

# HEE Genomics Education Programme Nursing and Midwifery Round Table

Monday 27<sup>th</sup> January 2020 - The Wesley Hotel &  
Conference Venue, London.



Developing people  
for health and  
healthcare

[www.hee.nhs.uk](http://www.hee.nhs.uk)

	Item	Lead	Slide
1	Welcome and Introductions	Lord Willis of Knaresborough	
2	National Update	Professor Mark Radford, Chief Nurse (Interim) Health Education England & Deputy Chief Nursing Officer for England, NHS England and NHS Improvement	
3	Update on the NHS Genomic Medicine Service	Professor Dame Sue Hill Chief Scientific Officer & SRO for Genomics, NHS England and NHS Improvement	
4	Embedding Genomics into Nursing and Midwifery Practice	Professor Janice Sigsworth Director of Nursing, Imperial College Healthcare NHS Trust	
5	Genomics Education and Training for Nurses and Midwives	Dr Anneke Seller Scientific Director, Genomics Education Programme, Health Education England	
6	Views from the Waiting Room: reflecting on patient and family experience of genomics to date	Leanne LeRiche, 100,000 Genomes Project Participant Panel member and parent	
7	Macmillan Genomics Portfolio of Activity	Dany Bell, Strategic Advisor Treatments, New Medicines and Genomics, Macmillan Cancer Support	
8	Mainstreaming in practice – The Polyposis Registry example	Vicky Cuthill, Lead Nurse/Manager, The Polyposis Registry and Family Cancer Clinic, St Marks Hospital	
9	Discussion: Awareness, Reach and communication	Professor Mark Radford, Chief Nurse (Interim) Health Education Eng;and & Deputy Chief Nursing Officer for England	
10	Next Steps, Reflections and Close	Lord Willis of Knaresborough & Professor Mark Radford, Chief Nurse (Interim) Health Education England & Deputy Chief Nursing Officer for England	
11	Close		

## Introduction

The Chair, Lord Willis of Knaresborough, welcomed everyone to the meeting.

Professor Mark Radford, Director of Nursing HEE & Deputy Chief Nurse NHSE&I was introduced to the Round Table members.

Lord Willis briefly explained the background and work of the Round Table and asked attendees to contribute to the discussion with views, suggestions and ideas to continue to take this agenda forward.

Lord Willis complimented colleagues on the fast pace of progress and implementation of the Genomic Medicine Service, commenting that a great deal has happened since the first Round Table meeting in January 2017.



## **Professor Mark Radford, Director of Nursing HEE & Deputy Chief Nurse NHSE&I - National update**

Professor Radford, updated the group on the national plans and standards affecting the nursing and midwifery professions.

The building of services across specialties is evident in the NHS Long-Term Plan. How the workforce operates will change over the next 10 –15 years. Work began 18 months ago on the People Plan and a national workforce strategy is being drafted, which will set out the vision for how those working in the NHS will be supported to deliver care. The government is committed to increasing the nursing workforce, but we have a ‘once in a generation’ opportunity to look at what healthcare practitioners are doing in practice, rather than thinking solely about workforce numbers.



The nursing and midwifery standards mark the beginning of a fundamental change for how we train nurses and midwives and equip staff with skills that are adaptable as healthcare evolves.

There will be one committee nationally looking at how the standards are delivered. With the addition of genomics in the NMC standards, it is essential that we educate future and current nurses, midwives and AHPs so that they at the very least understand what genomics is, its implications for healthcare, patients and their families and how/where to signpost to specialist services. This key area of the workforce will play a crucial role in mainstreaming genomics in healthcare and supporting patients.

# **Dame Prof Sue Hill- Update on the Genomic Medicine Service (GMS) and Long-Term Plan**

Professor Dame Sue Hill (NHS England and Improvement) provided an update on the NHS Genomics Medicine Service (GMS).

The Government have agreed to triple investment to NHSE/I for the GMS over the term of the Long-Term Plan. Baroness Blackwood is leading on the genomics element of the NHS People Plan including academic investment, plans to work with industry and the devolved nations.



The aim of the GMS is to have a national approach to patient choice, a single mandated genomic test directory, standards, contracts and consistent KPIs. 7 Genomic Laboratory Hubs (GLHs) will drive the rapid adoption of the service, building on the model used in the 100,000 Genomes Project and working across geographies. From the 1<sup>st</sup> April 7 Genomic Medicine Service Alliances will also be created.

The Genomics Clinical Reference Group (CRG), chaired by Professor Bill Newman, Professor of Translational Genomic Medicine in The Manchester Centre for Genomic Medicine at the University of Manchester will finalise the annual review process for the National Genomic Test Directory, developing the future operating model for the regional clinical genetics' services, and supporting mainstreaming genomic medicine across end to end clinical pathways and clinical specialties. This group will also look at the potential for the nursing role and how this could link with genetic counselling.

# NHS Genomic Medicine Service Update

Professor Dame Sue Hill,  
Chief Scientific Officer and Senior Responsible Officer for  
Genomics

Email [sue.l.hill@nhs.net](mailto:sue.l.hill@nhs.net) Twitter @CSO

Nursing and Midwifery Roundtable  
27<sup>th</sup> January 2020



# The NHS Long Term Plan has set the strategic focus on genomics for the next 10 years



## National genomics healthcare strategy will set out broader government ambition

### Technology

Targeted investment in areas of innovation that we believe will be transformative, particularly genomics

NHS Genomic Medicine Service will sequence **500,000 whole genomes by 2023/24**.

Improving recruitment to clinical trials and supporting research

### Cancer

Rapid Diagnostic Centres  
Access to personalised care  
Stratified follow-up pathways  
Earlier diagnosis

Extended access to molecular diagnostics

**Genomic testing routinely offered to all people with cancer**

### Children & Young people

A strong start in life for children and young people

**All children with cancer to be offered WGS**

Seriously-ill children likely to have genetic disorder to be offered WGS

### Cardiovascular

Early detection and treatment  
Rapid identification of high-risk conditions

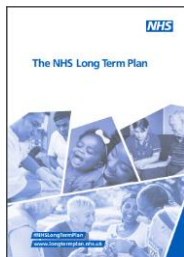
Expanding access to genomic testing for **Familial Hypercholesterolaemia**

### Research

Patient benefit from research and discovery

**Linking and correlating genomic, clinical data and patient data**

Provide routes to new treatments, diagnostic patterns and help patients make informed decisions about their care



# NHS Genomic Medicine Service





**The NHS Genomic Medicine Service will provide consistent & equitable care for the country's 55 million population with:**

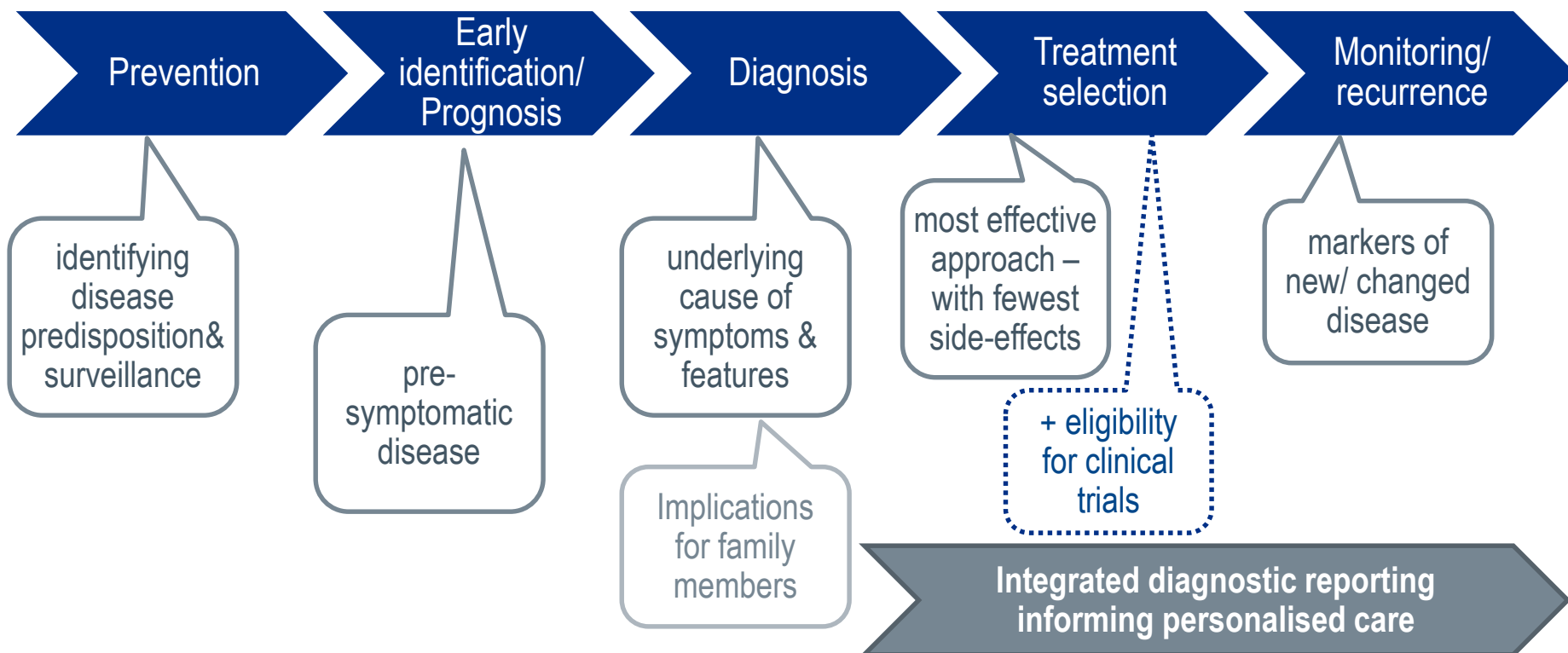
- **National approach** to patient consent, patient & public involvement & strong ethical framework
- **Common national standards**, specifications, protocols, contracts, activity monitoring & pricing frameworks
- **A consolidated national genomic laboratory network** made up of seven Genomic Laboratory Hubs with quality at the core & driving rapid adoption of technology
- **Partnerships across local providers to support front-line delivery** -building on NHS GMC model & integrated with clinical specialties across secondary/tertiary care to primary care & with linked workforce development

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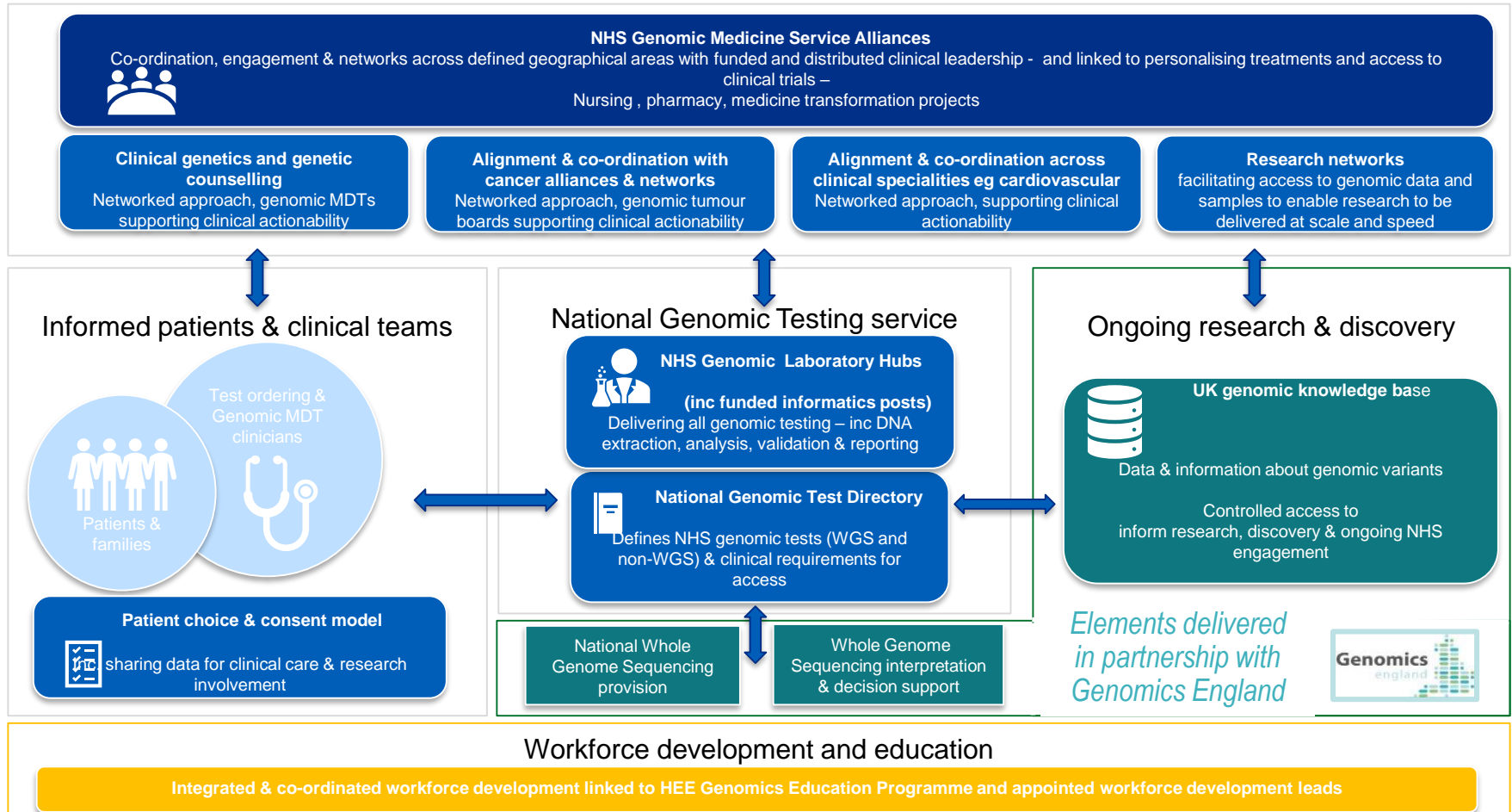
- **A single national testing directory** – covering use of all technologies from single gene to whole genome sequencing inclusive of cancer.
- **A national genomic knowledge base** to inform academic & industry research & discovery including clinical trials recruitment.
- **Single, national co-ordination and oversight of the service** from the genomics unit in NHS England & NHS Improvement

***Launched in October 2018 with an 18 month transition period***

# Mainstreaming : A focus on end-to-end pathways



# The new national genomic infrastructure



# Establishing the national laboratory hub network



**Establishing clear governance structures, an end state delivery model, consolidation of testing within hub laboratories for both rare disease and cancer and associated workforce and organisational development** –includes planning across geographies reflecting new boundaries and workforce development



**Testing and integrating the national genomics informatics system** includes end to end and safety testing , mapping Genomics Test Directory to Hub laboratory LIMS systems, establishing data entry practitioners, training future users



**Delivering a high quality and efficient service and ensuring equitable access to testing** including planning for core and specialist test provision and repatriation of testing ; establishing referral pathways across the geography; plans for capturing patient choice & key clinical data; monitoring of turn around times; benchmarking practice ; model for supporting genomic MDTs



**Contributing to innovation and future genomics projects & broader NHS Long Term Plan commitments** inclusive of FH, Lynch syndrome testing, introduction of DPYD testing and any additional genomics projects through Life Sciences Industrial Strategy & with Genomics England



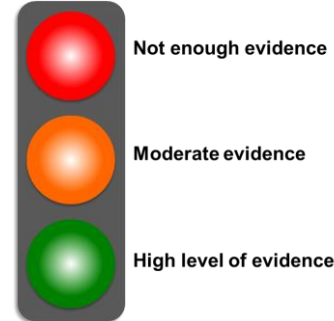
**New contract, data and funding model inclusive of cancer linked to NHS Genomic Test Directory** with additional funding to support cancer panel development and

# Standardisation of non-WGS rare disease testing & analysis

- Tests for over 300 rare disease clinical indications covering circa 3200 rare diseases identified across 18 test technologies with some 25% previous testing replaced by newer technologies.
- Estimated activity based on historical levels plus incidence and prevalence of conditions and expected presentation of unmet clinical need
- Identifies core tests provided by all 7 Genomic Laboratory Hubs (59 clinical indications) together with 16 specialist test areas (283 clinical indications) provided by a limited number of GLHs.
- On 1<sup>st</sup> April 2020 testing will move to the new model

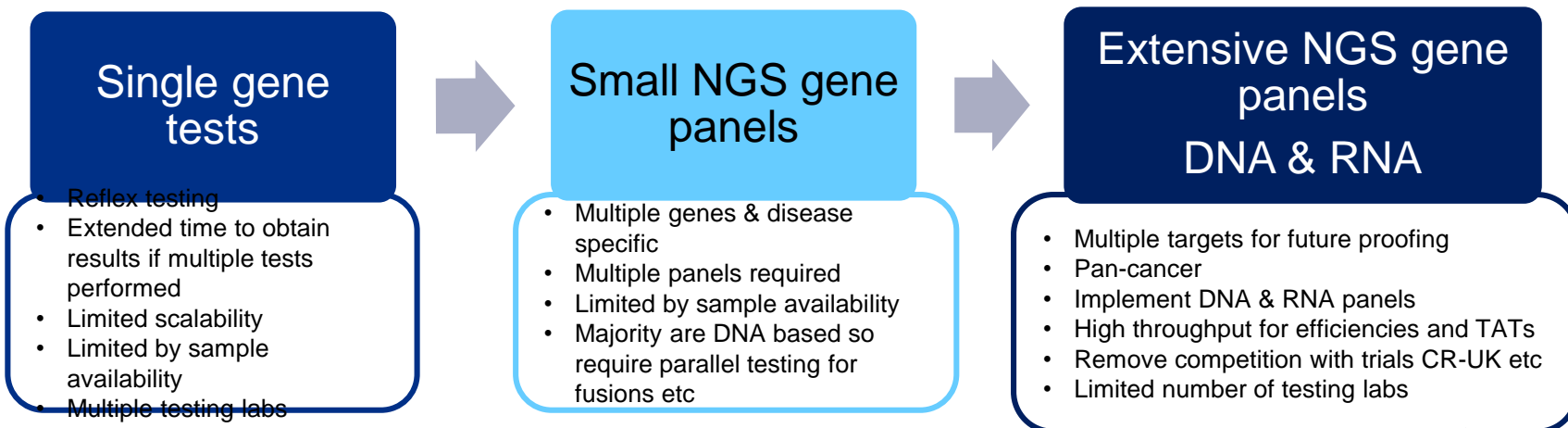
## Gene panel consensus

- 155 panel tests on the National Genomic Test Directory
- Tests split into one of 16 specialist test groups (2 to 4 GLH providers) and neurology and core tests (all 7 GLH providers)
- Working groups formed for each specialist group – clinicians and scientists from the GLHs that will be providing the tests plus other renowned experts
- Genomics England PanelApp used as the platform to gather consensus for the diagnostic grade genes to be included on each panel

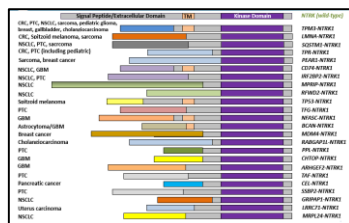


RD Clinical Indication Groups	Number of specialist testing GLHs
Cardiology	4
Endocrinology	3
Eyes	3
Gastrohepatology	2
Haematology	4
Hearing	2
Immunology	2
Inherited cancer	3
Metabolic	3
Mitochondrial	3
Musculoskeletal	3
Neurology	7
NIPD	2
Renal	2
Respiratory	4
Skin	2

# Standardisation of cancer panel testing



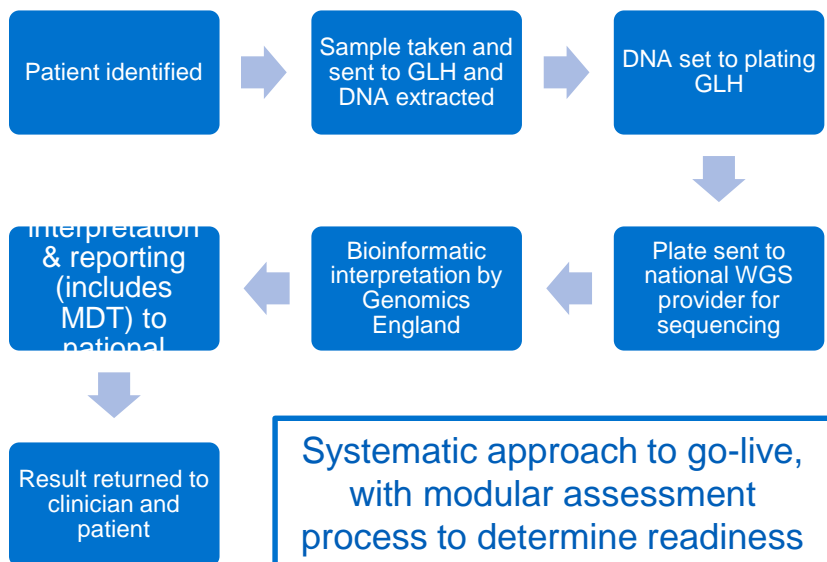
**NTRK gene fusions** are associated with  $\geq 20$  tumor types (1% of solid tumours)  
 Awaiting NICE approval of pan-cancer drugs  
 Require clinical testing for eligible patients from outset



## Biomarker testing – gathering pace

- BRCA testing in other tumours*
- Tumour Mutation Burden for stratifications*
- Mismatch repair deficiency genes*
- HRD scars*
- RET - pan-cancer drug awaiting approval*
- Liquid biopsies*

# Introducing WGS into routine clinical services



Test ID	Clinical Indications for WGS – Phase 1	Test ID	Clinical Indication WGS – future phases
R89.3	Ultra-rare and atypical monogenic disorders	R125.1	Thoracic aortic aneurysm or dissection
R27.3	Congenital malformation and dysmorphic microarray and sequencing	R146.1	Disorders of sex development
R29.4	Intellectual disability – microarray, fragile sequencing	R147.1	Growth failure in early childhood
R69.5	Hypnotic infant with a likely central cause	R31.1	Bilateral congenital or childhood onset cataracts
R104.3	Skeletal dysplasia	R15.1	Primary immunodeficiency
R100.3	Rare syndromic craniosynostosis or isolated synostosis	R352.1	Mitochondrial DNA maintenance disorder
R143.4	Neonatal diabetes	R353.1	Mitochondrial disorder with complex I deficiency
R98.2	Likely inborn error of metabolism - target possible	R354.1	Mitochondrial disorder with complex II deficiency
R54.3	Hereditary ataxia with onset in adulthood	R355.1	Mitochondrial disorder with complex III deficiency
R55.4	Hereditary ataxia with onset in childhood	R356.1	Mitochondrial disorder with complex IV deficiency
R59.3	Early onset or syndromic epilepsy	R357.1	Mitochondrial disorder with complex V deficiency
R61.4	Childhood onset hereditary spastic paraplegia	R63.1	Possible mitochondrial disorder - nuclear genes
R83.3	Arthrogyposis	R58.1	Adult onset neurodegenerative disorder
R381.2	Other rare neuromuscular disorders	R60.1	Adult onset hereditary spastic paraplegia
R84.4	Cerebellar anomalies	R62.1	Adult onset leukodystrophy
R85.2	Holoprosencephaly - NOT chromosomal		Hereditary neuropathy or pain disorder – NOT PMP22 copy number
R86.3	Hydrocephalus	R78.1	Congenital muscular dystrophy
R87.3	Cerebral malformation	R79.1	Congenital myasthenic syndrome
R88.3	Severe microcephaly	R80.1	Congenital myopathy
R109.3	Childhood onset leukodystrophy	R81.1	Limb girdle muscular dystrophy
R193.4	Cystic renal disease	R82.1	Neuromuscular arthrogyposis
		R266.1	Proteinuric renal disease
		R195.1	

Cancer WGS clinical indications
Sarcoma
Acute Myeloid Leukaemia
Paediatric tumours



# Rapid exome sequencing for NICU/PICU

- Nationally commissioned service commenced on October 1st 2019 provided by South West Genomic Laboratory Hub through the Exeter laboratory.
- Rapid genomic test for acutely unwell babies/children with a likely monogenic disorder where a genetic diagnosis would change management
- Put in place:
  - Standardised national order form for both NICU/PICU (and Fetal exome )
  - Standardised testing and reporting strategy
  - Benchmarked tariff
  - Protocols agreed by stakeholders
- Currently over 90 referrals
- Approximately 40% diagnosis rate
- Results issued in mean 9 days (range 6-13)
- Outcomes include (a) referral to specialist services (b) annual screening for diabetes or (c) redirection to palliative care



# Meet Henry...

- Premature baby born at 26 weeks' gestation
- Spent 14 months in NICU
- Rasopathy ( *developmental syndromes associated with genes that control signal transduction*) suspected by clinical geneticist
- Exome sequencing identified an *HRAS* disease-causing variant not detected in either parent
- Diagnosis of Costello syndrome
- Increased risk of certain cancers and heart problems
- Rhabdomyosarcoma identified 5 months later and then a liver tumour



# Henry now...



*“Without the exome sequencing, Henry’s diagnoses and treatment would likely have been delayed. Knowing that Henry has Costello syndrome provided the missing piece of the puzzle and means that he receives the medical management that he needs to make sure his needs are met and to help him achieve his full potential.”*

**Lauren Dunn, Henry’s mum**

**Daily Mail**  
**Gene test hope for critically ill babies**

Rare diseases diagnosed within days on the NHS

'Revolutionary' DNA test can detect thousands of rare diseases in children

NHS rolls out new DNA tests for critically ill babies



Breakthrough... DNA

DNA test tonic for sick kids

**i** News > Health

**Babies with rare diseases to benefit from DNA test that will 'speed up diagnosis'**

The technique, known as whole exome sequencing, doubles the chance of a diagnosis and can reveal what is wrong with patients

The Telegraph  
**'Game-changing' NHS tests diagnose sick children in days**

**ILL BABIES' LIFESAVER DNA TESTS**

Genetic mutations pinpointed

# Genomic Sequencing assisting outcome in safeguarding case

**11 year old girl presented with an acute abdomen and found to have a ruptured spleen.**

No history of significant trauma

Minor slip on stairs several days before admission

History of easy bruising and poor wound healing

**Social services were involved as there were concerns about possible non-accidental injury.**

## **Rapid trio WGS**

de novo mutation in COL3A1, consistent with a diagnosis of diagnosis of Ehlers-Danlos Syndrome, vascular type.

Diagnosis removed social service investigation and informed long term management decisions.

# Roles and responsibilities of NHS GMS Alliances



- Clear governance structure with evidence of CEO support from all major providers across the geography operating as part of a **GMS Alliance Partnership Board** and single **national representative**
- Operate an **effective partnership model** across primary, secondary and tertiary care providers to support and oversee the delivery of the relevant NHS Long Term Plan commitments and genomic medicine for the population.
- **Standardise and implement models of care** for genomic medicine across the geography for all conditions included within the national genomic test directory, inclusive of:
  - reducing variability and improving equity of access
  - national consent processes - monitoring uptake and addressing barriers
  - development of data and informatics infrastructure
  - access to clinical genetic and genetic counselling services
  - alignment and ways of working with cancer alliances, PCNs, pathology and other relevant specialist networks
  - genomic medicine multidisciplinary teams
  - access to personalised treatments and interventions based on genomic information

# Roles and responsibilities of NHS GMS Alliances



- Establish **strong clinical direction and multi-professional leadership** for genomic medicine across the geography, creating a clear relationship with the Genomic Laboratory Hub
- Create **strong and effective patient and public engagement and involvement** to raise awareness and enable service users and their families to shape the development of genomic medicine
- Develop and implement a strategy to **create a genomically informed multi-professional workforce** including participation in national programmes for medical, nursing and pharmacy
- **Facilitating genomic research, development and innovation** across the geography through systematic alignment and involvement of clinical and laboratory services, and inclusive of:
  - Completion of legacy activities of the 100,000 Genomes Project
  - active participation within the national genomic research network and delivery of flagship projects
  - coordinating local participation into agreed strategic research collaborative projects and initiatives
  - evaluating impact and outcomes of testing and of genomic medicine initiatives
  - access to clinical trials

# GMS Alliance Programmes

**NHS England is supporting three national programmes of work:**

## Pharmacy programme

- Creating national networks to support and develop the role of pharmacists in genomics and driving personalised medicine through the sharing of good practice and standard job roles

## Medical Programme

- Working with the Academy of Medical Royal Colleges to support the systematic roll out of genomic medicine within clinical pathways and to ensure clinicians have access to the right education and information at the right point

## Nurse & Midwifery led Genomics Collaborative

- Working with 7 Projects across England to systematically and sustainably embed genomics into nursing & midwifery roles and responsibilities



# Patient and Public Involvement

- Ensuring patients and the public are at the centre of the work we do to embed genomics into routine NHS care is crucial to ensure we design and deliver the best possible services.
- **National Level** - the Genomics Unit are currently recruiting a number of Patient and Public Voice Partners – two to sit on the Genomics Programme Board, three for the Genomics Clinical Reference Group, and three for the national test directory working groups.
- **Regional Level** - Genomic Medicine Service Alliances will be expected to set up appropriate governance mechanisms to ensure patients and the public have their voices heard in the set up and delivery of services and research.

# Genomic Strategy Group with Macmillan, Health Education England & other groups



- This group has brought together 11 cancer charities, the NHS, HEE and a network of professionals, patients and internal experts to work together in supporting and designing activities leading to the mainstreaming and launch of the Genomic Medicine Service.
- The aims were:
  - Bringing other cancer charities together to collectively drive this agenda
  - That people with cancer have the information and support they need to make informed decision
  - The workforce is skilled and knowledgeable and can provide person centred care and support around conversations of genomic testing and results.
- **Outcomes so far:**
  - Survey of GP's to assess their knowledge of genomics
  - Scoped information available to people with cancer and professionals on genomics and targeted treatment across all the organisations in the group to identify gaps and sign post to resources across our organisations on websites
  - Access charity user groups to obtain feedback for NHSE on their patient information around genomics
  - Delivered a patient and public involvement event (PPI) and subsequently formed a PPI Genomics group
  - Shaped a cascade training model targeting all professional groups to be launched early 2020

# New Genomics Clinical Reference Group

- Chaired by Professor Bill Newman Professor of Translational Genomic Medicine in The Manchester Centre for Genomic Medicine at the University of Manchester and Honorary Consultant at Manchester University NHS Foundation Trust.
- Multiprofessional membership
- Early priorities for the Genomics CRG will include finalising the annual review process for the National Genomic Test Directory, developing the future operating model for the regional clinical genetics services, and supporting mainstreaming genomic medicine across end to end clinical pathways and clinical specialties.
- The group will also include three patient and public voice representatives who are currently going through the final stages of recruitment.

# Embedding genomics into nursing & midwifery practice



Professor Janice Sigsworth, Director of Nursing, Imperial College Healthcare presented an update on the the nursing and midwifery collaborative work being undertaken.

NHS England and Improvement are supporting three national genomic collaborative programmes of work relating to pharmacy, medicine and nursing and midwifery. Janice emphasised the need to align work across all three programmes.

Professor Sigsworth announced the appointments in the new national team at NHSE/I who will be implementing work over the next 2 years. Marie Batey will support Janice in her role as National Professional Lead for embedding genomics into nursing and midwifery practice. Donna Kirwan will be national clinical midwifery lead, Charlotte Hitchcock clinical lead for Cancer and Gill Moss clinical lead for Rare Disease.

The team will identify and work with the 7 Chief Nurses from the 7 Lead GMS Alliances (once announced). In the meantime the team will start conversations about the work ahead to help build strategies and implementation plans across geographies and plan to continue the successful Masterclasses, which have been organised in collaboration with Health Education England.

# **Embedding Genomics – NHS Nursing & Midwifery Transformation Programme in England**

**Professor Janice Sigsworth @SigsworthJanice**

**National Professional Lead - Nursing &  
Midwifery Genomics Transformation  
Programme**

Lord Willis Round Table - 27<sup>th</sup> January 2020



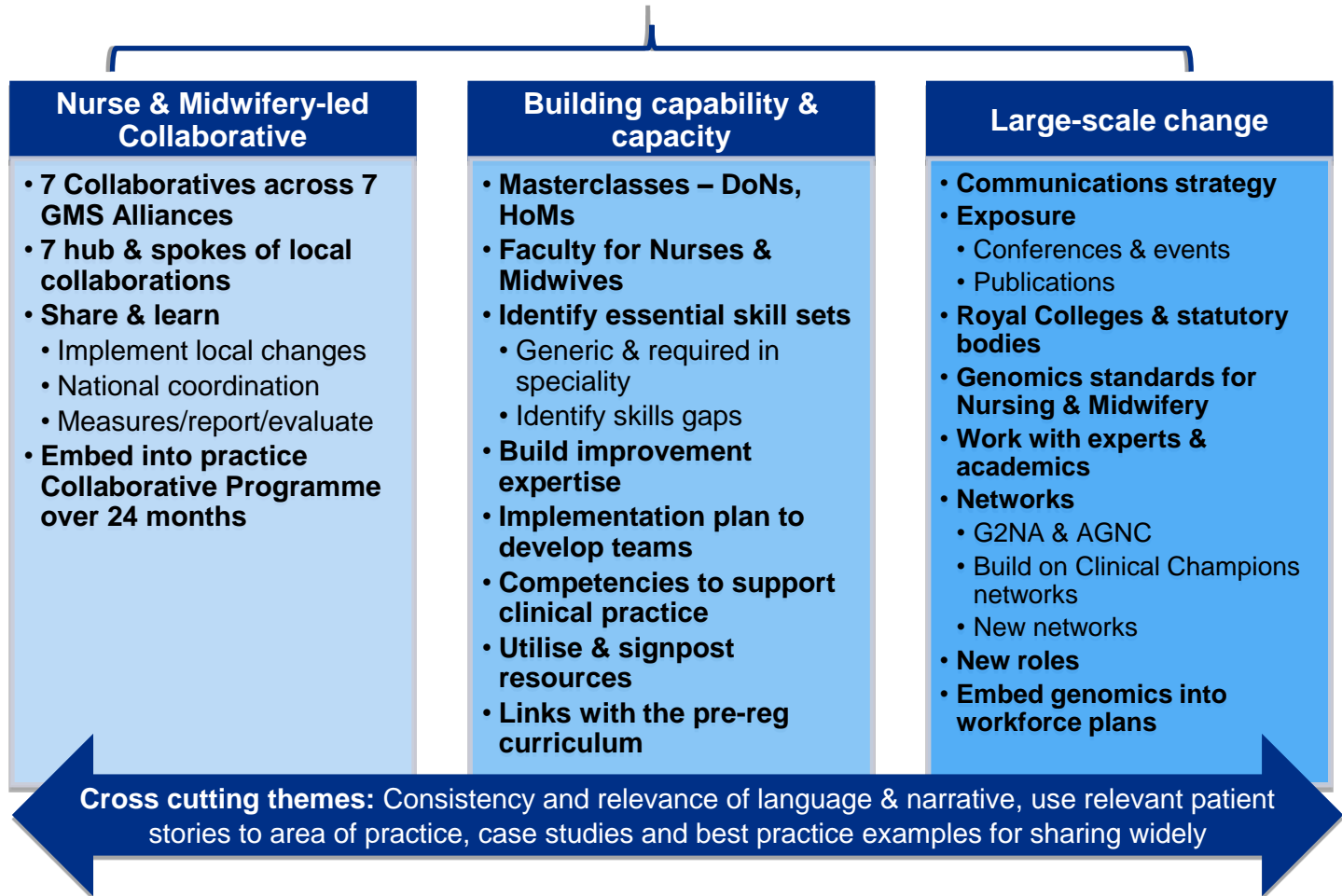
# YEAR 1: Designing the programme

**Ambitious programme to mainstream  
genomics in clinical practice**

To deliver such an ambitious programme across nursing and midwifery professions in England, we have engaged with **experts, clinicians, front-line staff & academics** to test the approach, refine thinking & our approach to implementation

# 3-STRAND APPROACH

## Embedding Genomics into the Nursing & Midwifery workforce



# Leadership



CSO Professor Dame Sue Hill



CNO Dr Ruth May



Professor Mark Radford Chief  
Nurse HEE; & Deputy CNO

**Leadership at *every level*** – key to achieving a step-change  
in clinical care delivery  
**AND**  
**System transformation** across the nursing & midwifery  
professions



# Aim: Embed genomics into nursing & midwifery workforce plans

## Levers - Policy & system level:

- DH – Rt Hon Mathew Hancock MP
- NHS Long-term Plan (Mainstream Genomics)

## Senior leaders:

- CSO Sue Hill + CNO Ruth May NHS
- Chief Nurse HEE/Deputy CSO Mark Radford

## National Professional leadership:

- Professor Janice Sigsworth & Senior Nurse

## Operational System Leaders:

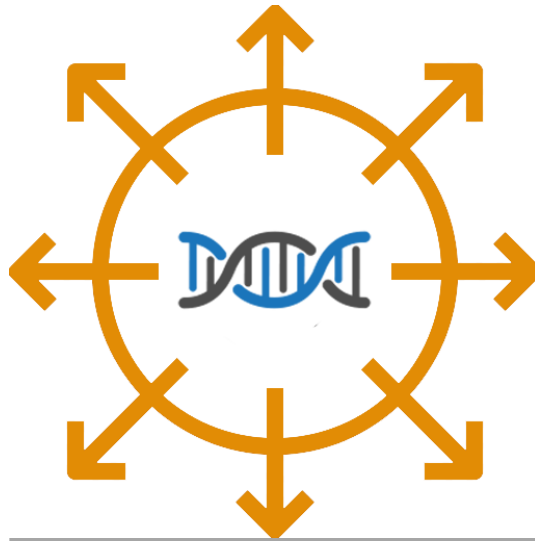
- Directors of Nursing + Heads of Midwifery

## Senior Clinical Leadership:

- Nurses & Midwives - leading change in the Collaboratives



# NHS Genomic Medicine Service Alliances (7)



**YEAR 2**

**Implementing the programme**

# Sustainable implementation: developing service and workforce models



## NHS England is supporting three national programmes of work:

### Pharmacy programme

- Creating national networks to support and develop the role of pharmacists in genomics and driving personalised medicine through the sharing of good practice and standard job roles

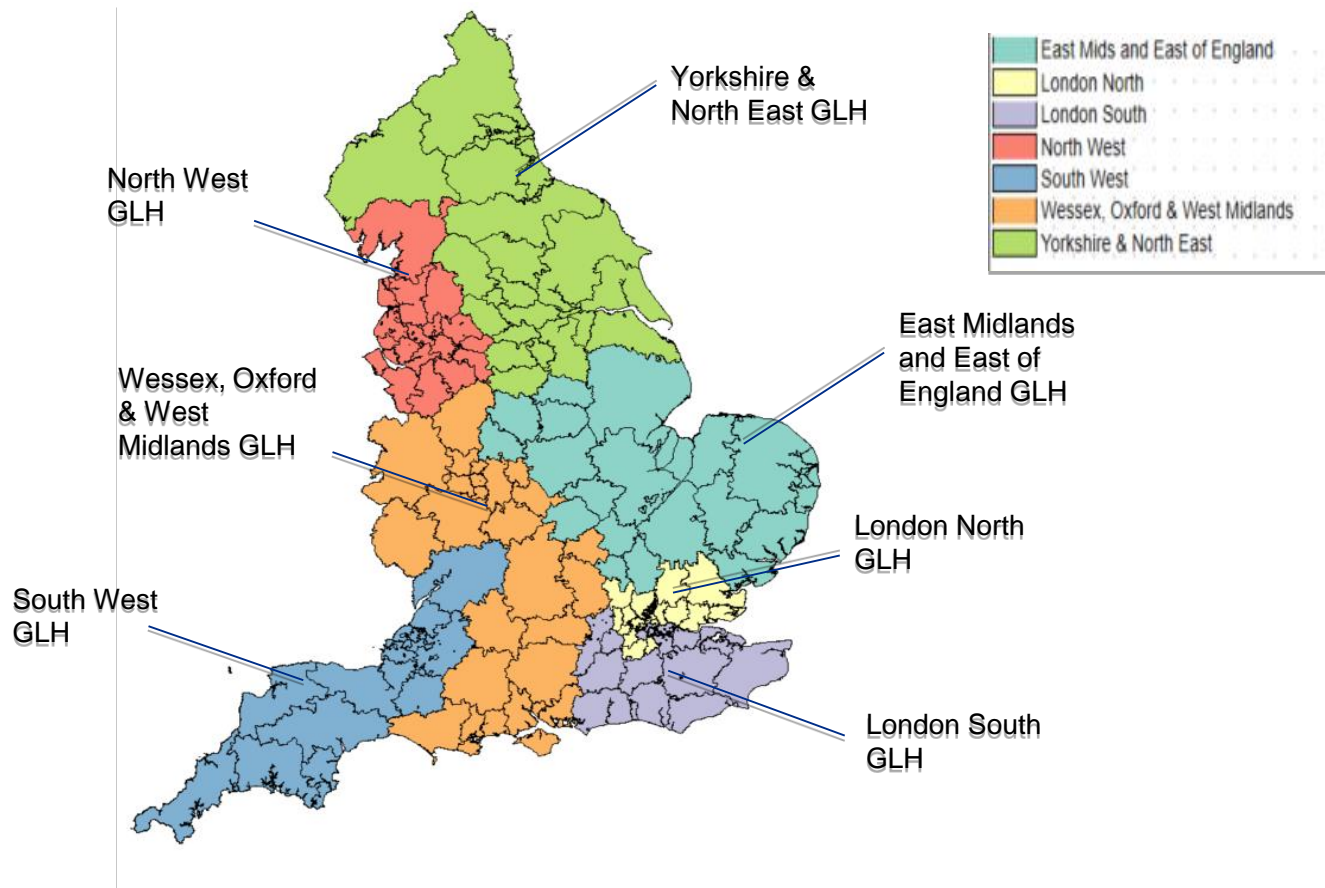
### Medical Programme

- Working with the Academy of Medical Royal Colleges to support the systematic roll out of genomic medicine within clinical pathways and to ensure clinicians have access to the right education and information at the right point

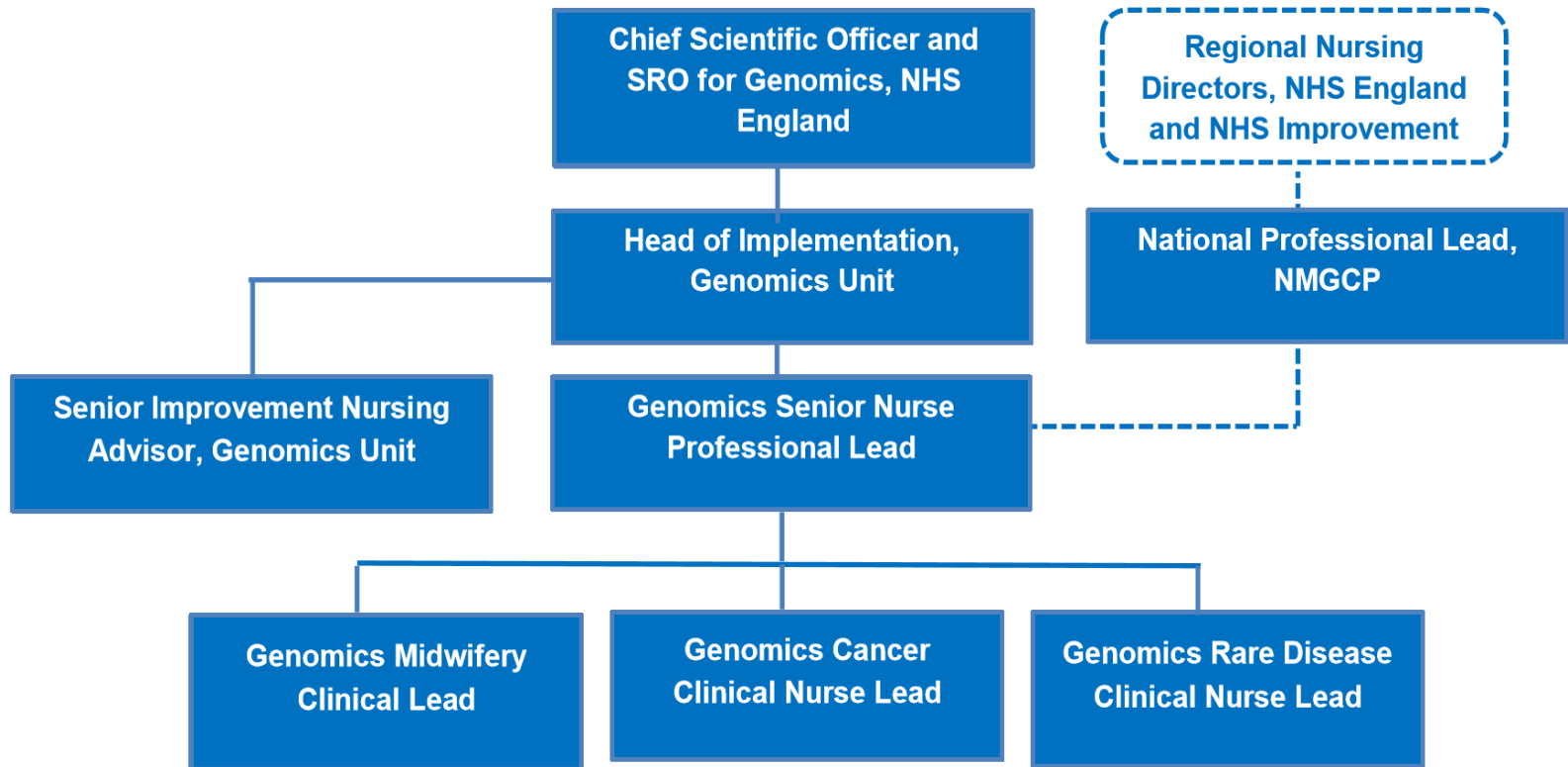
### Nurse & Midwifery led Genomics Collaborative

- Working with 7 Projects across England to systematically and sustainably embed genomics into nursing & midwifery roles and responsibilities

# Nurse & Midwifery Transformation Programme: 7 Collaboratives - 1 across each Genomic Medicine Alliances geography

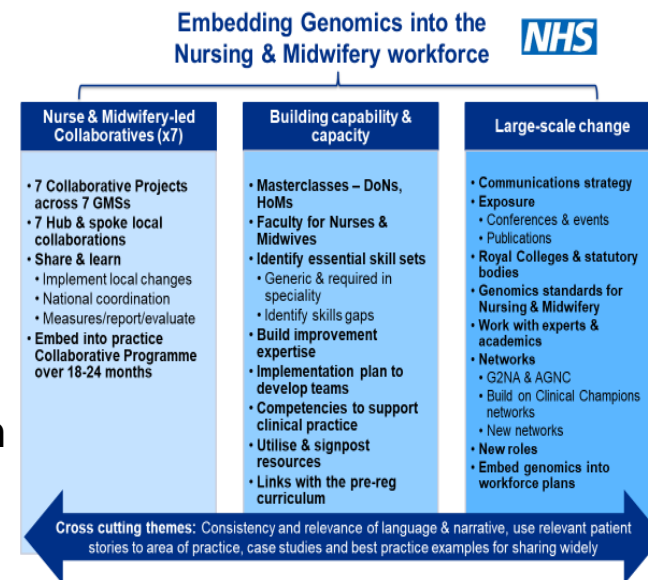


# Genomics Nursing & Midwifery Transformation Programme Team



# Planned Activity 2020-21

- Work with 7 Chief Nurses (and key team members) in 7 GMS Alliances to help build their strategies and implementation plans across their geographies
- Clinical Leads - offer expert advice and support
- Connect up the system
- Meaningful baselines & measures for improvement to assess progress
- Capture learning and develop new & existing sharing mechanisms
- Tap into existing Networks, create new Networks and connect these
- Events (various) across England
- Further Masterclasses and potential for e.g. roadshows
- Large scale change event/s working with Horizons team
- Work with the Medical and Pharmacy programmes to align
- Engage senior nursing & midwifery leaders – present at all relevant national events
- Extensive comms, engagement and media plan to reach Midwives
- Joined up work with the HEE GEP Team



# Dr Anneke Seller - update on the Genomics Education Programme

Dr Anneke Seller presented the work being undertaken by the Health Education England Genomics Education Programme (GEP).

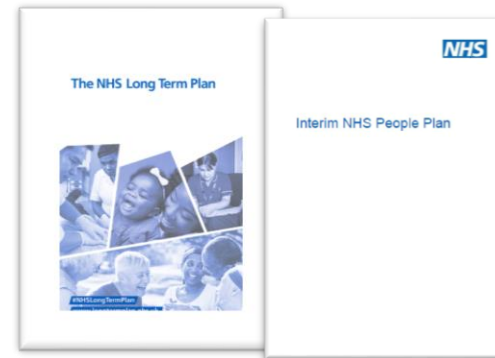
As the service delivery landscape changes, the Genomic Medicine Service evolves and GMS Alliances are created, the GEP will be developing its workforce development strategy aligned to the emerging service model and the work of the three collaboratives.

The GEP will assess workforce development, education and training priorities to fully support implementation. Considering the requirements for the prospective and current workforce. Recognising where and when genomics is relevant and identifying what resources are required.



# The current genomic landscape

- Publication of NHS Long-Term Plan and Interim People Plan
- Transition to the NHSE Genomics Medicine Service
- NHSE plans to develop service and workforce models across:
  - Nursing and midwifery
  - Pharmacy
  - Medical









***What are the underpinning workforce development, education and training priorities to support?***



# GEP in summary

## HEE's Genomics Education Programme is:

-  Providing co-ordinated national direction and leadership in **education and training** to support the development of a 21st century genomics workforce
-  Initiating **workforce planning and intelligence** activity to inform training commissions, workforce numbers to deliver GMS activities.
-  Focusing on **upskilling the existing workforce** through targeted CPD opportunities.
-  **Developing a broad range of innovative resources** in-house and with our partners/collaborators.
-  **Providing expertise** to the development of curricula and competency frameworks.
-  Working with networks to **influence** and aid **spread and adoption**.

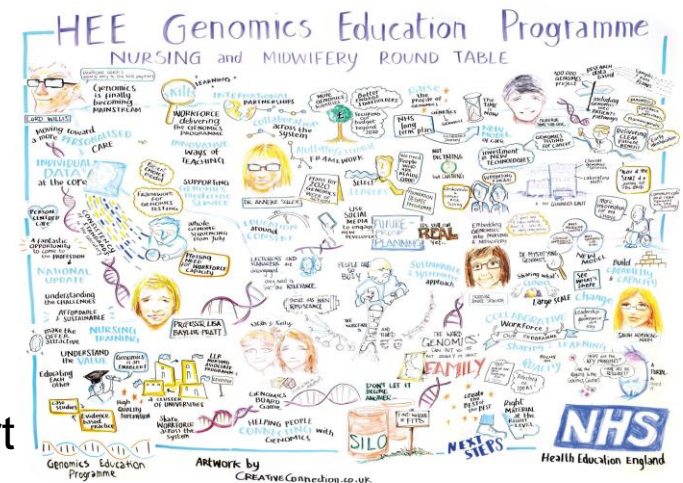
# How the GEP is supporting the nursing and midwifery workforce

- **Strategic:**

- HEE Nursing and Midwifery Roundtable
- NHSE Nursing and Midwifery collaborative

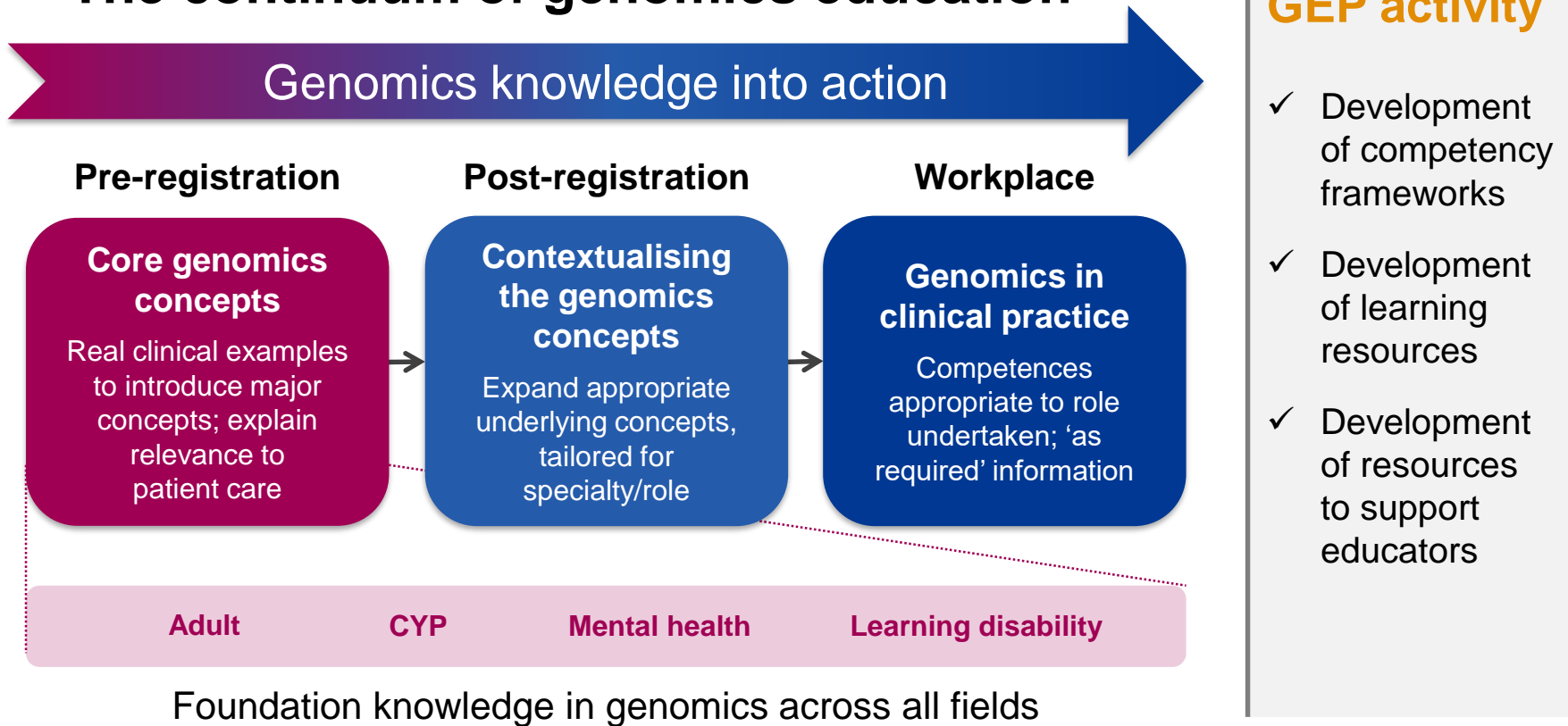
- **Education and Training:**

- Future workforce:
  - Educators toolkit; competencies to support standards of proficiency
- Current workforce:
  - Funding MSc framework in genomic medicine
  - Funding Macmillan Genomic Champions residential course
  - Competency frameworks
  - Resource development

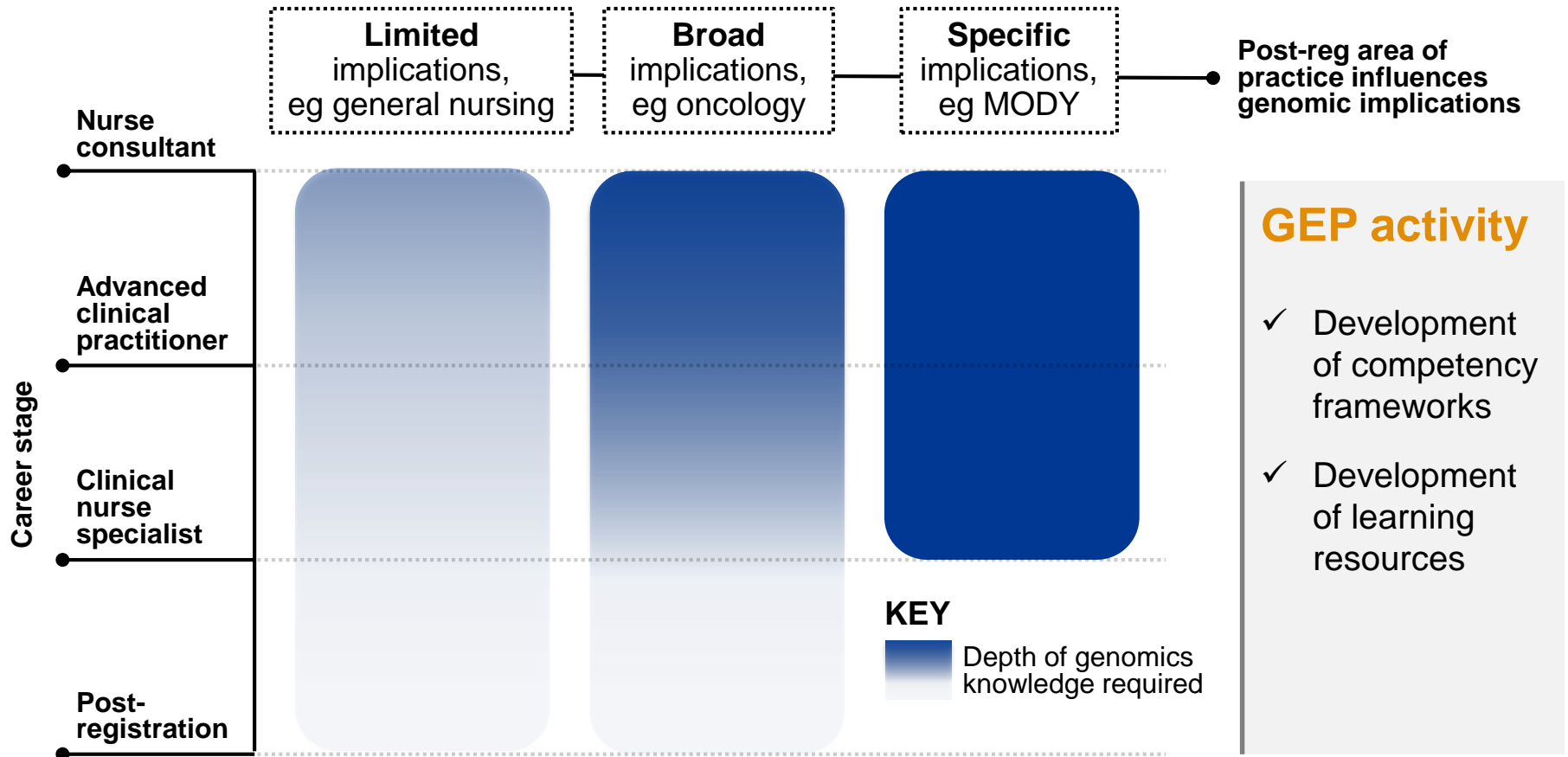


# Recognising where genomics is relevant

## The continuum of genomics education



# Nursing in genomics: knowledge map



# Competency frameworks

- Building on previous work by Kirk et al 2011.
- Similar model to that we have already used to produce competency frameworks for facilitating patient choice and returning patient results
- Working with subject matter experts
- Designing a competency framework covering three aspects:
  - Undergraduate nurses
  - General nursing workforce
  - Specialist nursing workforce – e.g. Advanced Nurse Practitioners
- Thinking about what every nurse needs to know e.g. what genomics is, it's implications for patient care and signposting to specialist services right through to the specialist knowledge and skills required by advanced practitioners



# Educational opportunities



## Taught courses

Funded CPPD modules; PG certs (can self-fund to Master's level)



## Online courses

30+ courses, from introductory to specialist



## Training sessions

Genomics in nursing, variant interpretation



## Practical guidance

Taking a family history



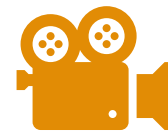
## Just-in-time information

Factsheets, guides



## Educator resources

Competency frameworks, toolkits, board games



## Film and animation

60+ educational videos

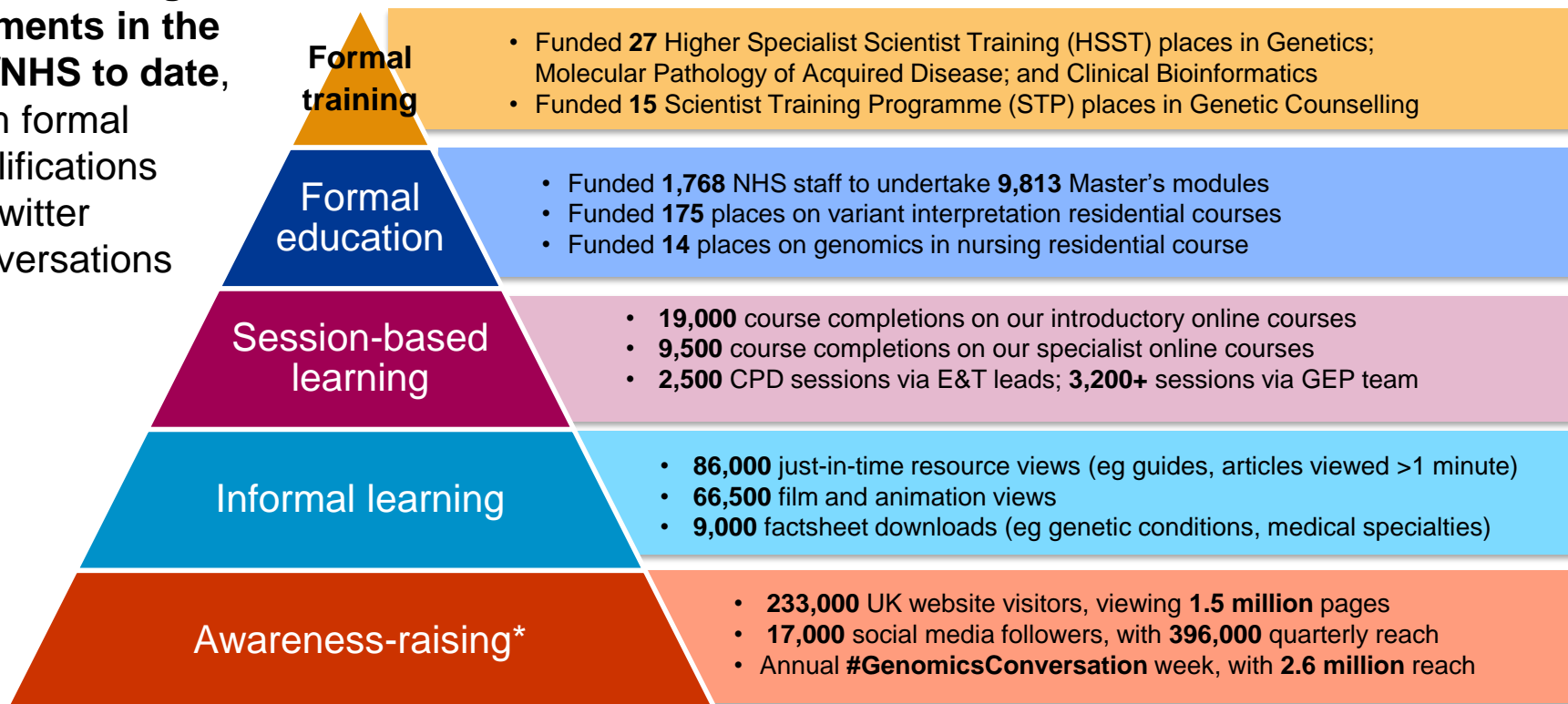


## Awareness raising

Social media campaigns, blogs

# Educational opportunities

Total learning moments in the UK/NHS to date, from formal qualifications to Twitter conversations



\* Social media figures are worldwide; all others are NHS/UK. Figures correct at 31<sup>st</sup> Dec 2019, online data rounded to nearest 500.

# GEP Resources: Week of Action 2020

## Genomics Conversation March 16-20<sup>th</sup> 2020

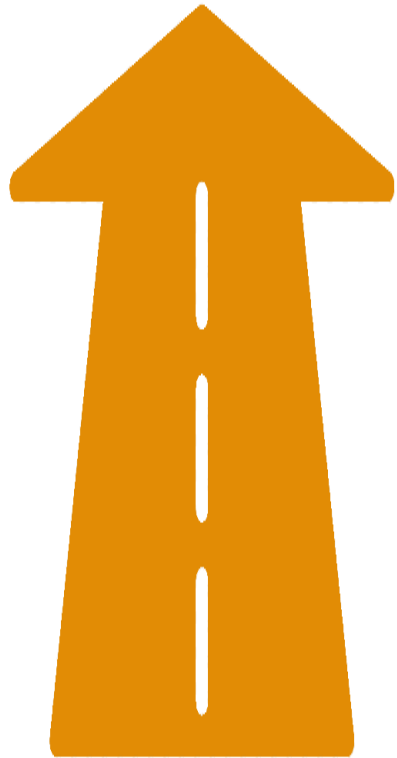
- National week of action to raise awareness of genomics in healthcare.
- Featuring events, podcasts, twitter chats and the launch of resources.
- The focus of this year's event is:



***“to dispel the myths, explain the facts and outline the clinical impact of genomics for nurses and midwives”***



# Going forward.....



- In process of agreeing **workplan for 2020/2021**
- **Aligned to workstreams** emerging from NHS England
- Linking **workforce development** to **service development**
- Developing **stronger links** within HEE, between NHS England, GLHs and GMS Alliances
  - Workforce development group
    - Include Education & Training leads

# Key ongoing activities

- Masters in Genomic Medicine (new funding model)
- AoMRC: Appointment of an Education Lead
  - Link in with GMS Alliances
- Competency frameworks
  - Intraprofessional (e.g. Nursing)
  - Interprofessional: Clinical activities along the patient pathway
- Resource development where need is identified
- Workforce planning and remodelling
  - Collecting metrics about current workforce
  - Consider new ways of working and what training is needed

# Find out more

**Web:** [www.genomicseducation.hee.nhs.uk](http://www.genomicseducation.hee.nhs.uk)

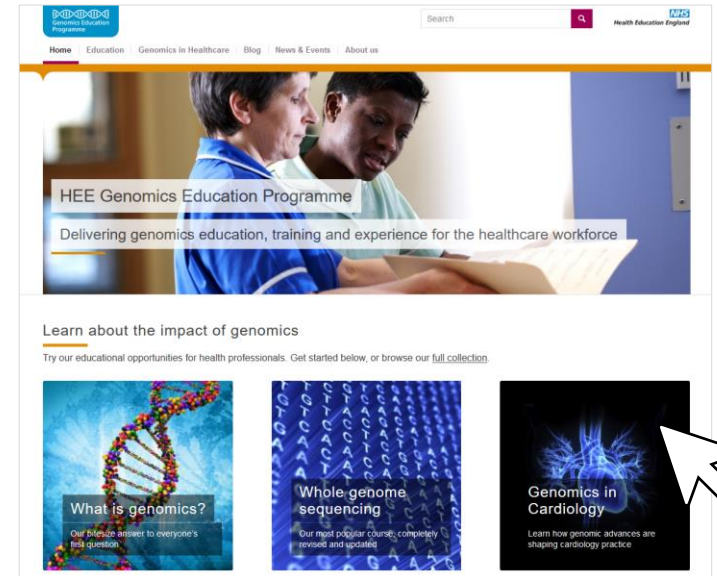
- Articles and guides
- Just-in-time resources, eg factsheets
- Links to all our courses
- c29,000 visitors every month
- Fully revised and relaunched in June 2019

**Social media:** @genomicsedu

- Twitter, Facebook and LinkedIn
- c16,000 followers

**Multimedia:** Vimeo, YouTube, Flickr

- Educational films, animations and infographics
- Free to embed / download for use in your presentations




# Leanne LeRiche - Views from the Waiting Room: reflecting on patient and family experience

Leanne LeRiche is a parent to a daughter who has an undiagnosed genetic condition. Leanne discussed her family's experience and highlighted the importance of the role of the nurse throughout the patient care pathway.

Leanne emphasised that nurses often build longer term relationships with patients and their families; they can spend longer with them and may get asked questions that patients feel they can't ask other healthcare professionals.

Leanne discussed consenting and the impact of genomic testing, as well as the value that nurses and midwives can add to the patient and parent experience.

***Healthcare will become increasingly dependent on technology – but no amount of technology can compensate for an empathetic nurse or midwife***



Views from and beyond the Waiting Room:  
reflecting on patient and family experience of  
genomics to date

Leanne LeRiche

100,000 Genomes Project Participant Panel

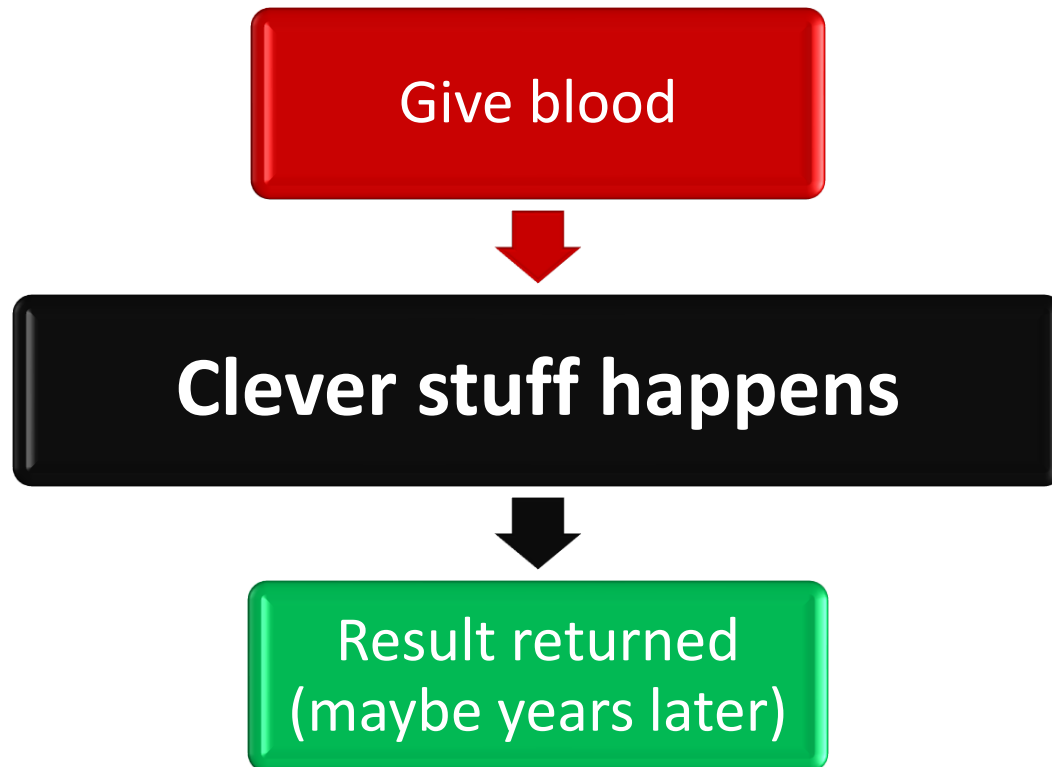
Nursing & Midwifery RT Jan 2020

- Parent to daughter Emily who has undiagnosed genetic condition
- Genomes sequenced in 2012 as part of NHIR pilot study – negative result returned Winter 2017
- Participant Panel member since April 2016
- Actively involved in overseeing research within Genomics England
- Here today to reflect on the patient experience of genomics so far and point out where we think nurses and midwives will be especially important in the future
- Attempt to give my own perspective and gratitude relating to undertakings such as your round table collaborative and how important they are for us patients, and how, for many, unbeknown to us, beyond the waiting room, the unprecedented task of how best to incorporate genomics into our National Health Service, the NHS, and its workforce has and continues to take place.

# Life is for living ...



# Patient experience so far



- Exciting new technology with the potential to bring an end to very long diagnostic odysseys
- But giving access to our whole genomes is a leap into the unknown
- So gaps in knowledge have to be filled by TRUST



# The growth of genomics

- Up to now, genomics has been a specialist subject: we mainly see consultants and counsellors
- But expansion of genomics into mainstream health care brings many more patients and families into contact with genomics
  - Rare conditions
  - Cancer
  - Pharmacogenomics
  - Screening programmes
- There is a huge need for staff with nursing skills and natural empathy to help families feel comfortable when deciding whether or not to give their consent for testing, and to cope with whatever results come back

# Before consenting to genomic tests

- Patients come from different perspectives
- In most cases a significant health problem is already evident; testing is proposed in order to understand and treat it better
- Patients and their families typically want to know at this point:
  - What will the NHS do with this ‘encyclopaedia of me’?
  - Who else will see this data and what will they do with it?
  - Where is the benefit to me or people like me?
  - What results will the NHS tell me, how and when?
  - What will we **not** be able to find out?
- In some cases there may be concerns before birth; antenatal testing may be appropriate and may have even more significant consequences

# Impacts of genomic testing

- Positive result: diagnosis => treatment pathway
- Negative result: rules out many scenarios but no diagnosis
- Uncertain result: what now??
- Impacts can be wide ranging, depending on each patient's circumstances:
  - Reproductive decisions
  - Family dynamics
  - Future health concerns
  - Socio economic impacts
- Effective and compassionate communication at this stage is essential

- In the foreseeable future, pharmacogenomics will tell us more about how our bodies react to different drug ingredients
- Some of these findings will have significant impacts on our treatment pathways
- Nurses and midwives will need to recognise these findings and continually act upon them

All for one and one for all .... Together we  
can



# Conclusions

- Nurses will play a key role in bridging the gap between genomic discoveries and their translation into clinical care:
  - More time to relate to patients
  - Ability to build their trust
- Healthcare will become increasingly dependent on technology – but no amount of technology can compensate for an empathetic nurse or midwife

# Beyond the waiting room - What it takes....



## Every organisation providing healthcare questioning how can they raise the bar to continue to provide world-class healthcare....

- **Workforce awareness** - *knowledge = comfort not self-doubt. How much? No one-size fits all*
- **Perspective** - *clinical, educational & research practice*
- **Drive change** - *local, national & global – embrace & unify genomics across the board*
- **Establish consensus** - *identifying opinions & taking into account work already undertaken*
- **Utilisation of existing skills** - *learning from best practice such as antenatal screening, genetic counselling, ethics & consent, specific roles & fields where genomics is already present*
- **Upskilling** - *existing staff making the most of genomic technologies*
- **Expectation management** - *patient & wider public, healthcare workforce*

data security

‘cures’

ethics

ensuring consent

consistency

common & transparent language



- **Patient public engagement** - awareness of genomics, what it means to the patient - personalised health, children & young people – schools, colleges & 6<sup>th</sup> forms
- **Emphasis** - treating the whole person, holistic, non pigeon holing as is currently the case
- **Evaluation** - impact & learning from the 100k Genomes Project – see where it is/has made a difference using case studies, profession specific information & guidance, social media etc
- **Focus** – patient at the very core in all that is done, enhancing society for all - for me - with me
- **Commitment** – everyone – healthcare workers, government bodies, industry, patients
- **Engagement** – engaged staff will think and act in a positive way about the work they do, the people they work with and the organisation that they work in
- **Innovation** – empower healthcare workforce to put forward ways to deliver better and safer services for patients and their families, support technological advancement
- **Standards** – introduced and embedded into practice – crucial for consistency
- **Collaboration** – every opportunity, every level, incorporating & involving - embrace mutual benefit

Genomics is happening, it is here and I thank you, individually and collectively.

Each and every one of you here today are paving the way, transforming unmet need as standard and routine.

Leanne LeRiche

Member, 100,000 Genomes Project Participant Panel

# Suggestions

- **Genomic App**
  - Quick reference, guidance, key points, refresher reading etc for healthcare workforce
  - Health games – schools, nurseries, well-being centres , general public, patients
- **Health Day**
  - Schools, clubs, colleges, retail, day centres, care homes, food bank collections within schools on the day
  - HEE, NHS, Charities, DoE, local communities
  - Sponsored health events – charity fundraising alongside
  - School sports day part of health day initiative? Parents drop in events throughout the day
  - Reach diverse communities/minorities
  - Incorporate current aspects of health drive including obesity, mental health, well-being whilst introducing genomic health
- **Careers Advice**
  - Any coverage linking to health & social care or science curricula?
  - National Careers Service, educational in house career advisers, college careers advisors , National Careers Service. Gov
- **Spotlight on**
  - Videos introducing the work of different various roles such as biomathematician & researcher
    - Spark curiosity
    - Increase interest

Leanne LeRiche  
Member, 100,000 Genomes Project Participant Panel

# Dany Bell - Macmillan Genomics Portfolio of Activity

Dany Bell, Strategic Advisor Treatment, New Medicines and Genomics presented the portfolio of work that Macmillan Cancer support have undertaken in relation to genomics to date.

Macmillan have been working with NHSE/I and HEE to drive forward a genomics work plan. The plan will ensure their workforce is skilled and knowledgeable and that those with cancer can have conversations with staff who have the information and support they need to make informed decisions, especially regarding genomic testing and obtaining results.

It was noted that organisations should work collaboratively to avoid replication, for example, signposting from websites rather than duplicating. The Genomic Strategy Group, which is hosted and chaired by Macmillan, meet each month with membership from across 14 organisations.

14 Genomics Champions are to be funded and recruited by Macmillan to help educate and support teams within the Genomic Laboratory Hubs (GLHs). Health Education England have also funded the individuals to undertake a Gateway to Genomics course, developed by the University of the West of England (UWE) as part of their training.

**MACMILLAN.**  
**CANCER SUPPORT**

# Macmillan Genomics Activity

**Dany Bell**

Strategic Advisor Treatment, New medicines and Genomics  
Jan 2020

**MACMILLAN**  
**CANCER SUPPORT**

# Macmillan Cancer Support's purpose

**TO HELP  
EVERYONE  
WITH CANCER  
LIVE LIFE  
AS FULLY  
AS THEY CAN**

# Considerations

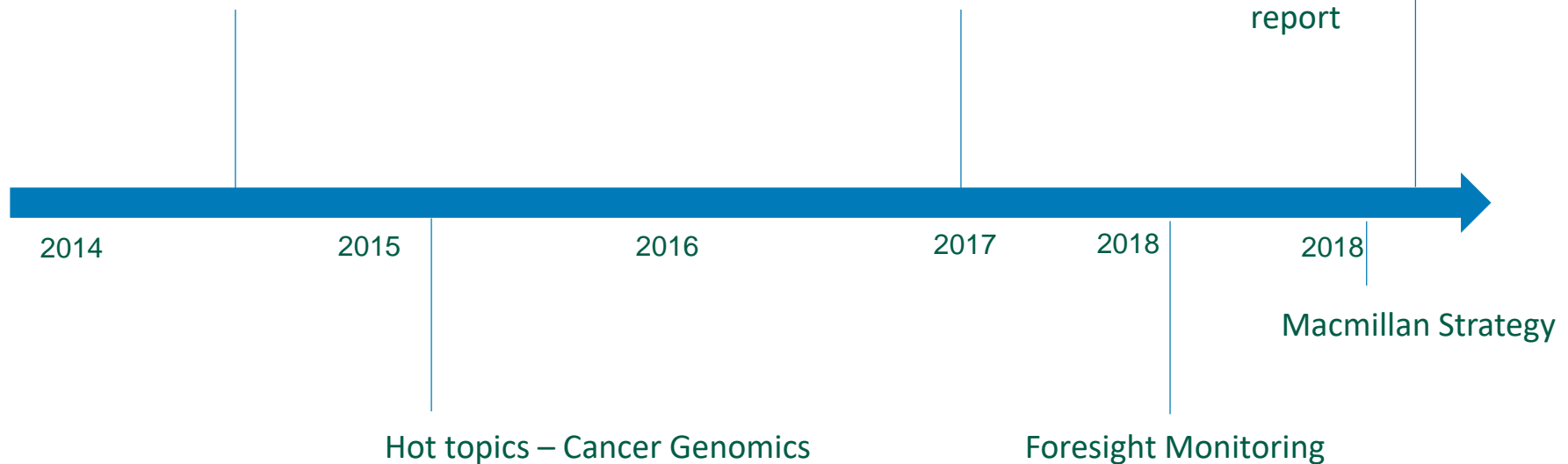
- How do we **ensure equity** and understanding for harder to reach groups around genomics?
- How do we ensure people **understand** their options and make good choices?
- How do we ensure the **workforce** is equipped to support patients when having conversations?
- What do internal teams need?
- Strategic implications

# Macmillan evidence and insight to date for Genomics and Personalised Medicine

Treatment and Recovery *Understanding future trends in technology, targeting, specialisation and location*  
*How personalised will genomic therapies be by 2030?*

Foresight on the Cancer Experience

Personalised treatments insight report







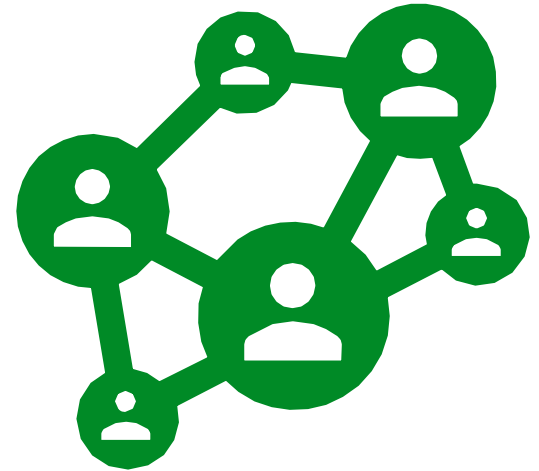
## Asking PLWC to be involved

# Collaboration with NHSE/HEE

## Ambition

Collectively drive Genomics agenda to ensure:

- People with cancer have the information and support they need to make informed decisions
- Workforce is skilled and knowledgeable and can provide Person Centred Care and Support around conversations for genomic testing and results giving
- Provision of consistent messages, reduce duplication and use our networks productively and collectively

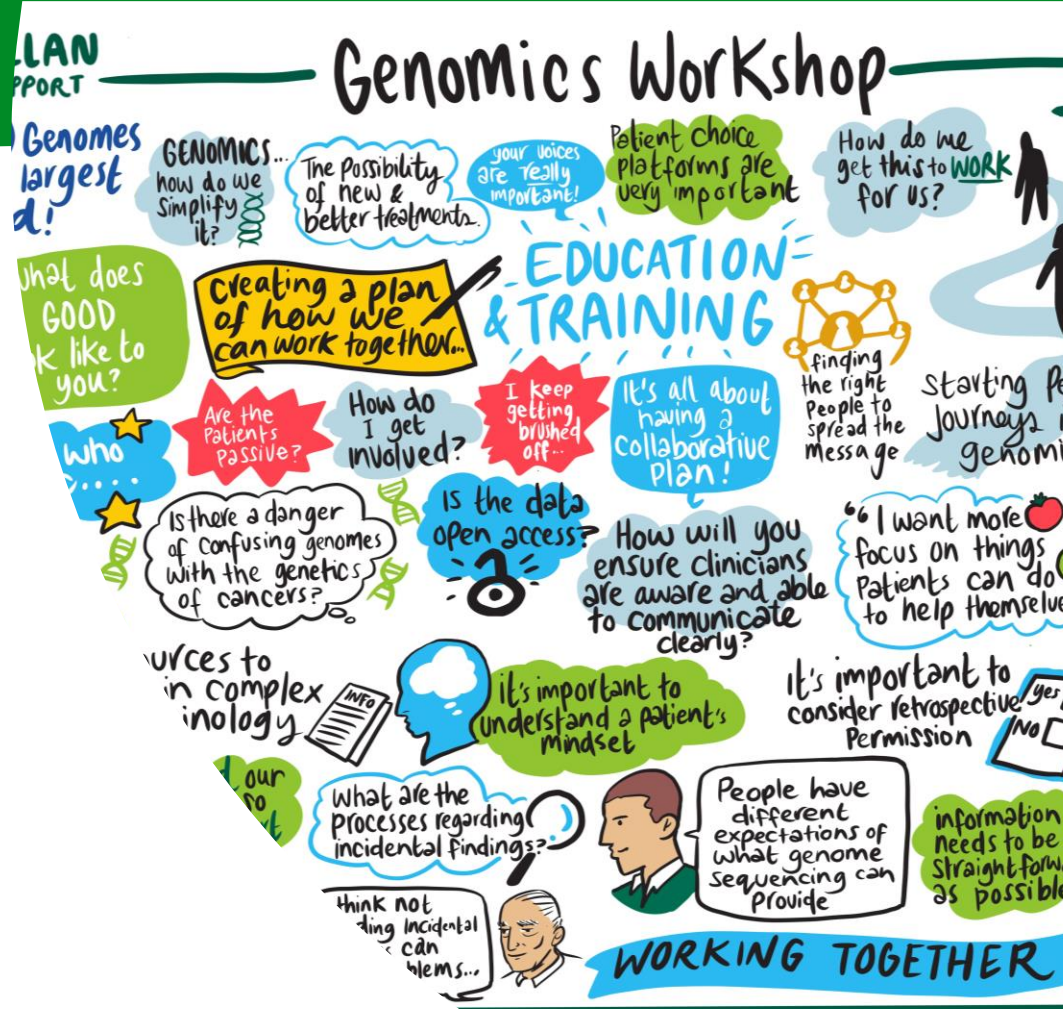


# Leadership, governance and involvement

- A Genomic Strategy Group meets monthly with membership across 14 organisations, is hosted and chaired by Macmillan and co chaired by NHSE/I
- There is a structured workplan with timescales
- All partners feed into the agenda for meetings giving the opportunity to think and discuss wider than the workplan
- All partners have a seat around the table and can suggest additional membership
- PPI group was formed from across all partnership specialty areas, so we have a valuable mix of people

# Achievements

- Formation of a Strategic Genomic Medicine Group across 14 organisations
- Formation of patient and public pan-cancer genomic user forum
- Mapped resources across all organisations
- Informing content and language of NHSE patient information leaflet
- Sharing networks and expertise e.g. taking the HEE RCGP genomics toolkit out to our networks of GP's and education forums
- Enabling smaller charities with less infrastructure and funds to participate actively in this agenda via this group
- Survey of GP's understanding of BRCA to help target engagement and education of GP's
- Fed into patient choice model and competency framework
- Workshops at Macmillan Professionals Conference





## Genomic Education Champion posts

- Identifying who requires training within their patch
- Educating individuals/teams using a blended learning approach
- First point of contact for clinical staff
- Undertake the patient choice conversation training via the competency framework
- Keep records of who has been trained and what and report back
- Linked to the Genomic Education and training leads for the GLH's
- Feed into the development of clinical pathways for patient choice conversations

# Professional Development and Knowledge Team

Head of Professional Development & Knowledge

Development  
Manager -  
PLWC

Development  
Manager -  
Professional  
Workforce.

Content  
knowledge  
Manager

Delivery  
Manager  
North

Delivery  
Manager  
South

Grants Manager

Development  
Lead - PLWC.  
x2

Development  
Lead  
Professional  
Workforce. x5

Content  
Knowledge Lead  
x2

Delivery Lead  
x7

Delivery Lead  
x8

# Membership for Collaborative Partnership



# True collaboration



Success and breadth of activity has been achieved due to the engagement of all members of the group and pooling resources to support NHSE/I/HEE :

- Access to networks of professionals the NHS wouldn't usually have access to
- Access by NHS to charity forums and to be able to deliver workshops, engage with professionals and understand their needs and concerns around
- NHS benefiting from influencing by charity policy teams across all members on Genomics.
- Dedicating a Genomics user group and our patient networks via our websites for NHS Genomics to access voice of people with cancer who are connected to local user groups and actively drive this agenda across different cancers in their local areas spanning all GLH's
- By working together influenced charities to develop their focus on this agenda within their remits spreading the workload across NHS and third sector



# Future ambition

- Development of targeted innovative training and education resources
- Embed and support cascade training posts and evaluate
- Actively engage and collaborate with royal colleges as a group
- Target different groups of professionals with awareness sessions
- Extend engagement and awareness raising beyond England to other nations
- Continue activity on workplan and add to it based on need, further engagement and insight

# Tomorrow's World



## Developing a Macmillan Genomics and Personalised Medicine Expert Reference Group

- To explore how best to inform and advise Macmillan with regards to future direction of genomics and personalised medicine
- To develop a shared understanding of Macmillan's interest and role in this space to date
- To inform a strategic horizon scanning exercise

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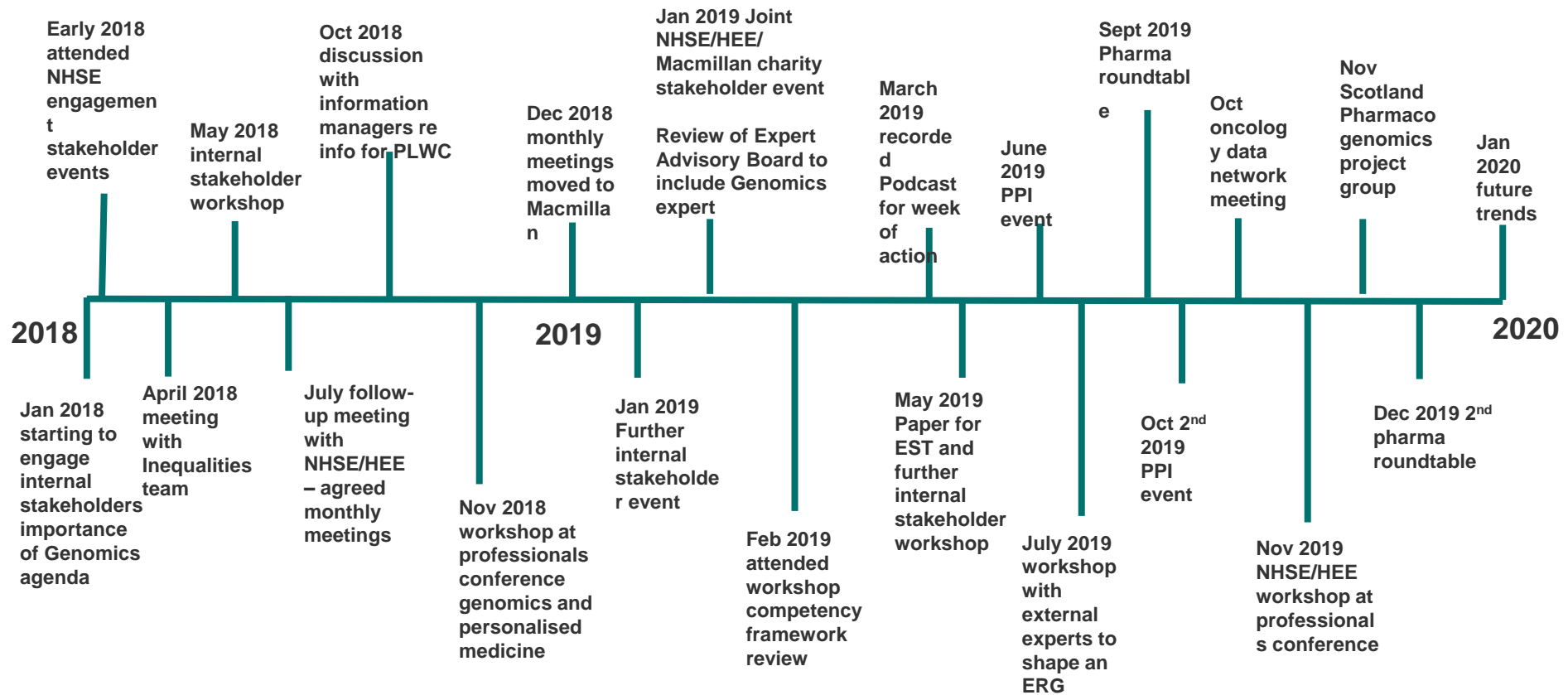
# Pharma roundtable - Understand the opportunities for horizon scanning to inform future support offers and start a dialogue and explore opportunities to collaborate

## Common priorities throughout the discussion

- Patient engagement– diversity, decision making, design of patient experience, and engagement as early in the design of trials and research and development as possible
- Genomics upskilling
- Education for healthcare professionals around new treatments – immuno-oncology and tumour agnostic drugs
- Influencing hard and soft conversations – holistic support for patients, and moving beyond the clinical
- How to improve patient access to information



# Activity timeline



# Vicky Cuthill - Mainstreaming in practice – The Polyposis Registry example

Vicky provided an overview of what polyposis is, the history of the condition and the integration of genomics into clinical practice for conditions that require lifelong clinical care.

The Polyposis Registry is unique as it is a unit that can deliver genetic diagnosis and lifelong care.

Genomics has shaped care in polyposis; as an example genotyping can influence surgical decisions.

For those with rare diseases a register can combined care for the patient and the ability to collect data and research with larger numbers of patients. The Polyposis Registry covers a range of activities including working with families with family tracking, cascading tests and genetic counselling.

The The Polyposis Registry staff are all NHS funded and this is an NHS service the database is funded by charity donations





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# Mainstreaming – The Polyposis Registry Example

**Vicky Cuthill**  
**Lead Nurse/Manager**  
**The Polyposis Registry and Family**  
**Cancer Clinic**  
**St Mark's Hospital**  
**27<sup>th</sup> January 2020**





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## Aims

- What is polyposis?
- A brief history of polyposis
- Mainstreaming in the context of conditions that need lifelong clinical care
- Why have a registry? An outline of what we do





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## What is polyposis?

“Polyposis is an umbrella term for a range of inherited conditions which cause polyps in the gut and can increase the risk of cancer”

Conditions classified according to type of polyp:

- Adenomatous polyps – FAP, MAP, PPAP
- Hamartomatous polyps – JPS, PJS, PTEN
- Serrated polyps - SPS



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## A little bit about the history of polyposis.....

- First description of multiple polyps dates back to 1721
- Prior to the 1860s, autopsy findings informed the literature – observational and linked to intestinal inflammation
- Sklifasowski published the first histologically verified case of adenomatous polyposis in Russia in 1881
- In 1882, inherited predisposition was highlighted when Cripp identified two affected siblings with a condition he termed “disseminated polyposis of the rectum”
- In 1907, the first review of FAP was published by Doering who found 44 references to FAP in the literature and 31/37 patients having died from cancer





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## Polyposis Registry Timeline

1924	Started as a research project, becoming first Polyposis Registry in the world
1940s	Inheritance pattern established
1948	First colectomy with IRA for FAP
1962	Morson publishes 'precancerous lesions of the colon and rectum'
1975	HJR Bussey thesis published 'Familial Polyposis Coli'
1978	First 'Parks Pouch' for FAP performed

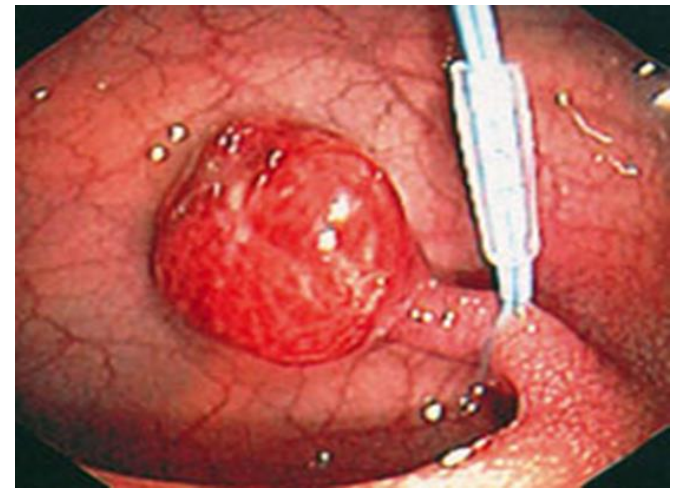
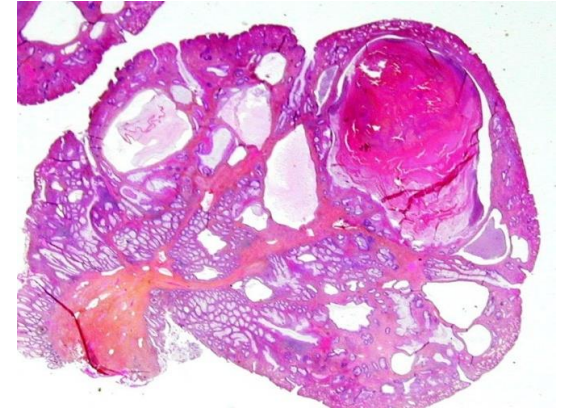




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<b>Polyposis Registry Timeline</b>	
1985	The Registry becomes a formal hospital department
1987	Bodmer et al. identifies region on Chr 5 of <i>APC</i> gene
1989	Spigelman et al. publish in The Lancet, initiating upper GI screening programme
1996	Clinically responsible for genetic counselling/NHS genetic testing commences
2000	First nurse practitioner/SPS first classified by WHO by Burt and Jass
2003	<i>MUTYH</i> gene identified – recessive inheritance pattern
2012	PPAP caused by germline mutations in exonuclease domains of <i>POLE</i> and <i>POLD1</i> cause PPAP
2014	World's first paediatric polyposis nurse practitioner



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## Mainstreaming and Polyposis

- Mainstreaming – a byword for the integration of genomics into standard clinical practice
- Difficult to achieve within the current confines of the NHS
- Usually focused on diagnosis
- The polyposis registry is a mature example of a unit that can deliver genetic diagnosis AND lifelong clinical care



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# How genomics has shaped clinical care in polyposis

- Surveillance differs according to genotype
- Genotype can influence surgical decision making in FAP
- Desmoid risk and genotype
- MAP and recessive inheritance
- SMAD4-HHT overlap for JPS



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## Why have a Registry?

- For any rare syndrome it's important that patients are cared for in specialist centres
- Especially important in premalignant conditions where cancer prevention hinges on early identification and appropriate treatment of patients
- Registries combine patient care and the ability to collect data and carry out research with larger numbers of patients
- Overall effect is cancer prevention through improved patient care with sound research base





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## What does the Polyposis Registry do?

- Acts as a hub for patients' clinical care
- Manages families, not individuals
- Family tracking, cascade testing and genetic counselling
- Patient education and open days
- Psychological support
- Secretariat for InSiGHT
- Teaching in-house, nationally and internationally





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## Patients attending Registry May 2018

FAP	896	48%
JPS	96	5%
PJS	142	8%
MAP	131	7%
SPS	253	13%
Other*	334	18%
Total	1,880	100%

\*PPAP + PTHTS + Unclassified Polyposis + screening pts



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## What happens when a patient is referred to the registry?

- Referral is triaged – consultant/nurse practitioner/telephone
- Checks to ensure pt does not belong to existing family
- Patient registered and added to database
- Appointment sent out with family history questionnaire
- New consultant appointment and family history appointment
- Family history taking, genetic counselling and testing as required
- Patient given contact details and information booklet
- Investigations organised/coordination of appointments
- Assessment of family and identification of at risk relatives
- Call up of at risk relatives/use of NHS database to track and trace



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## Patient Care and Research

- Custom made database
- FileMaker Pro 17
- Hosted by the Trust on a virtual server
  - Surgery, endoscopy, pathology, radiology
  - Phenotype and genotype
  - Appointment dates – due and booked
  - Extended family



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# Cancer Prevention by Cascade Testing

Code Entry	Patient Entry	Patient	External	Thames	Genetics	Gene Stats	Families	Operations	Cancer	ECM	Im
------------	---------------	---------	----------	--------	----------	------------	----------	------------	--------	-----	----

**General Reference**

Patient ID: 997.IV.3      Age:

Date of Birth:

Family Code: 997      [Details](#)

Family Name: Marks

Patient Name: James Count

Current Cat:       [Modify](#)

Current State: P

**Other Registry Information**

Family Code: 997

Family Name: Marks

Alt Patient ID:

[Enter >](#)  
[in both](#)

**Family Mutation Details**

Status: **Mutation identified**

Gene Name: APC

Exon: 15      Intron:       [MI](#)

Codon No.: 1309

Codon Prev:

Known As:

Sequence: 3926-3930 del GAAAG

Amino Acid Ch:

Polyposis Type:       [U/T Count](#)

Other type (text):

**Consent Forms**

	Comments
NHS Testing <input checked="" type="checkbox"/> Y	FAP
Genetic Research <input type="checkbox"/> Y	
Genetic Info <input checked="" type="checkbox"/> Y	1/1/2014
Medical Info <input type="checkbox"/> Y	
Tissue <input type="checkbox"/> Y	

**DNA Test Chronology**      **GENE STATS**

GT      DNA ID: 997GT      Code: GT

Index Case?  Y      DNA Test Urgent?  Y

First Sample	Second Sample
DNA Sample Type 1: Blood      Age: <input type="text"/>	Age: <input type="text"/>
Sample Taken Date: 01 Jan 2014	
Blood tested at KGC: <input checked="" type="checkbox"/> Y <input type="checkbox"/> N      Delays: <input type="text"/>	Delays: <input type="text"/>
Result received For (gene type): 03 Feb 2014      33	
Test Mutation Status: positive	

Full Gene Sequencing Done?  Yes  No       Yes  No

Result to Referrer on:      

Result to Patient on:       261

**HAS PATIENT BEEN INFORMED?**

Fup call due:      

Fup call done:      

Blood Bank Sample Taken:  BB      BB Sample Date:

22/10/14; 15:18; KFN; DNA Test 1 Outcome: positive  
 22/10/14; 15:18; KFN; DNA Test 1 Read: 03/02/2014  
 22/10/14; 15:17; KFN; Blood sample 1 tested at KGC; Y  
 22/10/14; 15:17; KFN; DNA Test Date 1; 01/01/2014  
 22/10/14; 15:17; KFN; DNA Sample Type 1; Blood



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# Cancer prevention by monitoring attendance

- Using the database we can monitor:
  - Relatives advised to be referred
  - Attendance of patients referred to St Mark's
    - Outpatient, endoscopy, radiology, admission
  - Babies and children who will require referral in future years



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# Cancer prevention by monitoring attendance

- Successful monitoring relies on
  - Staff understanding how to use the fields
  - Timely data entry of attendance
  - Regular preparation and cross referencing of database lists against hospital systems
  - Administrator's knowledge of the syndromes



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FileMaker Pro - [Patient (PP-SERVER)]

File Edit View Insert Format Records Scripts Window Help

NEW ENTRY HIDE SEARCH GET RECENT TOOLBARS ACTION FIND 1 of 49 Ext Surg Name Work KFN

Code Entry Patient Entry Patient External Thames Genetics Families Clinical History Cancer ECM Investigations Treatment Clinical Trials Menu

General Reference Patient History Other Details

Patient ID: 997.1.1 Yb: 1856  
 DB Unique Ref: P-000655 Yd:  
 Family Code: 997 Age: 0  
 Medical Card No. PolyNo  
 NP Hospital No. In UK?  
 Ext Hospital No.  
 Family Name: Marks

Date of Birth: 01 Jul 1856  
 Date First Seen  
 First Bowel Exam  
 Polyps First Seen  
 Last negative  
 Last OPD Visit  
 Date Last LGI Endo  
 Callup?  Y  N  
 LKTBA notified  
 LKTBA calo: 01 Jul 1856  
 Details Dead?  Dead  D  
 Date of Death  
 Polyposis Related?  
 Attending SMH?  Y  Lifestyle

Tel Home  
 Work  
 Mobile  
 Email  
 Other Contact  
 Contact Type  
 Contact Phone

No. ops  
 Operations

Record Created: 10 Nov 1997  
 Modification Date: 21 Dec 2007  
 Modification Time: 13:16:36  
 Modified By

Mark  Y Find Unmark all Exclude Find

Clinics

Next Clinic / Time  
 Next visit Year  
 Next visit Date  
 Clinic Surgeon Int  
 Send appt to patient?  Y  X  
 Print Appointment Cards

Non Attendance Letters

Letter 1 Set  
 Letter 2 Set  
 Letter 3 Set  
 Clear All

Status Information

Current Category  
 Current State B Modify  
 Referral Type 1

Genetic Information

Mutation Status  
 Family Name: Marks Details  
 Polyposis Type FU  
 Other type (text)  
 Genetic Type Genetic Region

My Action List Entry Delay F/up Date Appt Req Date Req For Date Histology? Histology Result Print Call Notes This Patient Selected Patients P Clear All

Call By  
 Call Date  
 Call Time  
 Entered  
 Call Notes

Consultation Staff Inits Action By Action Done?  Y

100 Browse





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# Cancer Prevention by monitoring surveillance

Identity of Patient		Date Last LGI Endo		Investigations				
Surname(s)	Lord	Callup?	<input type="checkbox"/> Y <input type="checkbox"/> N	OGD	<input type="checkbox"/> Y	Date Due	Date Booked	Notes
First Name	Cuthbert	LKTBA notified		Capsule	<input type="checkbox"/> Y			
Initials	C	LKTBA calc		Flexi/Col	<input type="checkbox"/> Y			
Sex	M	Details	Dead? <input type="checkbox"/> D	TCI	<input type="checkbox"/> Y			
Salutation		Date of Death		MRI	<input type="checkbox"/> Y			
Notes		Polyposis Related?		CT	<input type="checkbox"/> Y			
		Attending SMH?	<input type="checkbox"/> Y <b>Lifestyle</b>	USS	<input type="checkbox"/> Y			
				XRay	<input type="checkbox"/> Y			
<b>My Action List</b>   <b>Entry</b>   Delay F/up Date   Appt Req Date   Req For Date   Histology Result   Print Call Notes   <b>This Patient</b>   Selected Patients <input type="checkbox"/> P   Clear All								
Call By	KFN	9/10/11	<input type="checkbox"/> Y	Height cm	Weight kg	BMI Calc		
Call Date	2/10/11	This field is used for recording all contacts: clinic visits, telephone calls etc. The time and "entered" date will be automatically recorded but the "call date" can be amended to record the date the event occurred. For example the record of a clinic consultation may be entered the next day.						
Call Time	13:54							
Entered	03/10/11							
<input type="checkbox"/> Consultation	Staff Inits	KFN	Action By	KFN	Action	Done?	<input type="checkbox"/> Y	
Call By				<input type="checkbox"/> Y	Height cm	Weight kg	BMI Calc	
Call Date								
Call Time								



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# Cancer Prevention by monitoring surveillance

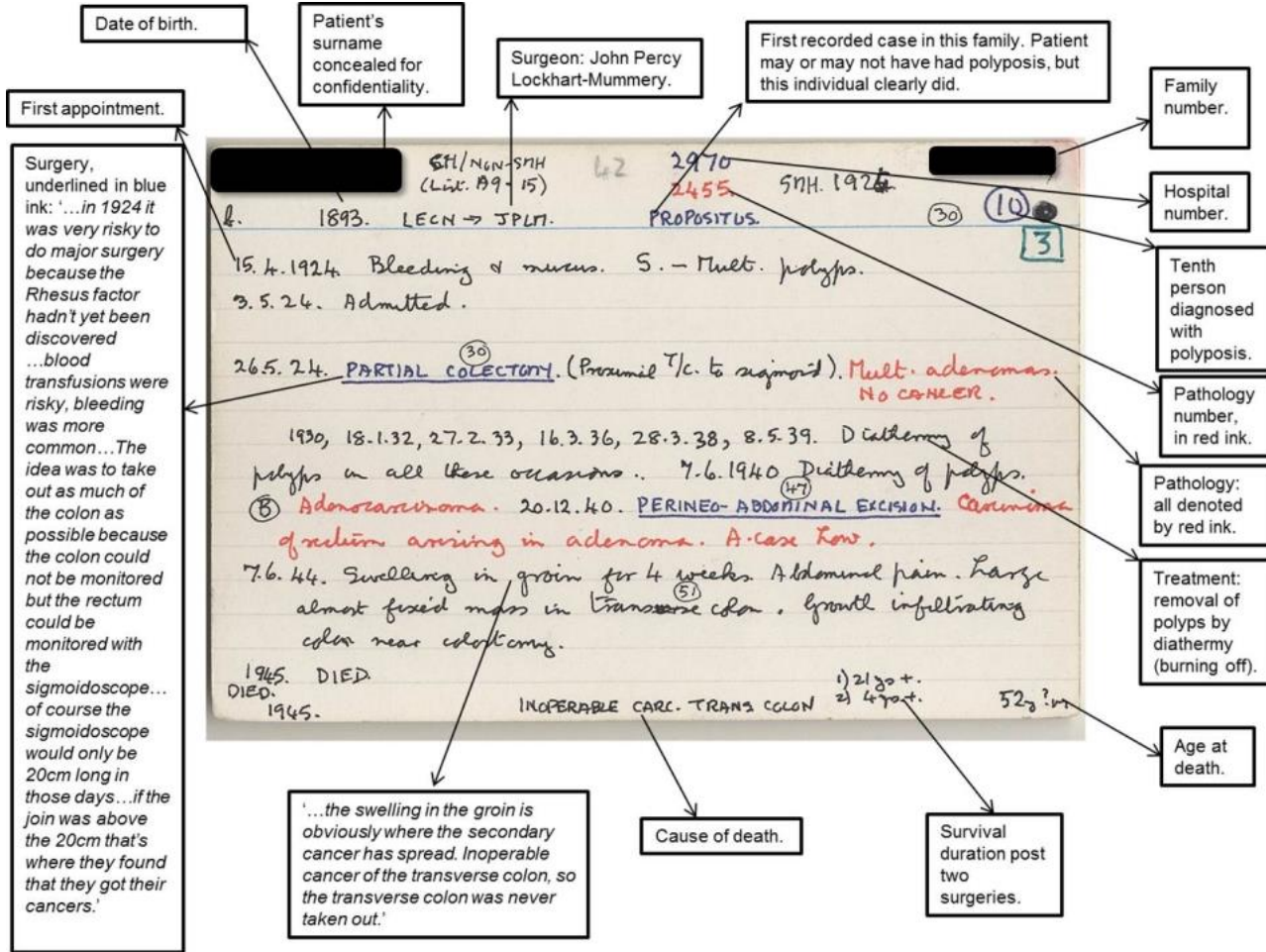
Lower GI Endoscopy Data		Upper GI Endoscopy Data		Scan Results		<input checked="" type="checkbox"/> SPIG IV		First Spig IV (Calo) Date: <input type="text"/>		Last Spig IV (Calo) Date: 21/11/12	
Gastric & Small Bowel Endoscopy Data						Duodenal Histology			Non-Duodenal Histology		
<input type="checkbox"/> DBE	Endoscopy Date	05/02/2014	Video polyps		UGI Histology	Tubular	<input checked="" type="checkbox"/>	Histology			02/04/14; 1
<input type="checkbox"/> Caps	Biopsies Taken?	Y No. 5	Hpylori		Dysplasia	Mild	<input checked="" type="checkbox"/>	Dysplasia			02/04/14; 1
<input checked="" type="checkbox"/> GA	Refused Trial	<input type="checkbox"/> Refused	pH		No. of polyps	5-20	<input checked="" type="checkbox"/>	Polyps No.		Size	02/04/14; 1
<input type="checkbox"/> MRI	Spigelman Grade	II Est	Endoscopist	AL	Size of polyps	5-10 mm	<input checked="" type="checkbox"/>	Location	Fundus		02/04/14; 1
	Ampulla Size mm		Description								02/04/14; 1
Notes: OGD: Multiple fundic gland polyps in fundus. Antrum normal. Ampulla hidden under a fold but did not appear											
<input type="checkbox"/> DBE	Endoscopy Date	21/11/2012	Video polyps		UGI Histology	Tubular-villous	<input checked="" type="checkbox"/>	Histology			08/01/13; 0
<input type="checkbox"/> Caps	Biopsies Taken?	Y No.	Hpylori		Dysplasia	Mild	<input checked="" type="checkbox"/>	Dysplasia			gland polyps
<input checked="" type="checkbox"/> GA	Refused Trial	<input type="checkbox"/> Refused	pH		No. of polyps	>20	<input checked="" type="checkbox"/>	Polyps No.		Size	>. 20 small
<input type="checkbox"/> MRI	Spigelman Grade	IV Est	Endoscopist	STG	Size of polyps	>10 mm	<input checked="" type="checkbox"/>	Location			mm in D2 v
	Ampulla Size mm		Description		Minimally enlarged						snared in si
Notes: OGD: Multiple gastric cystic gland polyps. Ampulla minimally enlarged. There are >											
<input type="checkbox"/> DBE	Endoscopy Date	22/04/2009	Video polyps		UGI Histology	Tubular	<input checked="" type="checkbox"/>	Histology			13/10/09; 0
<input type="checkbox"/> Caps	Biopsies Taken?	Y No.	Hpylori		Dysplasia	Mild	<input checked="" type="checkbox"/>	Dysplasia			13/10/09; 0
<input type="checkbox"/> GA	Refused Trial	<input type="checkbox"/> Refused	pH		No. of polyps	5-20	<input checked="" type="checkbox"/>	Polyps No.		Size	06/05/09; 1
<input type="checkbox"/> MRI	Spigelman Grade	II Est III	Endoscopist	STG	Size of polyps	5-10 mm	<input checked="" type="checkbox"/>	Location			06/05/09; 1
	Ampulla Size mm		Description								06/05/09; 1
Notes: Antral gastritis. Several fundic gland polyps. The duodenal bulb was inflamed and oedematous with a very											
<input type="checkbox"/> DBE	Endoscopy Date	16/04/2008	Video polyps		UGI Histology		<input checked="" type="checkbox"/>	Histology			16/09/10; 1
<input type="checkbox"/> Caps	Biopsies Taken?	N No.	Hpylori		Dysplasia		<input checked="" type="checkbox"/>	Dysplasia			16/09/10; 1
<input type="checkbox"/> GA	Refused Trial	<input type="checkbox"/> Refused	pH		No. of polyps	>20	<input checked="" type="checkbox"/>	Polyps No.		Size	16/09/10; 1
<input type="checkbox"/> MRI	Spigelman Grade	Est III	Endoscopist	NS	Size of polyps	5-10 mm	<input checked="" type="checkbox"/>	Location			16/09/10; 1
	Ampulla Size mm		Description								12/06/08; 0
Notes: (Sed) There are multiple polyps (>20 in number) in duodenum. A couple of polyps were 1cm in size and the rest											



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## Research

- The database can provide extensive information.
- As requirements for research change the database is modified.
- As we learn about more syndromes modifications are needed.
  - The integrity of the database depends on Registry staff who change from year to year
- It is important to be alert to this and work together for completeness of data



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## Importance of expert staff

- Administrative staff: excellent IT and database skills to support nursing team in ensuring patients receive endoscopic surveillance in timely manner
- Nursing staff: highly specialist skills e.g specialist genomic knowledge, family history taking, genetic counselling, nurse-led clinics, independent prescribing, IT skills, in-depth clinical knowledge
- Specialist endoscopists: uniform endoscopy reporting, complex therapeutic endoscopy skills e.g. IOE in PJS
- Specialist surgeons: genomics in surgical decision making, need for low risk prophylactic surgery in adolescents who are asymptomatic (IDSA), a knowledge of pouch surgery and pouch revisional surgery
- Complex decision making around desmoids – very rare tumours



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## Medical and endoscopic staff



- Director: Prof. Sue Clark: Surgeon
- Assistant director: Dr Andrew Latchford
- Laproscopic surgeon: Prof. Omar Faiz
- Paediatric gastroenterologist: Dr Warren Hyer
- Lead nurse/manager: Vicky Cuthill
- Nurse endoscopist: Ripple Man
- Research Fellows: Isabel Martin, Roshani Patel



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## Nursing and administrative team



**Front row from left to right:** Jeshu Chauhan (nurse practitioner), Vicky Cuthill (nurse manager), Janet Paul (senior administrator), Jackie Hawkins (paediatric nurse practitioner)

**Second row from left to right:** Menna Hawkins (nurse specialist), James Cockburn (nurse specialist), Denise Coleman (administrator), Avon Bastion (administrative support clerk).



**Unique: Paediatric Polyposis Nurse**



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## Everything we do hinges on **Cancer Prevention**

“It would be difficult to find a more promising field for the exercise of cancer control than a polyposis family, because both diagnosis and treatment are possible in the precancerous stage and because the results of surgical treatment are excellent.” (C.E. Dukes, 1958)



The lady who had the first colectomy with IRA at St Mark's in 1948 and lived into her 80s

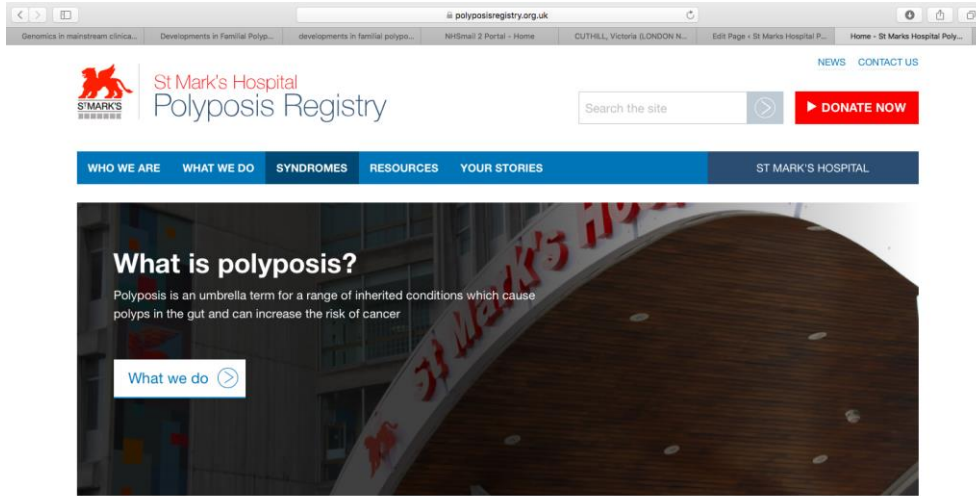




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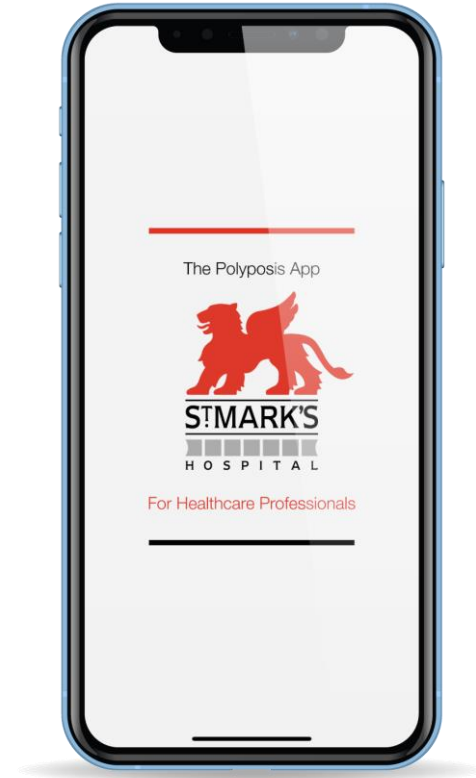


The Polyposis Registry supports people who have a polyposis condition, and their relatives.

Website: [polyposisregistry.org.uk](http://polyposisregistry.org.uk)

Twitter: @PolyposisRegUK  
@VickyChas3

International scientific group: [insight-group.org](http://insight-group.org)



New version launched March 2019!

# Discussion: Communication, Awareness and Reach

Professor Mark Radford led a general discussion regarding raising awareness and communication around genomics.

Round Table members were asked to provide examples of good practice and knowledge of what has worked well previously.

Good quality effective communication is needed. It was recognised that there is a requirement to have different messages for the NHS workforce and for patients, public and their families, but there needs to be a golden thread connecting these.

## **Key messages and points discussed included:**

- Gill Moss gave the example from her trust of using the GEP Genomics in Healthcare 101 module being included in mandatory training for band 6 and offered to 70 band 6/7 staff. It was suggested that this could be mandated further, but an additional slide needs to be included to explain its importance.
- Medicine is changing and nurses and midwives have a role to play.
- The word “genomics” can be off-putting and considered ‘too scientific’. The group felt that it was more about the fact that genomics is transgenerational, about families, communities and society and should be portrayed in that way to make it more accessible.
- The perception that nurses and midwives are already ‘doing genomics’ in their everyday practice e.g. taking family histories, screening etc.

# Discussion: Communication, Awareness and Reach cont..

- There is a need to normalise genomics – this could start with teaching genomics in schools, storylines in soaps and dramas, using real patient stories in advertising, showing how it helps to get answers and stops inappropriate tests.
- There are various one/two-minute animations and videos already available via the GEP website to aid individuals.
- The message could be there is a ‘new tool in the toolkit’ to improve health.
- We need to consider what we mean when we say ‘NHS’. We should think about it broadly and not just in terms of the acute sector.
- Any communication strategy needs to go across the board, teams should not be working or trained in silos. Everyone in the NHS should know something about genomics.
- Ethical issues – data storage and what happens with personal data needs to be clear.
- Golden thread – messaging that appeals to both the public and the workforce. Some overarching principles for a strategic campaign needs to be set before starting the approach,
- Any comms plan should include messaging about adding value and quality, better more efficient services, this needs to be carefully messaged so as not to raise people’s expectations.
- Every patient who comes into contact with healthcare service has the opportunity to discuss genomics with a healthcare professional.
- Need to think about what a campaign means, what awareness raising means and what success looks like.

# Actions and next steps

- GEP and NHSE/I to meet to discuss service need and priorities. To focus the GEP activity, HEE need to know in advance what education and training to develop.
- Institute of Health Visiting to link with Genomics Education Programme to discuss collaborative working.
- GEP to circulate the link to animations developed by the Wellcome Trust Genome Campus.
- Feedback forms will be circulated. Everyone is encouraged to complete these to assess how future meetings should work and improve.



# Summary

Lord Willis of Knaresborough thanked everyone for attending the day and their contributions to the discussions.

He summed up by saying 21<sup>st</sup> century healthcare is not about the individual but is about communities and society and genomics is a great example of families helping other families.

He suggested reading the Born in Bradford study, which is one of the largest research studies in the World, tracking the lives of over 30,000 Bradfordians to find out what influences the health and wellbeing of families. Findings are used to develop new and practical ways to work with families and health professionals to improve the health and wellbeing of communities.

<https://borninbradford.nhs.uk/>



# Round Table Participants

Name	Representing
Lord Willis of Knaresborough	Independent Chair
Professor Mark Radford	NHS England and Improvement & Health Education England
Alison Pope	Genomics Education Programme, Health Education England
Andrew Stewart	NHS England and Improvement
Anji Kadam	Council of Deans
Professor Anna Middleton	Wellcome Trust Genome Campus
Anne Trotter	Nursing and Midwifery Council
Dr Anneke Seller	Genomics Education Programme, Health Education England
Dr Cheryl Adams	Institute of Health Visiting
Dr Christine Patch	Genomics England
Dany Bell	Macmillan Cancer Support
Dawne Garrett	Royal College of Nursing
Dr Edward Miller	Genomics Education Programme, Health Education England
Emma Lord	NHS England and Improvement
Dr Emma Tonkin	University of South Wales
Ethel Rodrigues	Unite Union

# Round Table Participants

Name	Representing
Gail Johnson	Royal College of Midwives
Gill Moss	Manchester University NHS Foundation Trust
Professor Janice Sigsworth	Imperial College Healthcare NHS Trust
Kat Lynch	Genomics Education Programme, Health Education England
Leanne LeRiche	Genomics England Patient Participant
Marie Batey	Imperial College Healthcare NHS Trust
Sarah Armstrong-Klein	NHS England and Improvement
Dr Sharon Barrett	National Institute for Health Research
Sue Boran	Queen's Nursing Institute
Professor Dame Sue Hill	NHS England and Improvement
Vicky Cuthill	London North West University Healthcare NHS Trust

# The GEP team

**Amelia McPherson**  
Programme Editor



**Charlotte Murray**  
Engagement Officer



**Edward Miller**  
Senior Education Development Officer

**Aine Kelly**  
Managing Editor



**Anneke Seller**  
Scientific Director



**Alison Pope**  
Programme Manager



**Michelle Bishop**  
Education Development Lead

**Kathleen Lynch**  
Project Manager



**Claire Healy-West**  
Project Administrator



**Charlotte Szczepanik**  
Project Administrator



**Siobhan Simpson**  
Senior Education Development Officer