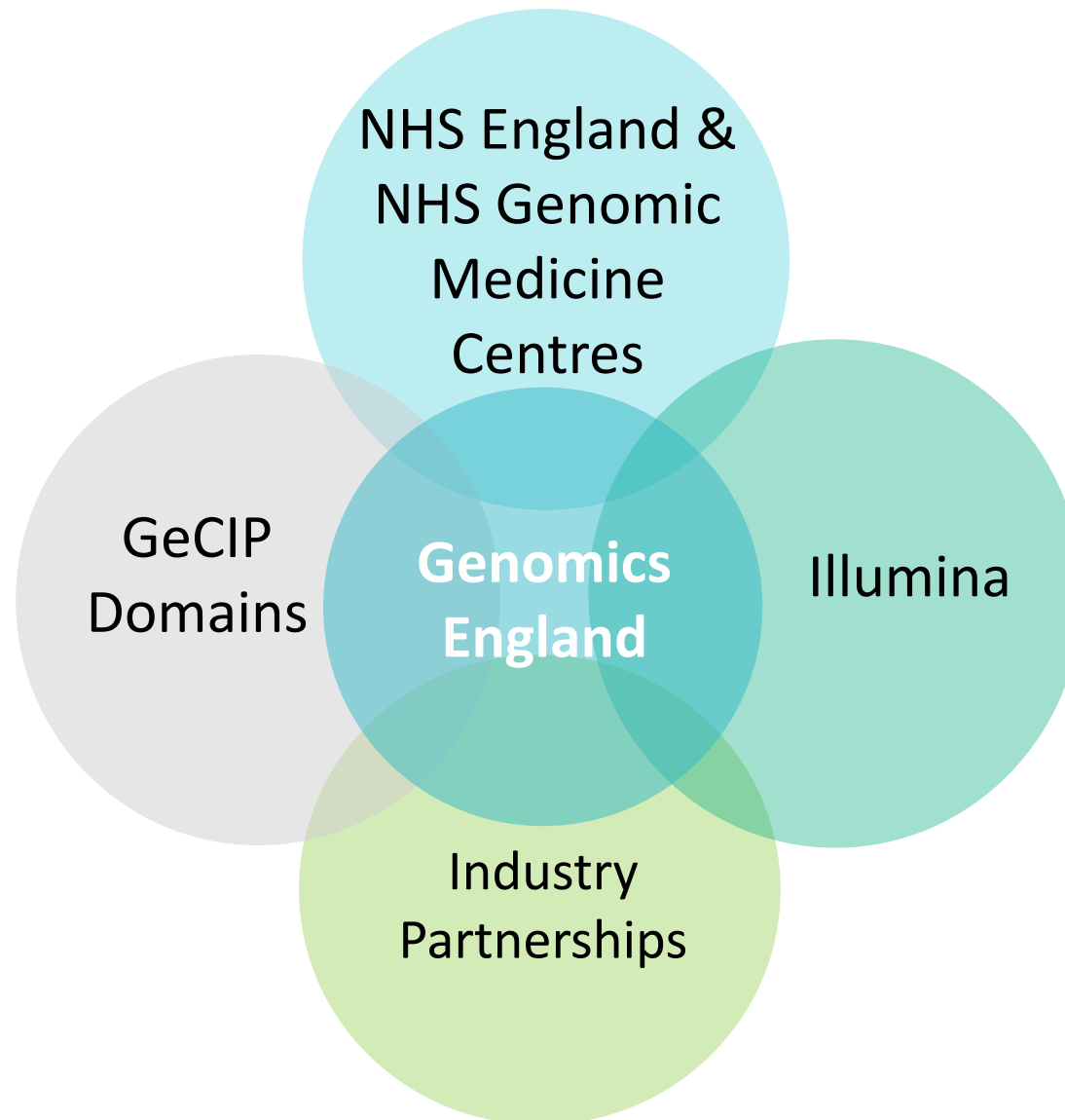
NHS Genomic Medicine Sequencing Centre



# 100,000 Genomes Project: Opportunities – Sample Diversity

- **Routine collection of samples through NHS clinics**
- **Single consent** for clinical diagnostics and research
- Collection of **additional samples** for:
  - Additional DNA - Epigenetics
  - Stabilised Tissue & blood – transcriptomics
  - Plasma, Serum – cell free DNA & metabolomics
- **Serial samples in time**, multiple samples in space
- Collection of **high quality clinical data**
  - Clinical follow-up data
  - Data linkage: life-course data
- **Re-contact patients**

# The Principal Partners



# Key Operational Features



- ▶ Genomics England - a company wholly owned by the Department of Health
- ▶ Sequencing 100,000 genomes
- ▶ Clinical service, major research project and NHS transformation
- ▶ Focusing on cancer, rare disease (and infection - Public Health England)
- ▶ Based on consent
- ▶ Linking phenotypic and medical record data with genome data
- ▶ **13 NHS Genomic Medicine Centres**
- ▶ Data held in a **secure and monitored environment** with strictly regulated access for researchers including industry
- ▶ Genomic medicine **education programme** lead by Health Education England is embedded in the NHS to upskill the workforce
- ▶ Over 2000+ scientists and clinicians engaged through disease-based **Genomics England Clinical Interpretation Partnership (GeCIPs)**
- ▶ Additional funding through MRC, the Wellcome Trust

# Stay in touch



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