

# Trust Board Update

## Genomic Medicine Service

26<sup>th</sup> May 2022

Delivered in partnership with:

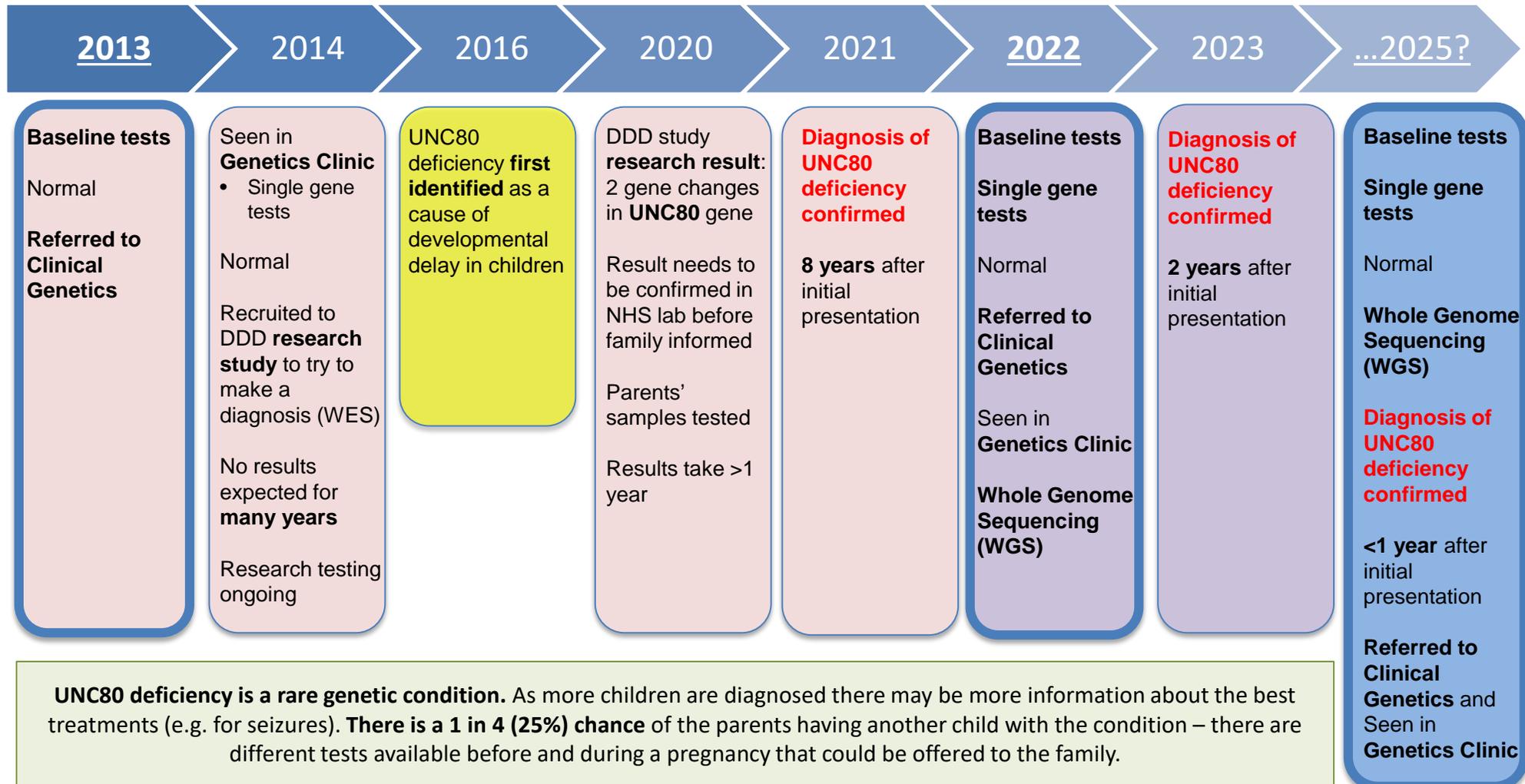
Sheffield Children's  
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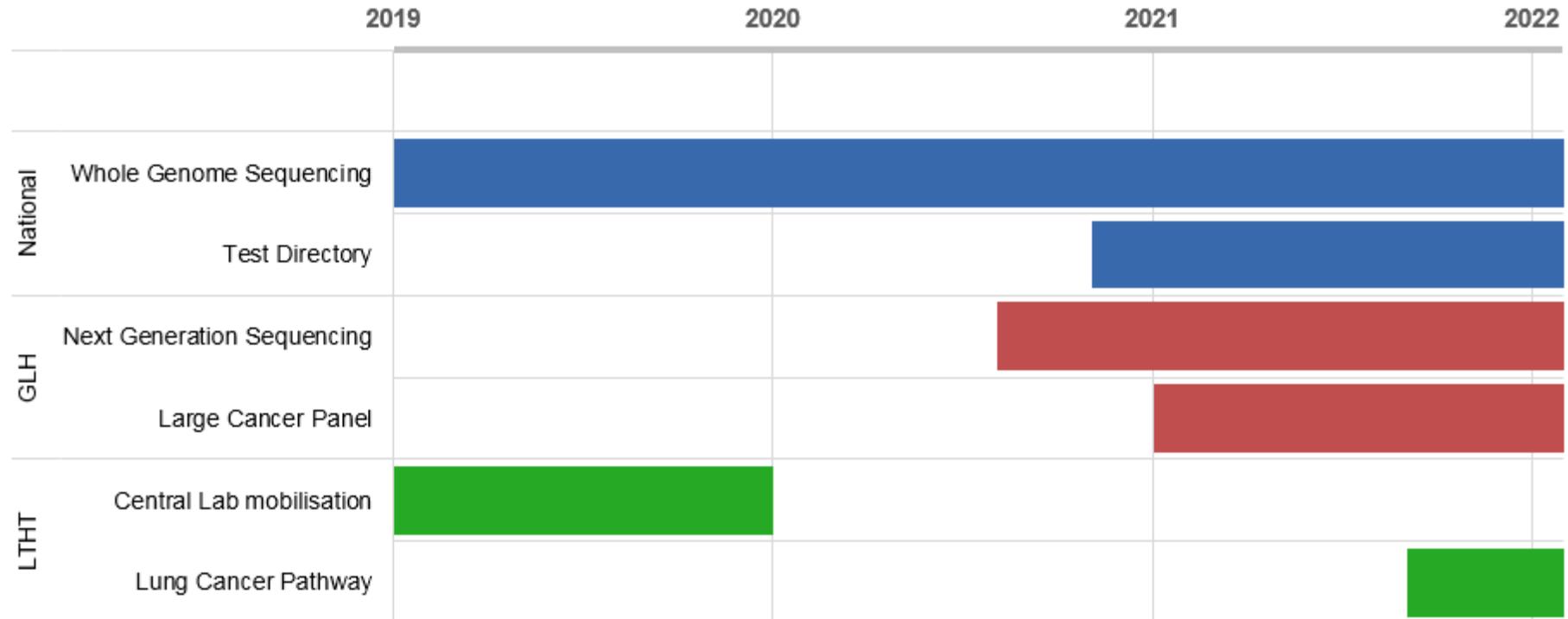
# A patient's journey through a decade of genomics

3-year-old boy, presents to paediatrics with delayed development (not sitting or crawling), lack of speech, fits, poor growth

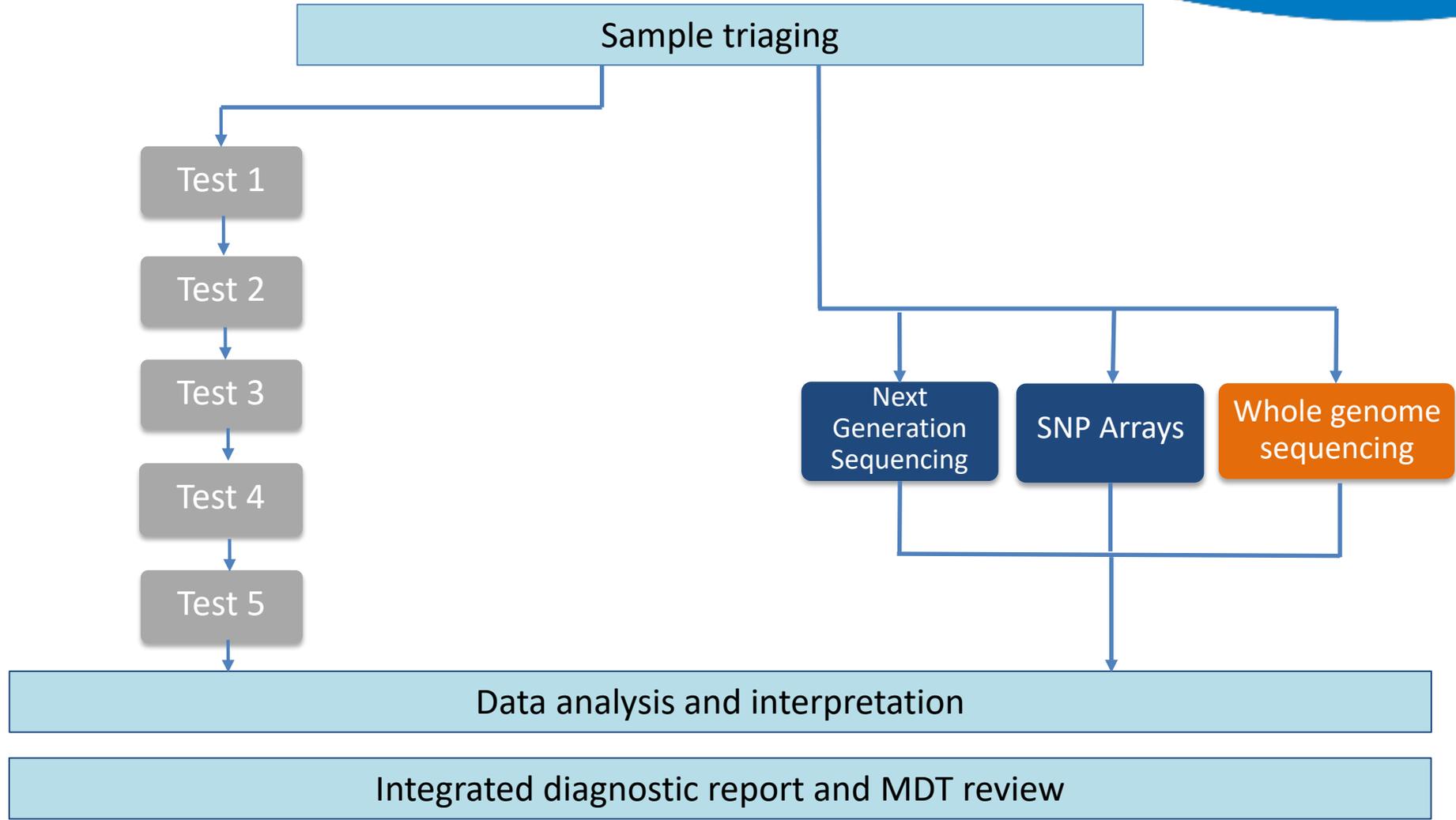


**UNC80 deficiency is a rare genetic condition.** As more children are diagnosed there may be more information about the best treatments (e.g. for seizures). **There is a 1 in 4 (25%) chance** of the parents having another child with the condition – there are different tests available before and during a pregnancy that could be offered to the family.

# Testing Pathway Developments Delivered



# Improved Cancer Diagnostic Pathway



Previous Standard of Care

Individualised patient management

## How are we making improvements?

Transformational, system-wide change driven by  
NHSEI Genomics Medicine Service

Technology  
developments

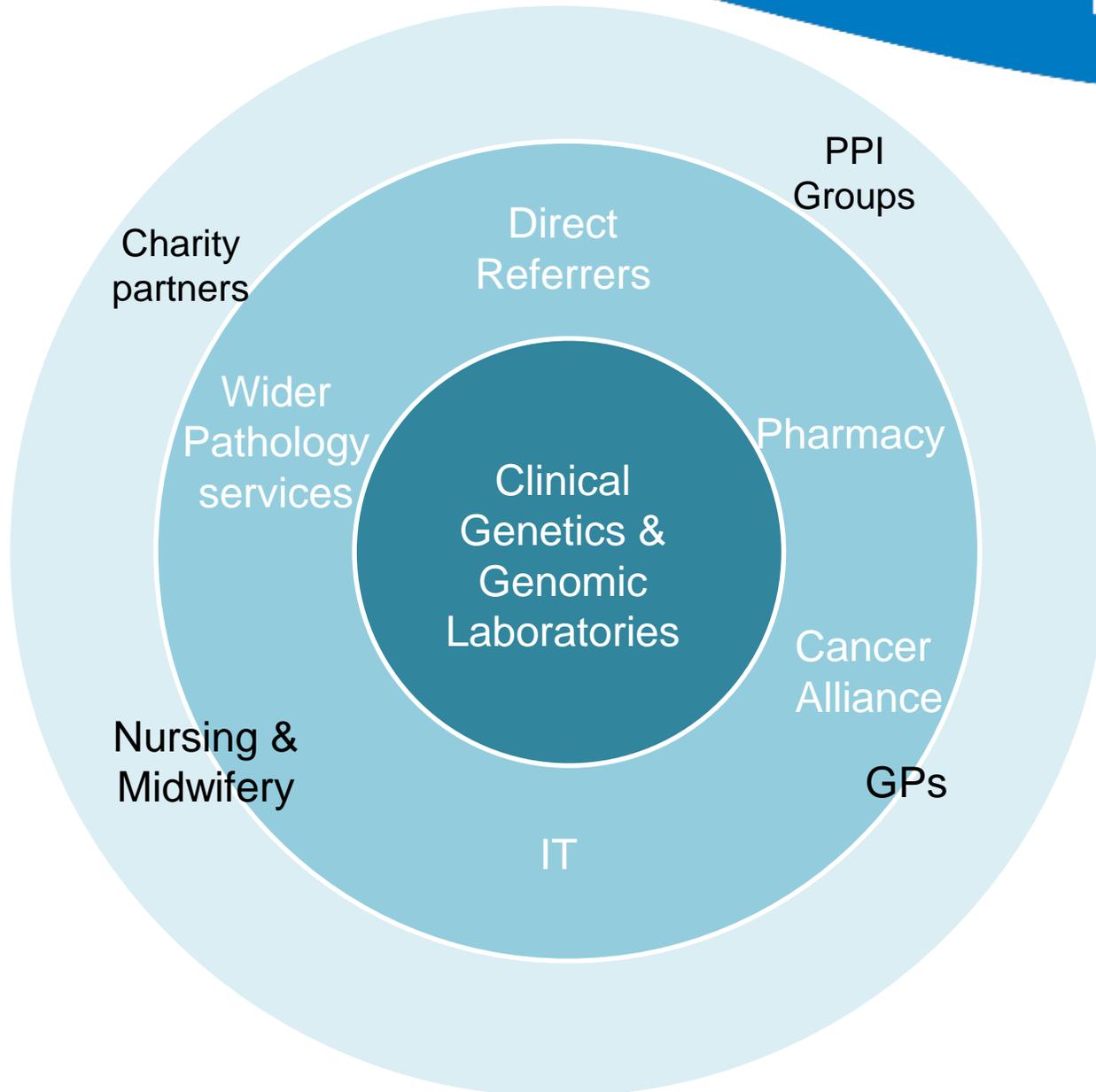
Local Projects

National  
Projects

Laboratory  
Transformation

Collaboration

# Workforce



# GMSA Core achievements in 2021/22

## Integrated Leadership

### Strong Clinical Leadership

- Medical, Nursing, Midwifery, Pharmacy, Research

### Integrated Partnership Board

- Exec level rep from partner Trusts

### Joint GMSA/GLH Executive Team

- Collaborative leadership approach developing

### Cancer Alliances Group

- Common purpose and goals alignment

## Partnerships and Networks

### Stakeholder Engagement

- Partnership mapping and prioritisation
- SRM system to support engagement plan delivery

### Collaboration

- Clinical networks developing, e.g. Pharmacogenomics
- Genomics champions emerging across multiple professions and projects, e.g. Lynch, MD, Nursing

## Workforce – Education and Training

### Baselining Genomics Knowhow

- Nursing and Midwifery – ASSIGN-E self assessment
- Joint GMSA national pharmacy staff survey

### Developments

- Collaborative working with HEE/GEP, RCNi
- Workforce development lead appointed

### Learning Opportunities

- Regular scheduled events on genomics-specific topics
- From Niche to Necessity – nurses/midwives webinar



## Research

- Key themes identified and strategic values and vision agreed
- Workshops, study days held to clarify & shape Research Collaborative offering
- NEY Genomic Research Network in progress & research proposal submissions to Research Collaborative from NEY

## Communications and PPI

### Communications and PPI Strategy

- Approved by NHSE/I and being implemented
- Joint communications advisory group being established

### Patient Involvement

- Interim PPI Chair appointed
- Model of involvement co-designed in collaboration
- Recruitment to Patient Forum progressing

# GMSA Project achievements in 2021/22

## Lynch Syndrome

- Baseline Colorectal and Gynae cancer MDTs survey for genomic tumour testing across NEY
- Regional lists of LS patients collated
- Regional LS Clinical Network – good engagement from NEY GMSA regional LS Clinical Leads
- NEY leading on 3 extension projects



## Rare Disease Case Finding



### Pilot project

- Innovative software used to identify Rare Disease cases in GP records and flag for review
- Aiming to reduce time to diagnosis

## Monogenic Diabetes

- 95% Trusts with trained doctor
- 95% Trusts with doctor champion
- 73% Trusts with trained nurse
- 68% with nurse champion



## Digital DNA



### Discovery phase in progress

- Collaboration with NW GMSA
- Reviewing systems, suppliers, IT work flows
- Region-wide engagement
- Aligned with National informatics work

## Nursing and Midwifery

### Developing nursing knowhow...

- ASSIGN-E assessments completed
- Gaps identified in each GMSA
- Training promoted/delivered for Nurses, Midwives, HVs



## DPYD

### Levels of Testing

- Baseline data established
- Dispensing process understood

### Sharing Best Practice

- Cancer Alliances via PPI Group
- Oncology Research Group (Ncle)
- Pharmacist Networks

# GMSA Familial Hypercholesterolaemia project

Support genetic testing for FH within Primary Care through pathway transformation (support Genomic Test Directory update)

- Engagement of 11 PCNs: PCN Pharmacists case-finding and delivering diagnostic FH testing within Primary Care setting



NEY responsibility for national workstreams:

- Education and Training: national 6-hour training course, collation of FH resources within Genomics Education Programme website: [Transformation project: Familial hypercholesterolaemia - Genomics Education Programme \(hee.nhs.uk\)](https://www.hee.nhs.uk/transformation-projects/familial-hypercholesterolaemia-genomics-education-programme)
- Digitalisation: Mapping of potential digital solutions to clinical pathway development:
- Clinical Pathway Initiative (Genomics Education Programme)



What have we achieved within the project?

- 11 PCN pharmacists trained and delivering diagnostic FH testing within the Primary Care setting
- Pathway development across Primary Care, specialist FH services and GLH
- Supporting informatics strategies: CDRC searches within S1, accurx development, electronic referral form
- Challenges: Covid-19 vaccination programme, indemnity,



Broader achievements

- Broad stakeholder engagement: across geographical and service boundaries
- Close collaboration with AHSN: AHSN SOP, shared learning, cross-pollination of similar workstreams
- Primary Care leadership
- Increasing engagement and workforce development of Pharmacy workforce
- National Leadership: Jude Hayward and Emma Groves

## Background

- Project driven by clinical need
- **SROs** based at LTHT
- Core multidisciplinary team has a **high proportion of LTHT staff**
- **Collaborative working** across region and specialties



# Transforming Maternal and Fetal Genomics Pathways

“Women and families do not have equitable access to genomics services related to pregnancy across the North East and Yorkshire”

## Aims

- To **reduce the current inequity of access** to prenatal and postnatal genomic testing.
- To **develop end-to-end clinical pathways** for pregnancies with a high chance of a genetic diagnosis
- To create a **multi-professional network** of care providers across the region to **embed genomics in to maternity care** and deliver **culturally competent services** to diverse populations
- **Contribute** to transformation projects in nursing, midwifery and medicine



4 workstreams, running in parallel, across the NEY region

Process mapping and pathway interventions

Best practice guidelines and patient registries

Education and training

Informatics and data collection and analysis

# Central Laboratory Capital investment

2019/20 - £500k equipment + 75k estate works



2020/21 - £1.3m Novaseq & IT infrastructure



2021/22 - £1m equipment

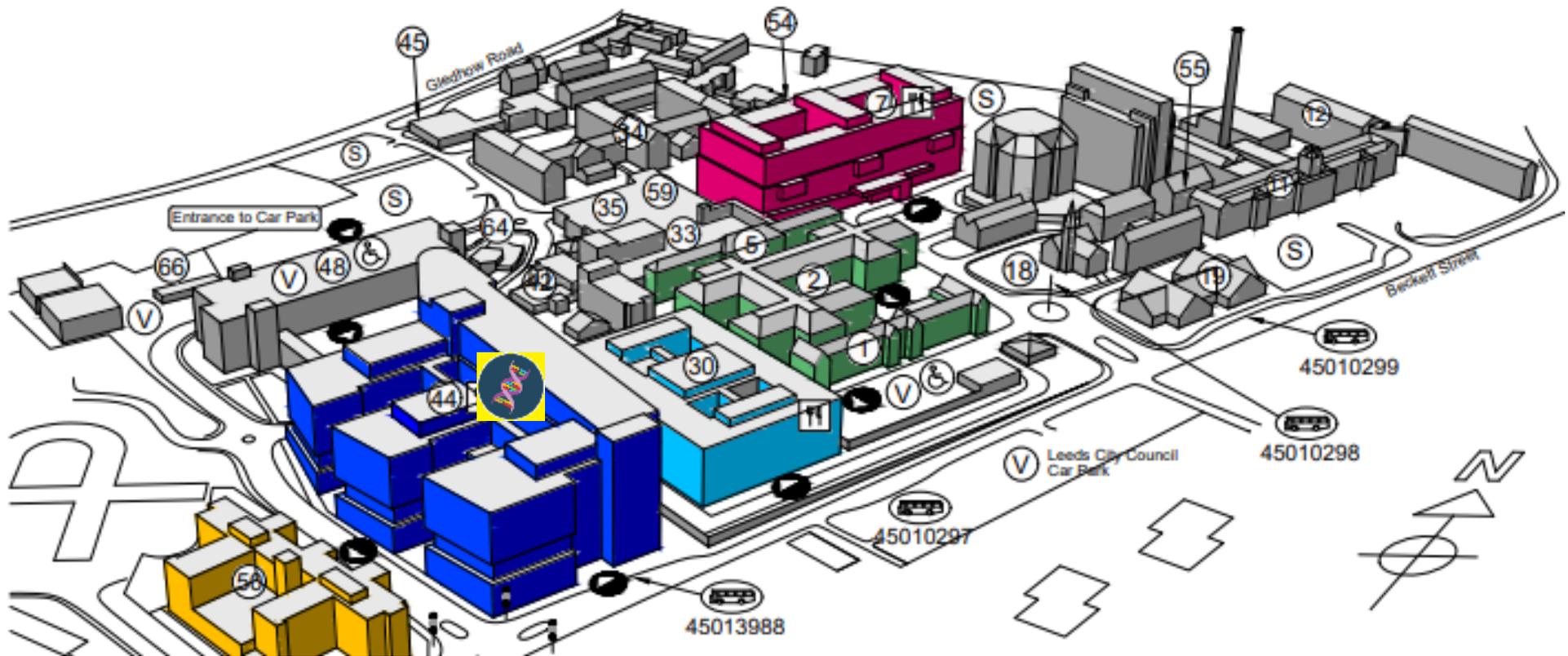


## Rare Disease NGS Pathway

60 individual process steps (in 12 stages)

Approximately 75 hours processing time

Let's see where the sample / paperwork travel...



Challenges	Opportunities	Next Steps
<ul style="list-style-type: none"> <li>• Technology</li> <li>• Changing Test Directory &amp; user education / communication</li> <li>• Demand &amp; Capacity</li> <li>• Shortage of skilled workforce</li> <li>• Estate</li> </ul>	<ul style="list-style-type: none"> <li>• NPIC Genomics England cancer research partnership</li> <li>• Translational Unit shared with University of Leeds</li> <li>• University of Sheffield pre-analysis NHSE bid</li> </ul>	<ul style="list-style-type: none"> <li>• LIMS replacement</li> <li>• North of England IT Strategy</li> <li>• Accommodation development</li> <li>• Cancer diagnostic pathway expansion</li> <li>• Amalgamation of GMSA &amp; GLH Strategic Plans</li> </ul>