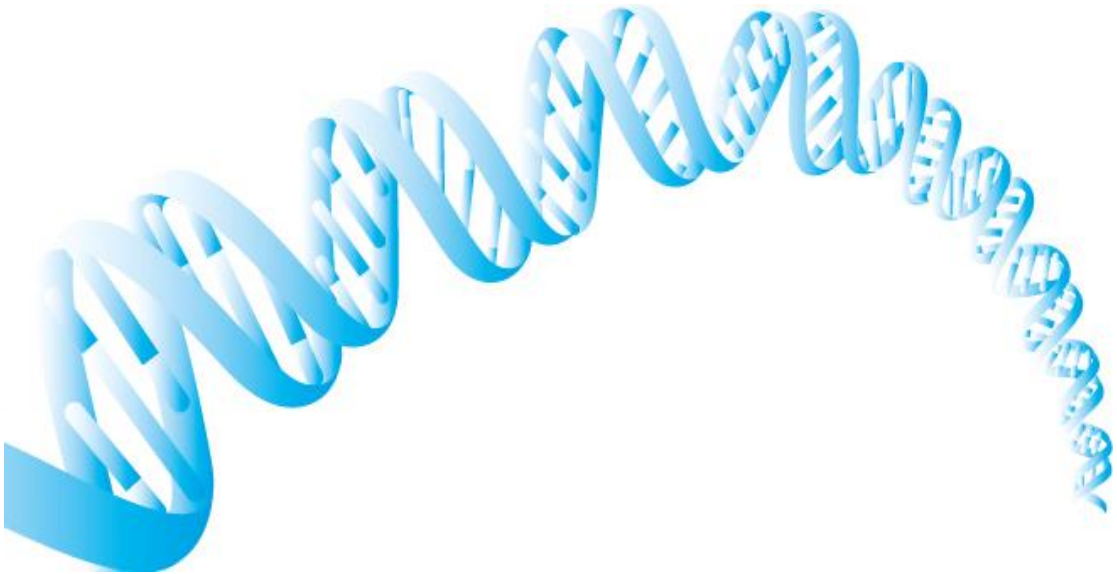
Genomics Education Programme

NHS
Health Education England

HEE Genomics Education Programme Nursing and Midwifery Round Table

Monday 27th January 2020 - The Wesley Hotel & Conference Venue, London.



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Genomics Education Programme

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	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		

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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.

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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, it's 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.

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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 1st 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 Twitter @CSO

Nursing and Midwifery Roundtable

27th 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

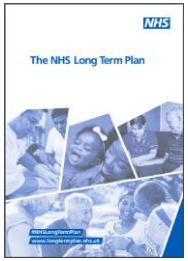
All children with cancer to be offered WGS

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**



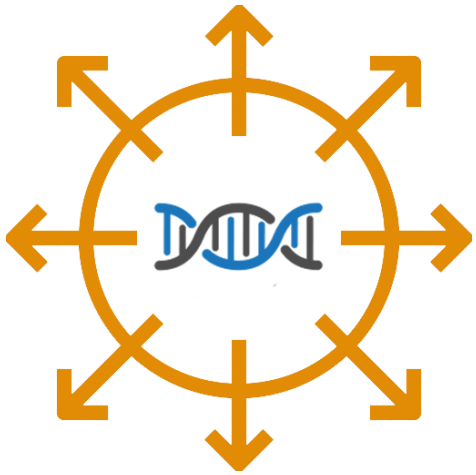
Research

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:

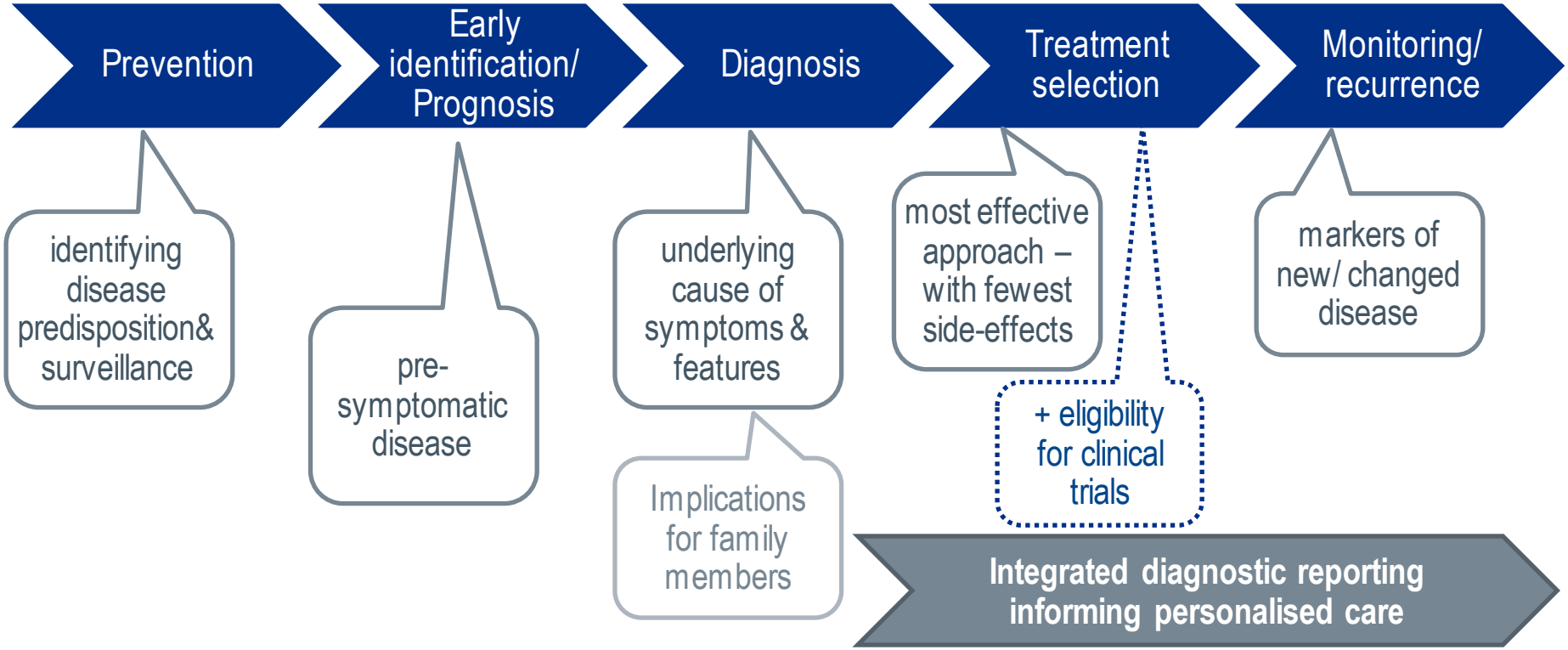
- **National approach** to patient consent, patient & public involvement & strong ethical framework
- **Common national standards**, specifications, protocols, contracts, activity monitoring & pricing frameworks
- **A consolidated national genomic laboratory network** made up of seven Genomic Laboratory 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

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

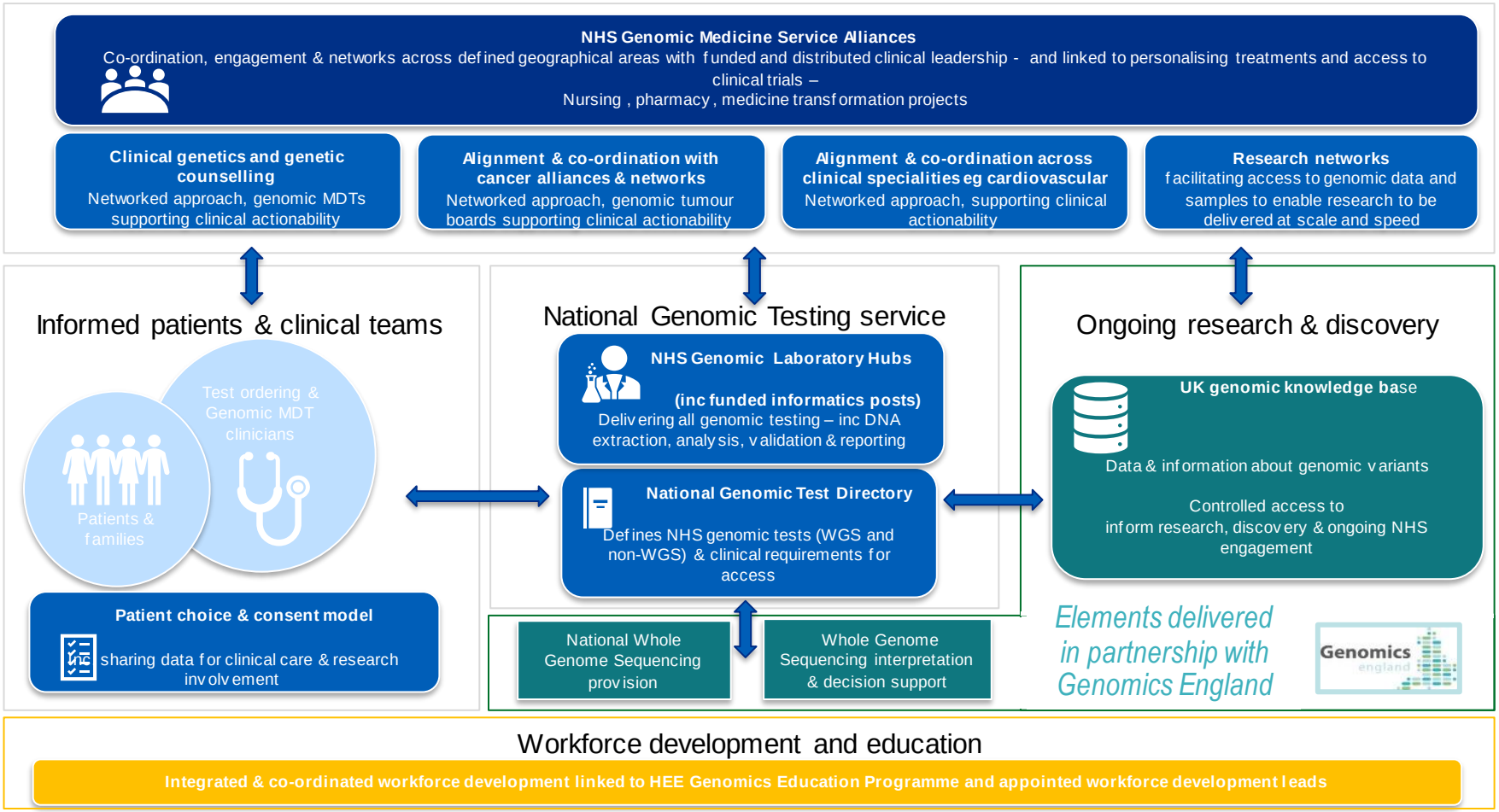
- **A single national testing directory** – covering use of all technologies from single gene to whole genome sequencing inclusive of cancer.
- **A national genomic knowledge base** to inform academic & industry research & discovery including clinical trials recruitment.
- **Single, national co-ordination 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*

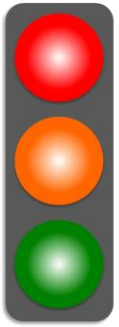
New contract, data and funding model inclusive of cancer linked to NHS Genomic Test Directory with additional funding to support cancer panel development and

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 1st 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

Single gene tests

- Reflex testing
- Extended time to obtain results if multiple tests performed
- Limited scalability
- Limited by sample availability
- Multiple testing labs

Small NGS gene panels

- Multiple genes & disease specific
- Multiple panels required
- Limited by sample availability
- Majority are DNA based so require parallel testing for fusions etc

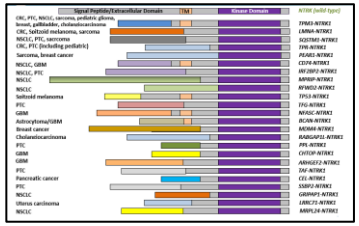
Extensive NGS gene panels DNA & RNA

- Multiple targets for future proofing
- Pan-cancer
- Implement DNA & RNA panels
- High throughput for efficiencies and TATs
- Remove competition with trials CR-UK etc
- Limited number of testing labs

***NTRK* gene fusions** are associated with ≥20 tumor types (1% of solid tumours)

Awaiting NICE approval of pan-cancer drugs

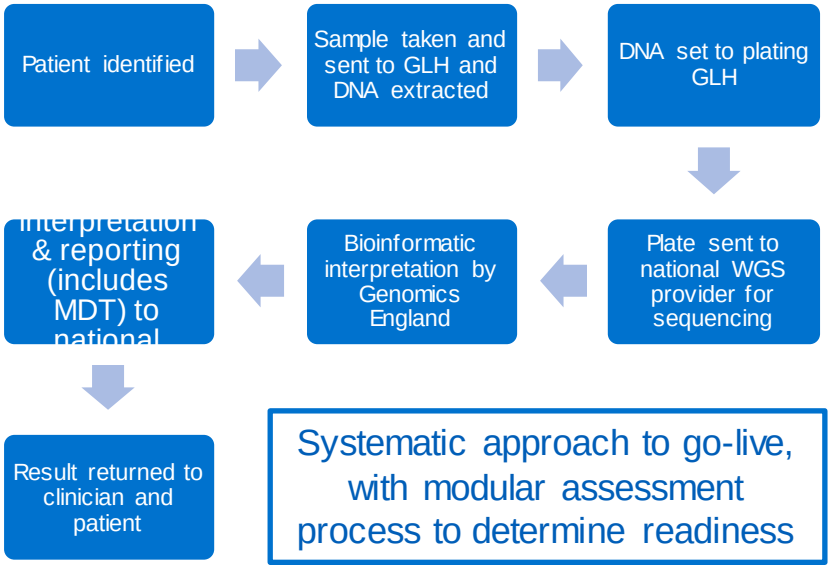
Require clinical testing for eligible patients from outset



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



Systematic approach to go-live, with modular assessment process to determine readiness

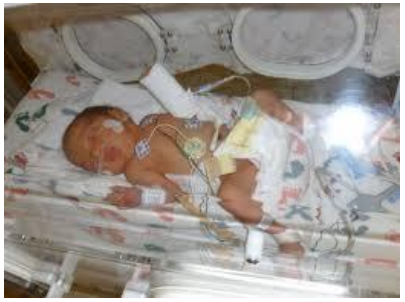
Cancer WGS clinical indications
Sarcoma
Acute Myeloid Leukaemia
Paediatric tumours

Test ID	Clinical Indications for WGS – Phase 1
R89.3	Ultra-rare and atypical monogenic disor
R27.3	Congenital malformation and dysmorph
R29.4	Intellectual disability – microarray, fragil
R69.5	Hypnotic infant with a likely central caus
R104.3	Skeletal dysplasia
R100.3	Rare syndromic craniosynostosis or iso
R143.4	Neonatal diabetes
R98.2	Likely inborn error of metabolism - targe
R54.3	Hereditary ataxia with onset in adulthoo
R55.4	Hereditary ataxia with onset in childhoo
R59.3	Early onset or syndromic epilepsy
R61.4	Childhood onset hereditary spastic para
R83.3	Arthrogryposis
R381.2	Other rare neuromuscular disorders
R84.4	Cerebellar anomalies
R85.2	Holoprosencephaly - NOT chromosoma
R86.3	Hydrocephalus
R87.3	Cerebral malformation
R88.3	Severe microcephaly
R109.3	Childhood onset leukodystrophy
R193.4	Cystic renal disease

Test ID	Clinical Indication WGS – future phases
R125.1	Thoracic aortic aneurysm or dissection
R146.1	Disorders of sex development
R147.1	Growth failure in early childhood
R31.1	Bilateral congenital or childhood onset cataracts
R15.1	Primary immunodeficiency
R352.1	Mitochondrial DNA maintenance disorder
R353.1	Mitochondrial disorder with complex I deficiency
R354.1	Mitochondrial disorder with complex II deficiency
R355.1	Mitochondrial disorder with complex III deficiency
R356.1	Mitochondrial disorder with complex IV deficiency
R357.1	Mitochondrial disorder with complex V deficiency
R63.1	Possible mitochondrial disorder - nuclear genes
R58.1	Adult onset neurodegenerative disorder
R60.1	Adult onset hereditary spastic paraplegia
R62.1	Adult onset leukodystrophy
R78.1	Hereditary neuropathy or pain disorder – NOT PMP22 copy number
R79.1	Congenital muscular dystrophy
R80.1	Congenital myaesthenic syndrome
R81.1	Congenital myopathy
R82.1	Limb girdle muscular dystrophy
R266.1	Neuromuscular arthrogryposis
R195.1	Proteinuric renal disease

Rapid exome sequencing for NICU/PICU

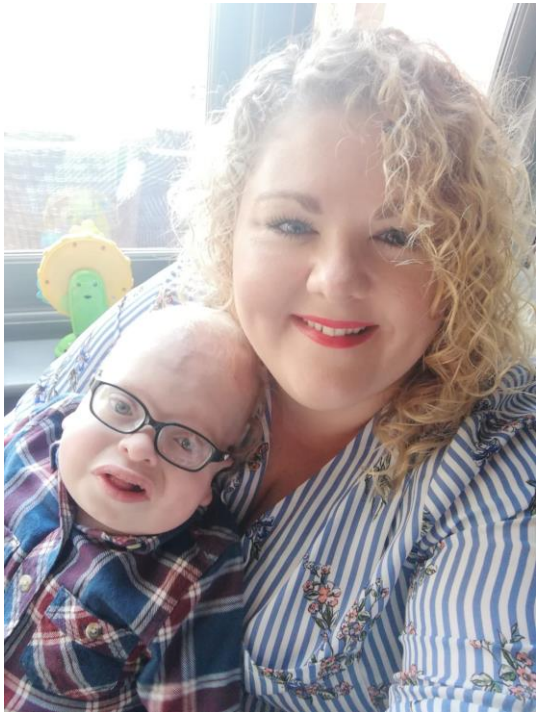
- Nationally commissioned service commenced on October 1st 2019 provided by South West Genomic Laboratory Hub through the Exeter laboratory.
- Rapid genomic test for acutely unwell babies/children with a likely monogenic disorder where a genetic diagnosis would change management
- Put in place:
  - Standardised 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

NHS

Daily Mail

Gene test hope for critically ill babies

Rare diseases diagnosed within days on the NHS

THE Sun

Breakthrough . . . DNA

DNA test tonic for sick kids

The Telegraph

‘Game-changing’ NHS tests diagnose sick children in days

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

sky news

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

ILL BABIES' LIFESAVER DNA TESTS

Genetic mutations pinpointed

DAILY Mirror

NHS rolls out new DNA tests for critically ill babies

INDEPENDENT

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 would 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.

Great Ormond Street Hospital for Children NHS Foundation Trust

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

FOR HEALTHCARE LEADERS HSJ AWARDS

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.

Genomics Education Programme

NHS Health Education England

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 - 27th 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

Nurse & Midwifery-led Collaborative

- **7 Collaboratives across 7 GMS Alliances**
- **7 hub & spokes of local collaborations**
- **Share & learn**
  - Implement local changes
  - National coordination
  - Measures/report/evaluate
- **Embed into practice Collaborative Programme over 24 months**

Building capability & capacity

- **Masterclasses – DoNs, HoMs**
- **Faculty for Nurses & Midwives**
- **Identify essential skill sets**
  - Generic & required in speciality
  - Identify skills gaps
- **Build improvement expertise**
- **Implementation plan to develop teams**
- **Competencies to support clinical practice**
- **Utilise & signpost resources**
- **Links with the pre-reg curriculum**

Large-scale change

- **Communications strategy**
- **Exposure**
  - Conferences & events
  - Publications
- **Royal Colleges & statutory bodies**
- **Genomics standards for Nursing & Midwifery**
- **Work with experts & academics**
- **Networks**
  - G2NA & AGNC
  - Build on Clinical Champions networks
  - New networks
- **New roles**
- **Embed genomics into workforce plans**

Cross cutting themes: Consistency and relevance of language & narrative, use relevant patient stories to area of practice, case studies and best practice examples for sharing widely

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

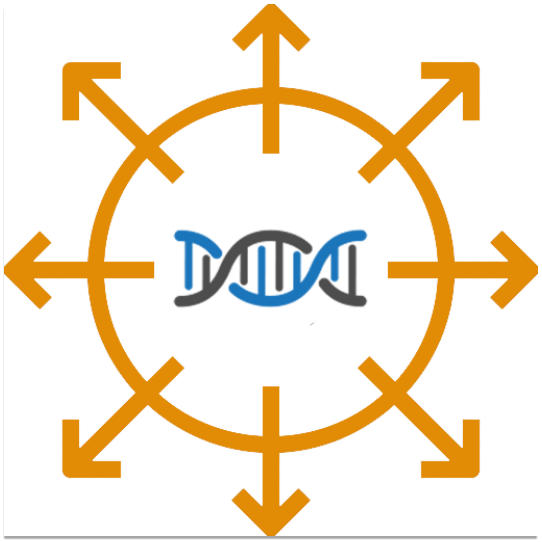
Operational System Leaders:

- Directors of Nursing + Heads of Midwifery

Senior Clinical Leadership:

- Nurses & Midwives - leading change in the Collaboratives

NHS Genomic Medicine Service Alliances (7)



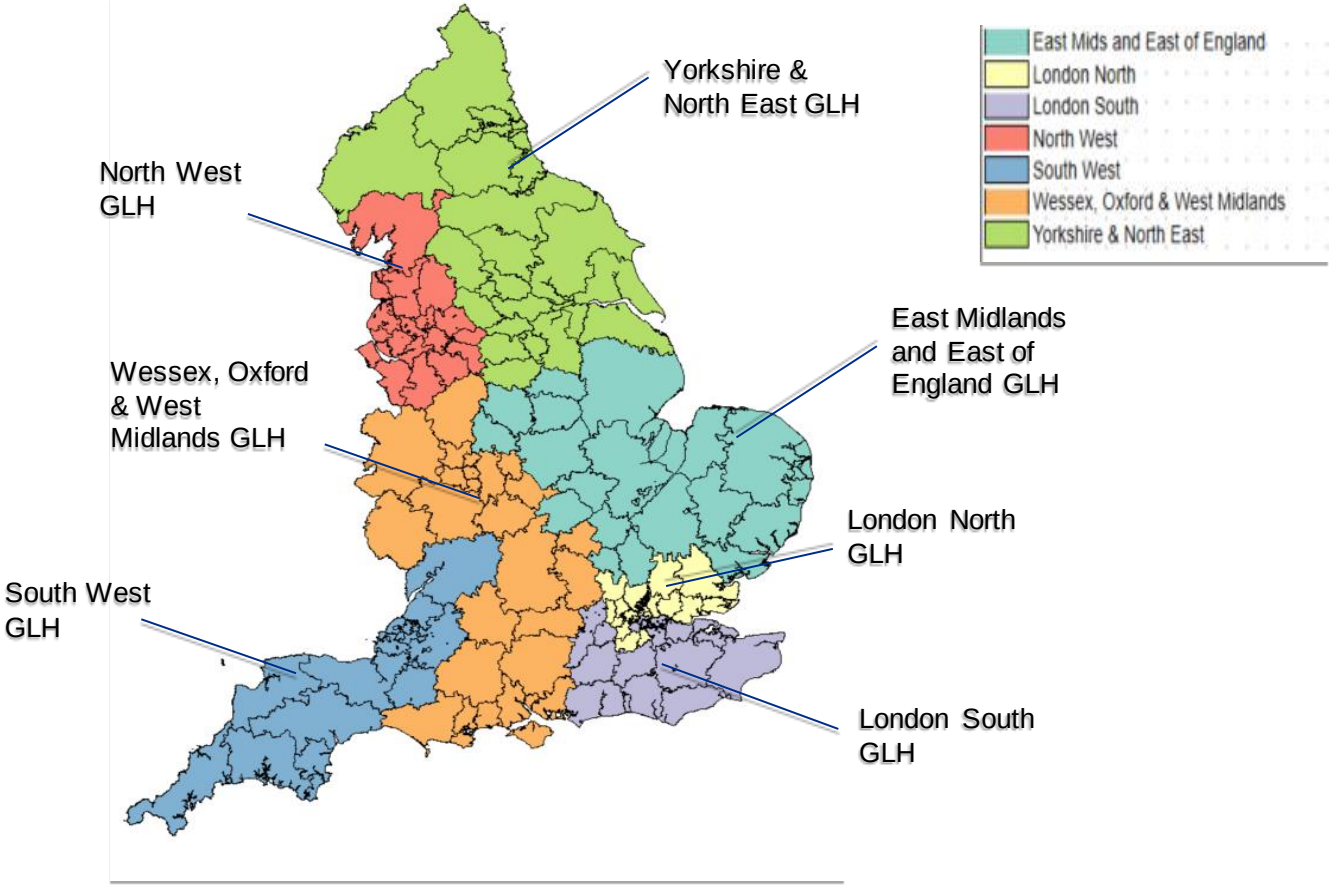
YEAR 2
Implementing the programme

Sustainable implementation: developing service and workforce models

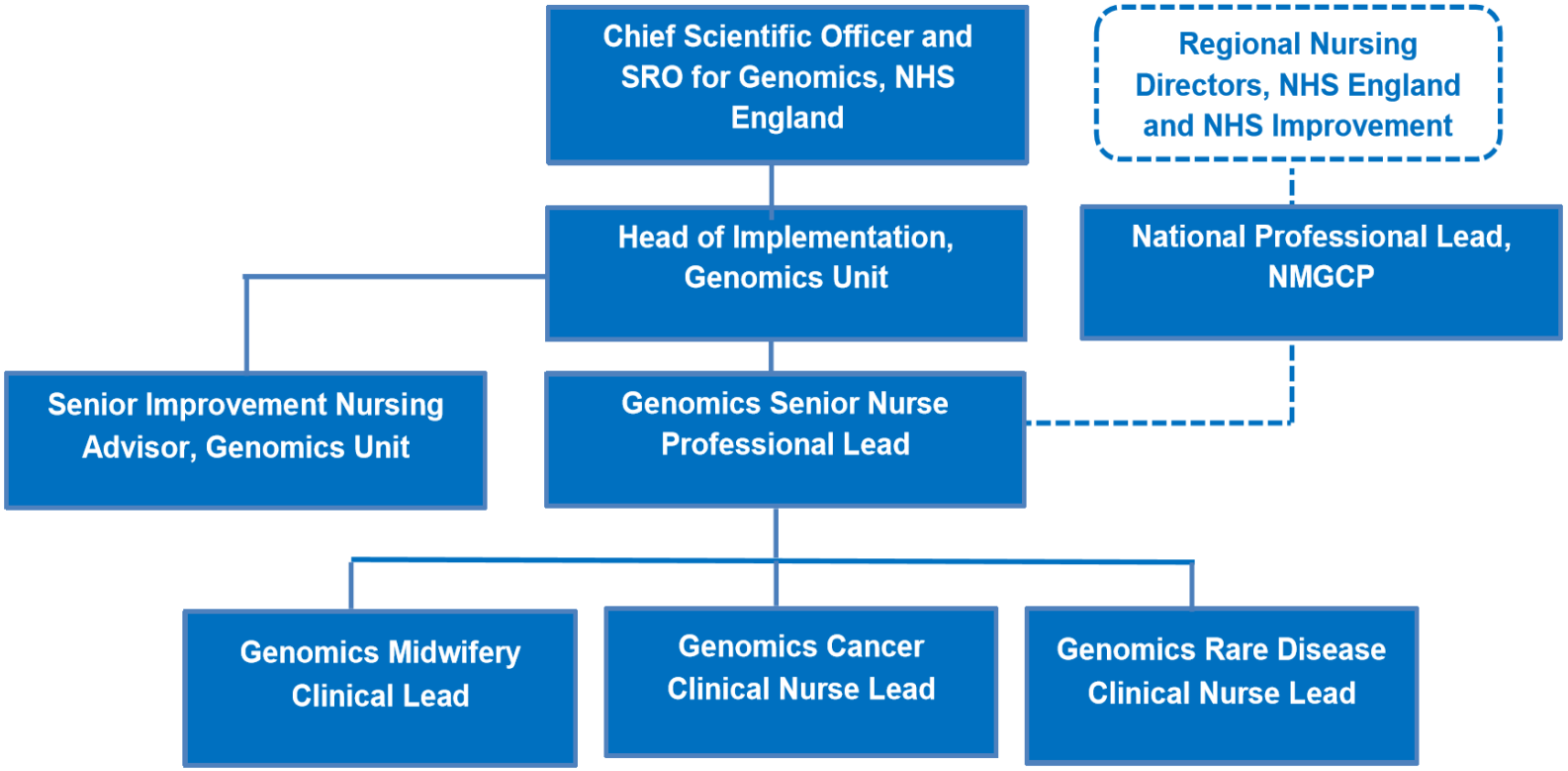
NHS England is supporting three national programmes of work:

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

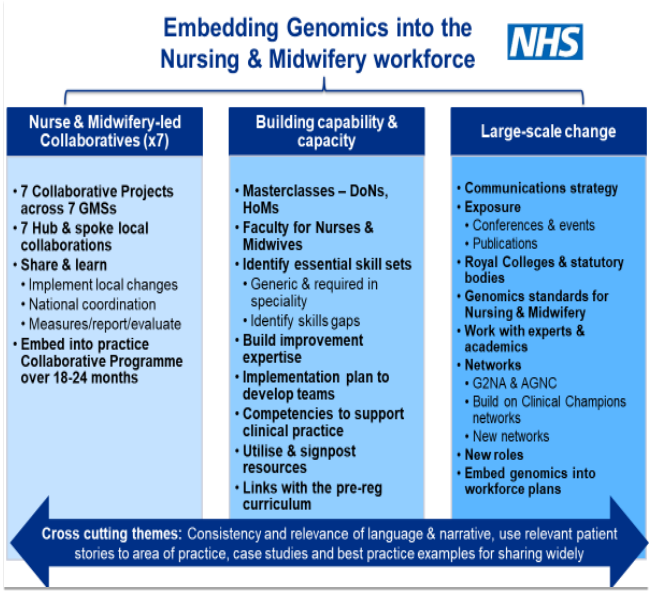


Genomics Nursing & Midwifery Transformation Programme Team



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



Genomics Education Programme

NHS
Health Education England

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.

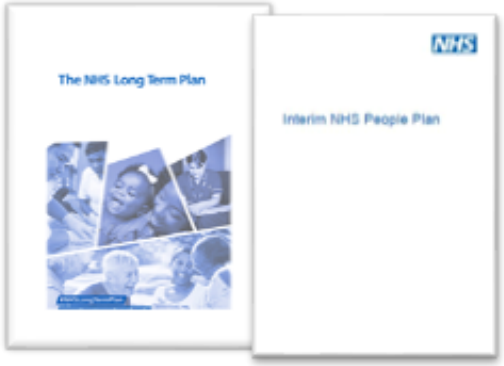
The GEP will assess workforce development, education and training priorities to fully support implementation. Considering the requirements for the prospective and current workforce. Recognising where and when genomics is relevant and identifying what resources are required.

Genomics Education Programme

NHS Health Education England

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?

Genomics Education Programme

NHS Health Education England

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**.

Genomics Education Programme

NHS
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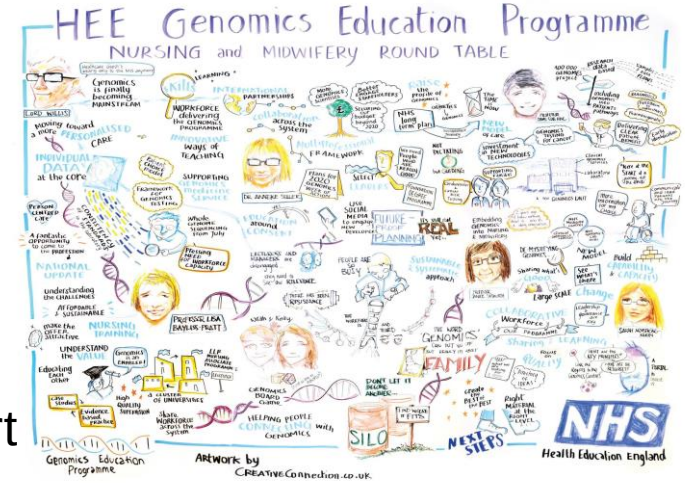
How the GEP is supporting the nursing and midwifery workforce

- **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

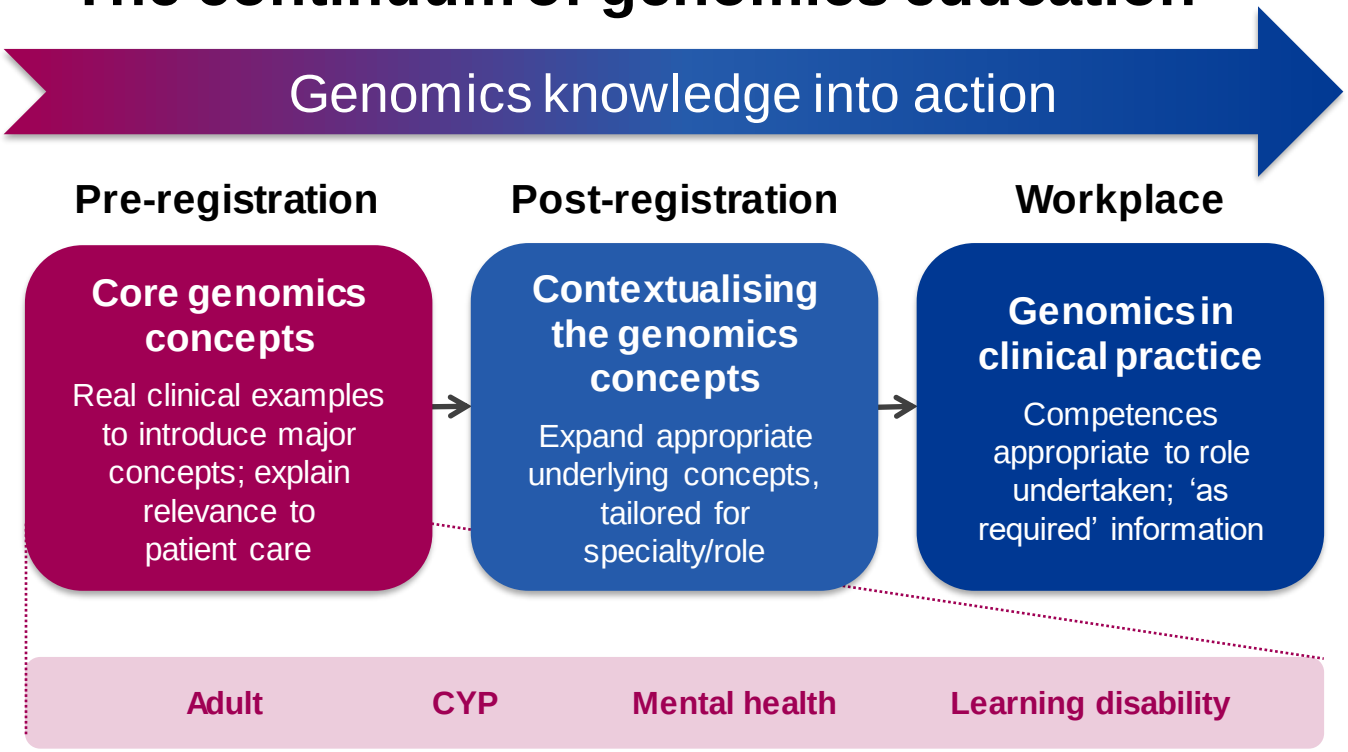


Genomics Education Programme

NHS Health Education England

Recognising where genomics is relevant

The continuum of genomics education



GEP activity

- ✓ Development of competency frameworks
- ✓ Development of learning resources
- ✓ Development of resources to support educators

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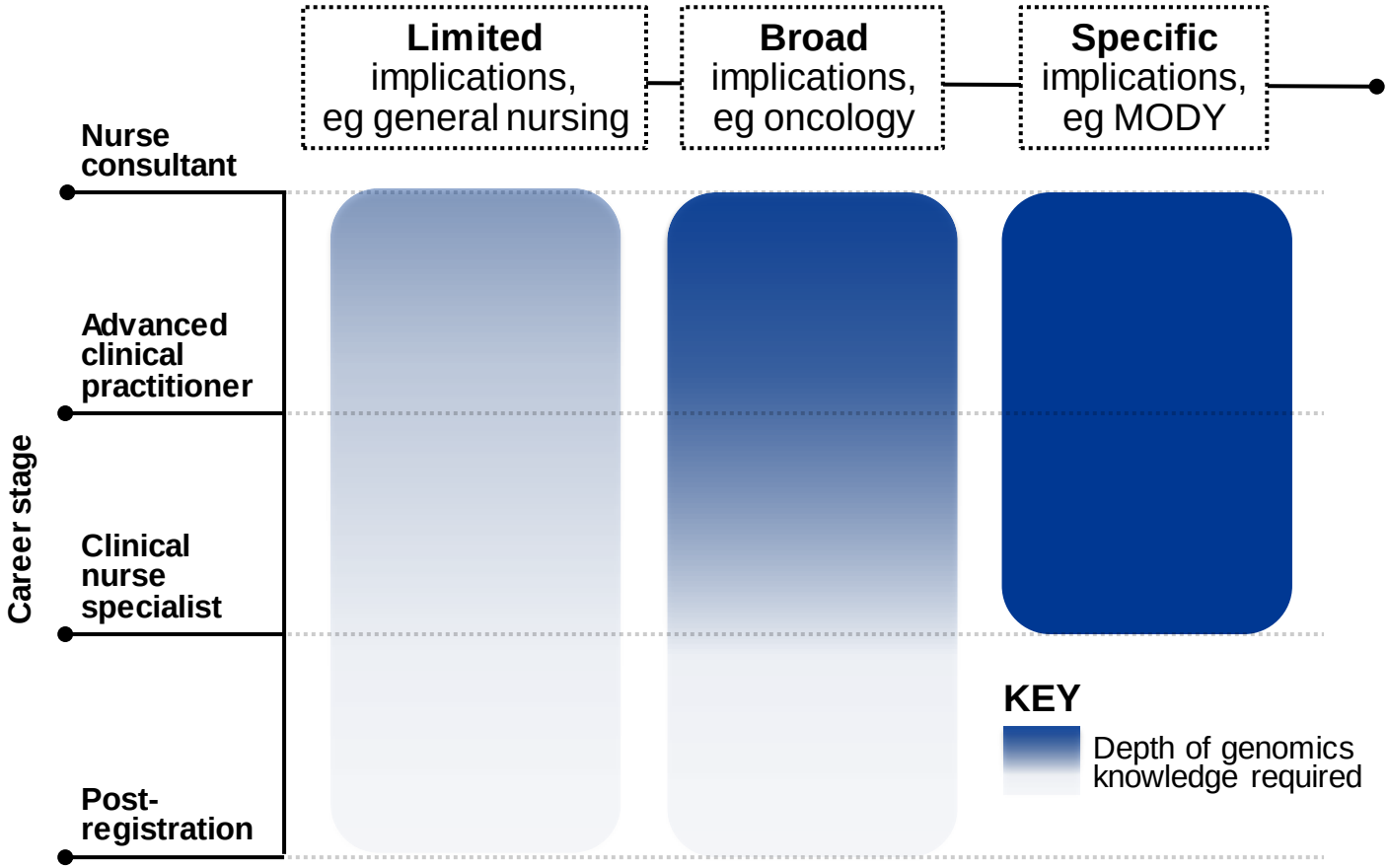
NHS Health Education England

Nursing in genomics: knowledge map



GEP activity

- ✓ Development of competency frameworks
- ✓ Development of learning resources

Genomics Education Programme

NHS Health Education England

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

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NHS Health Education England

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

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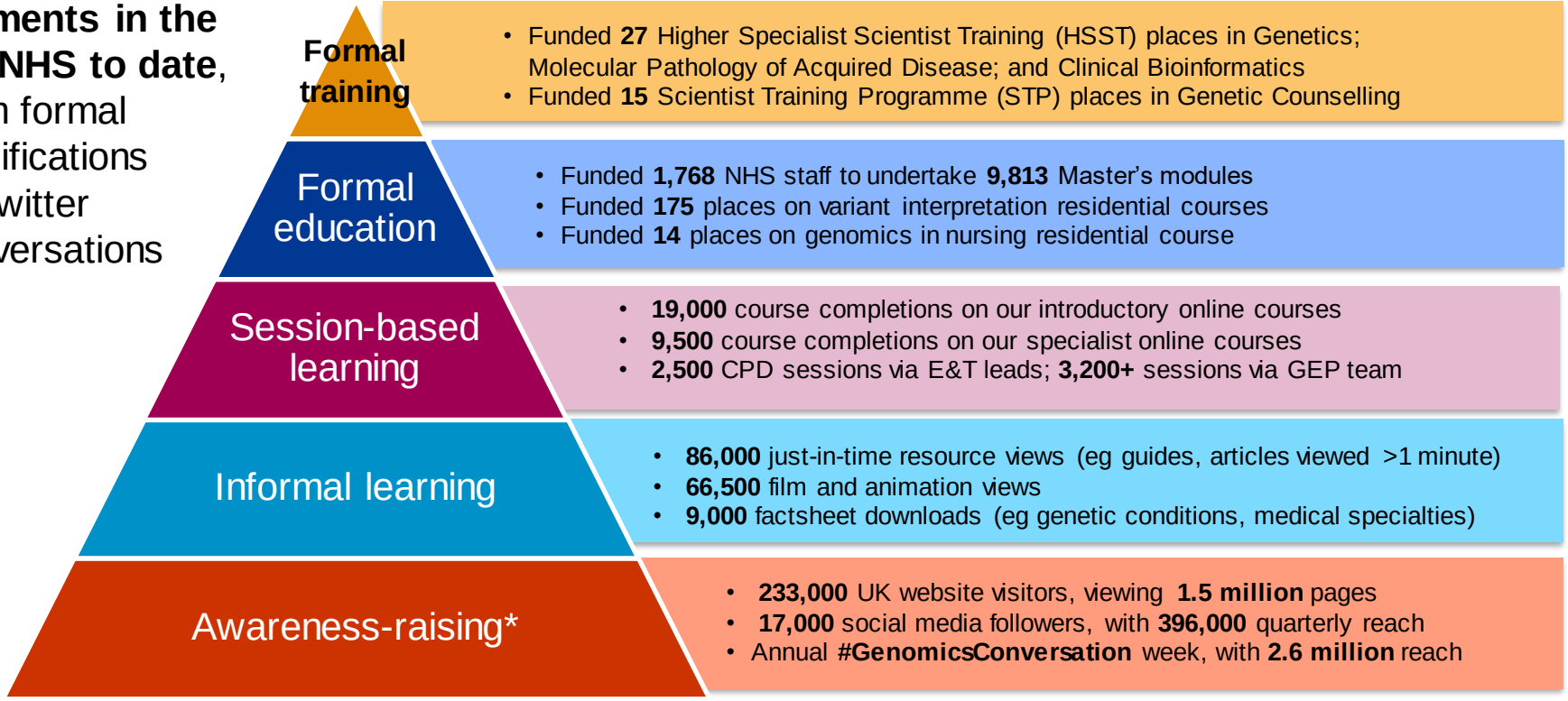
Educational opportunities

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 31st Dec 2019, online data rounded to nearest 500.

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GEP Resources: Week of Action 2020

Genomics Conversation March 16-20th 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"

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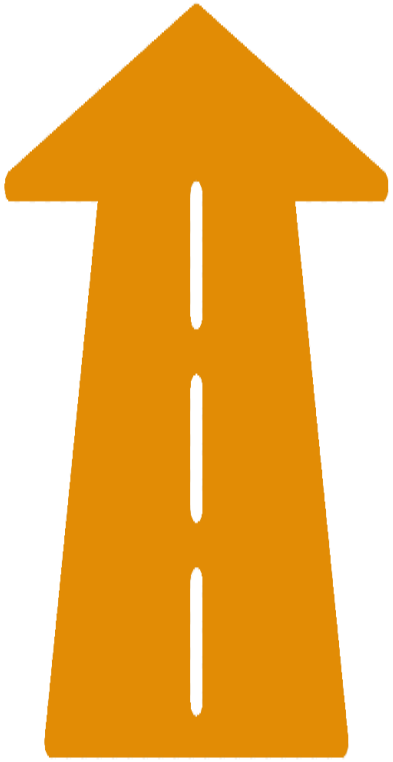
Going forward…….



- 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

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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

Genomics Education Programme

NHS
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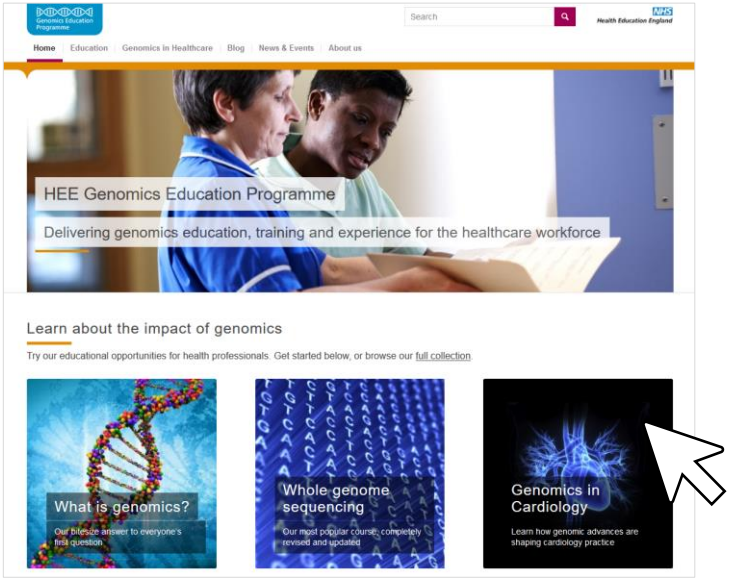
Find out more

Web: 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

Genomics Education Programme

NHS Health Education England

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

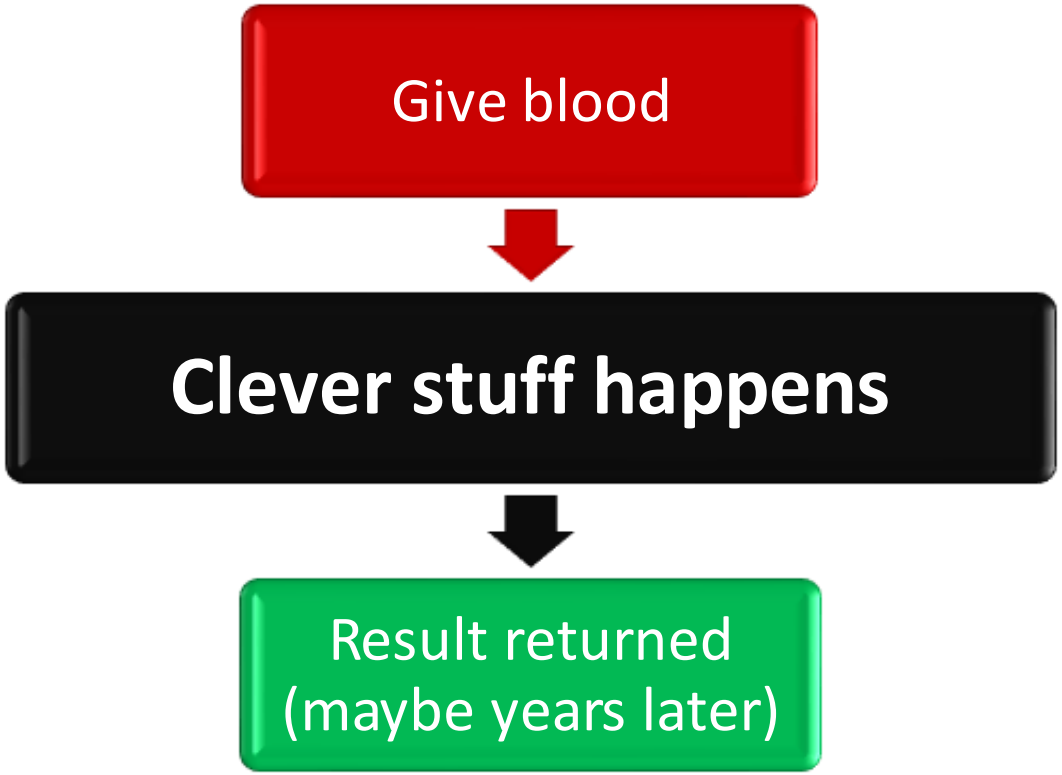
My story

- 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.

Genomics england

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

Pharmacogenomics

- 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

Genomics england

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 ethics consistency

'cures' ensuring consent common & transparent language

- **Patient public engagement** - *awareness of genomics, what it means to the patient - personalised health, children & young people – schools, colleges & 6th 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

Genomics Education Programme

NHS Health Education England

Dany Bell - Macmillan Genomics Portfolio of Activity

Dany Bell, Strategic Advisor Treatment, New Medicines and Genomics presented the portfolio of work that Macmillian 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

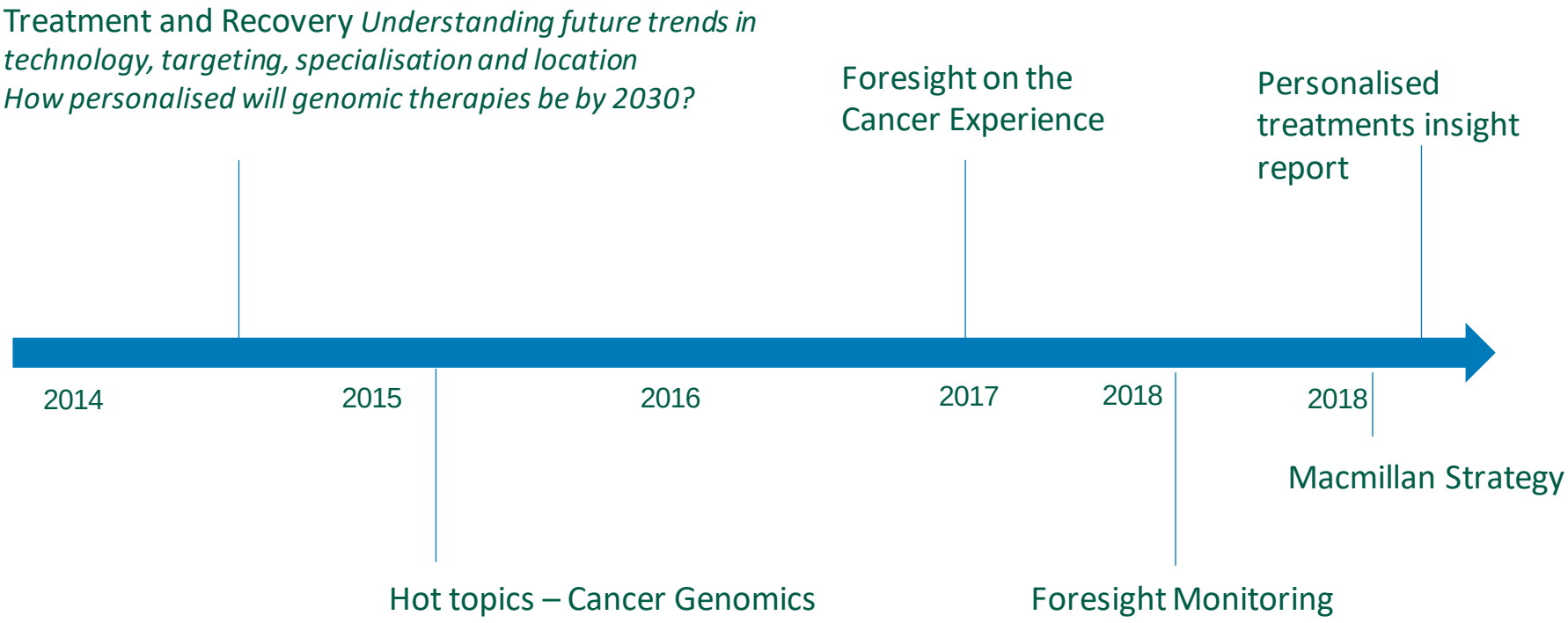
MACMILLAN CANCER SUPPORT

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 CANCER SUPPORT

Macmillan evidence and insight to date for Genomics and Personalised Medicine



Asking PLWC to be involved

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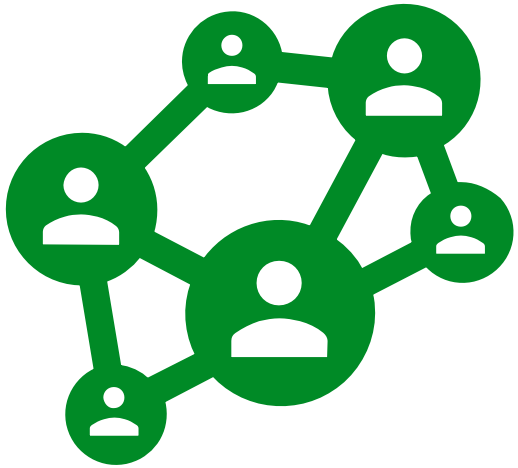
Collaboration with NHSE/HEE

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



MACMILLAN CANCER SUPPORT

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

MACMILLAN CANCER SUPPORT

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

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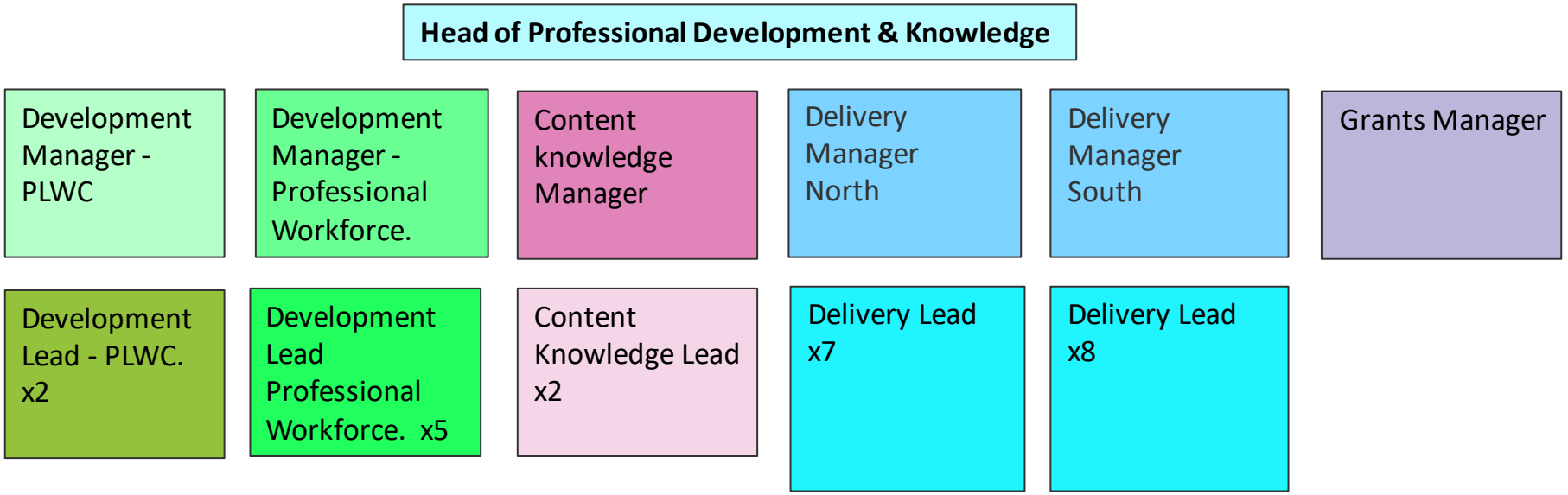
Genomic Education Champion posts

- 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



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Membership for Collaborative Partnership

Sarcoma UK
The bone & soft tissue cancer charity

the eve appeal

amrc
ASSOCIATION OF MEDICAL RESEARCH CHARITIES

Brain Tumour Research
Together we will find a cure

Bowel Cancer UK
Beating bowel cancer together

Bloodwise
The blood cancer research charity

CANCER RESEARCH UK

TARGET OVARIAN CANCER
For every woman. For life.

MACMILLAN CANCER SUPPORT

ovacome
ovarian cancer

NHS
Health Education England

NHS
England

Genomics england

BREAST CANCER NOW
The research & care charity

MACMILLAN CANCER SUPPORT

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

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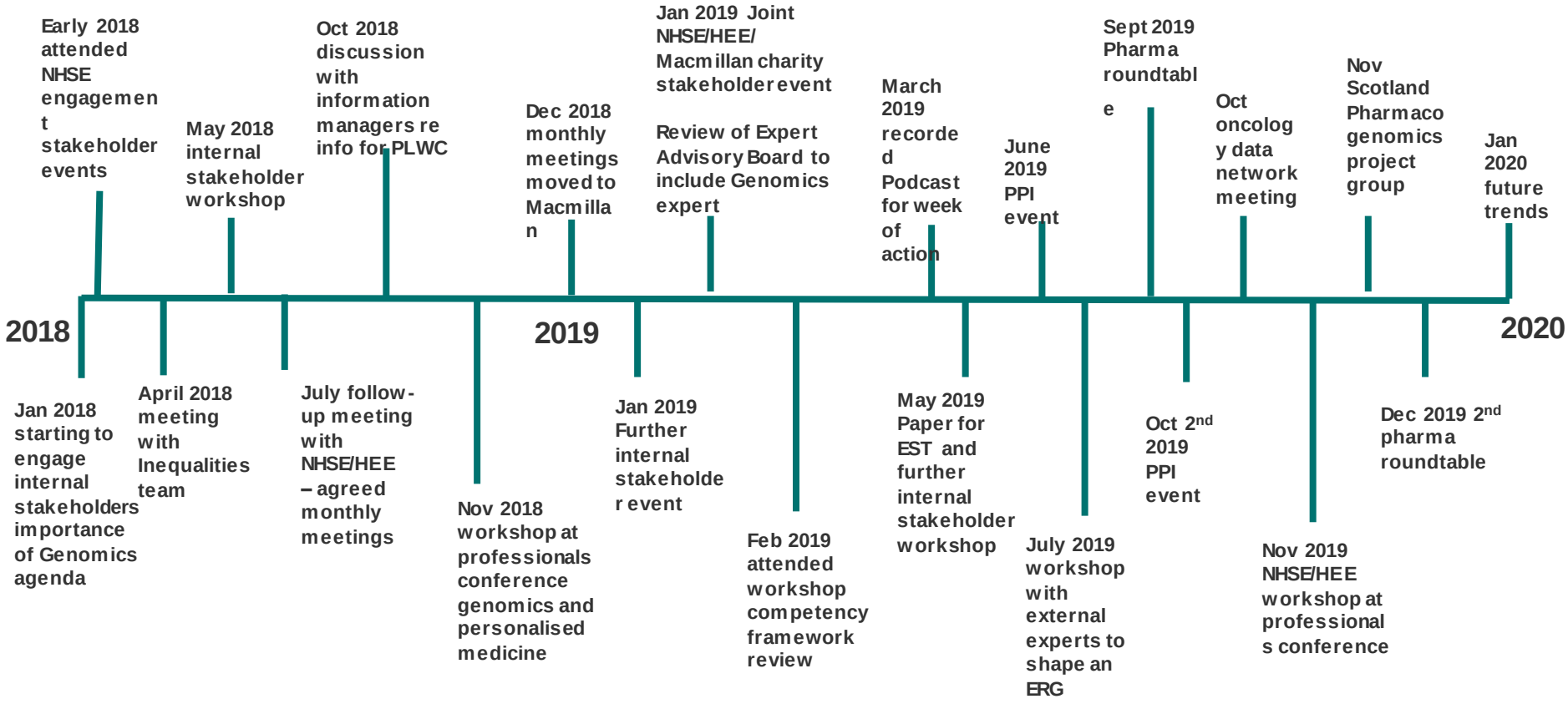
Pharma roundtable - Understand the opportunities for horizon scanning to inform future support offers and start a dialogue and explore opportunities to collaborate

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

MACMILLAN CANCER SUPPORT

Activity timeline



Genomics Education Programme

NHS Health Education England

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

Honesty Equality Accountability Respect Teamwork

Putting **patients** at the **HEART** of everything we do

St MARK'S HOSPITAL

NHS
London North West Healthcare
NHS Trust

Mainstreaming – The Polyposis Registry Example

Vicky Cuthill
Lead Nurse/Manager
The Polyposis Registry and Family Cancer Clinc
St Mark's Hospital

27th January 2020

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

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:

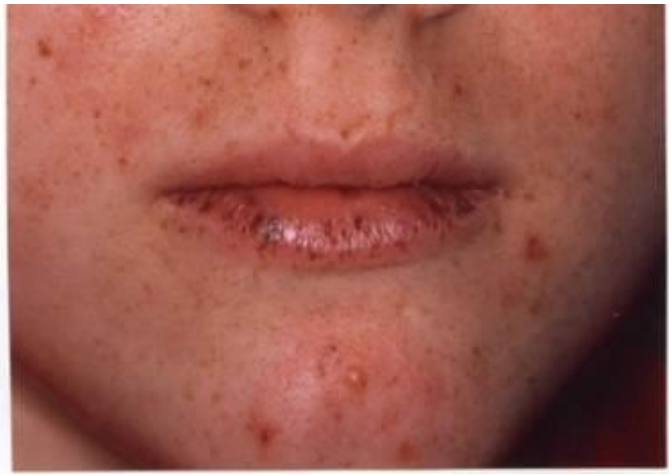
- Adenomatous polyps – FAP, MAP, PPAP
- Hamartomatous polyps – JPS, PJS, PTEN
- Serrated polyps - SPS

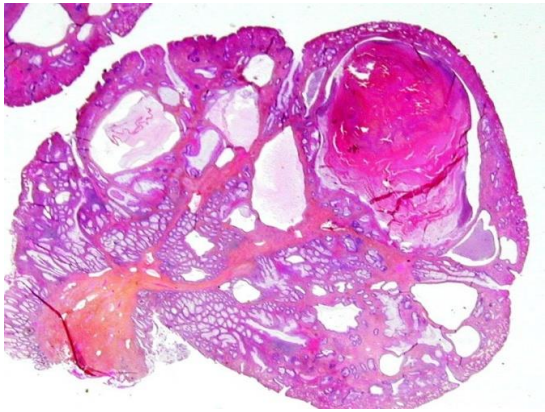
A little bit about the history of polyposis…..

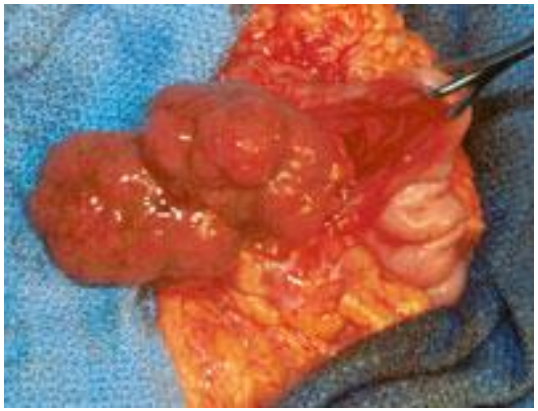
- First description of multiple polyps dates back to 1721
- Prior to the 1860s, autopsy findings informed the literature – observational and linked to intestinal inflammation
- Sklifasowski published the first histologically verified case of adenomatous polyposis in Russia in 1881
- In 1882, inherited 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

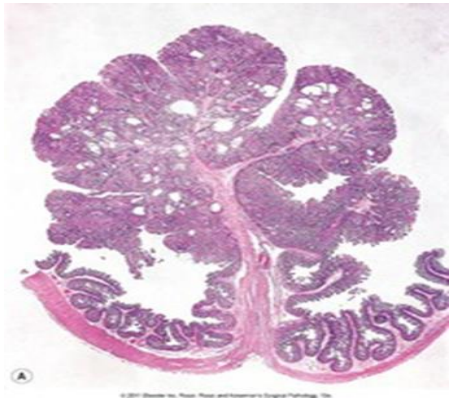
	Polyposis Registry Timeline
1924	Started as a research project, becoming first Polyposis Registry in the world
1940s	Inheritance pattern established
1948	First colectomy with IRA for FAP
1962	Morson publishes 'precancerous lesions of the colon and rectum'
1975	HJR Bussey thesis published 'Familial Polyposis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

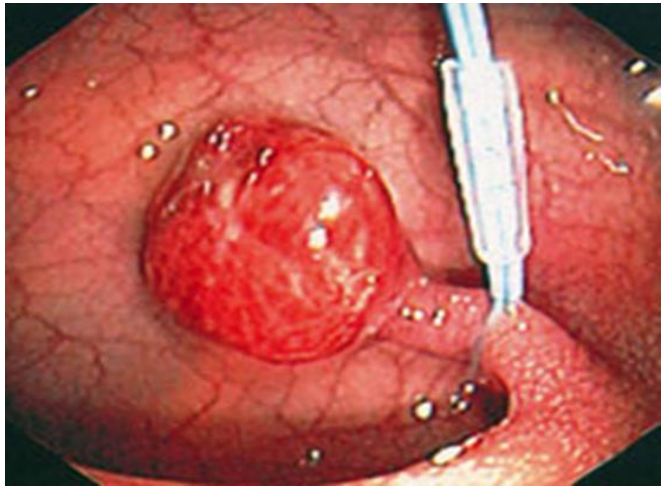










	Polyposis Registry Timeline
1985	The Registry becomes a formal hospital department
1987	Bodmer et al. identifies region on Chr 5 of *APC* gene
1989	Spigelman et al. publish in The Lancet, initiating upper GI screening programme
1996	Clinically responsible for genetic counselling/NHS genetic testing commences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

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

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

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

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

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

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

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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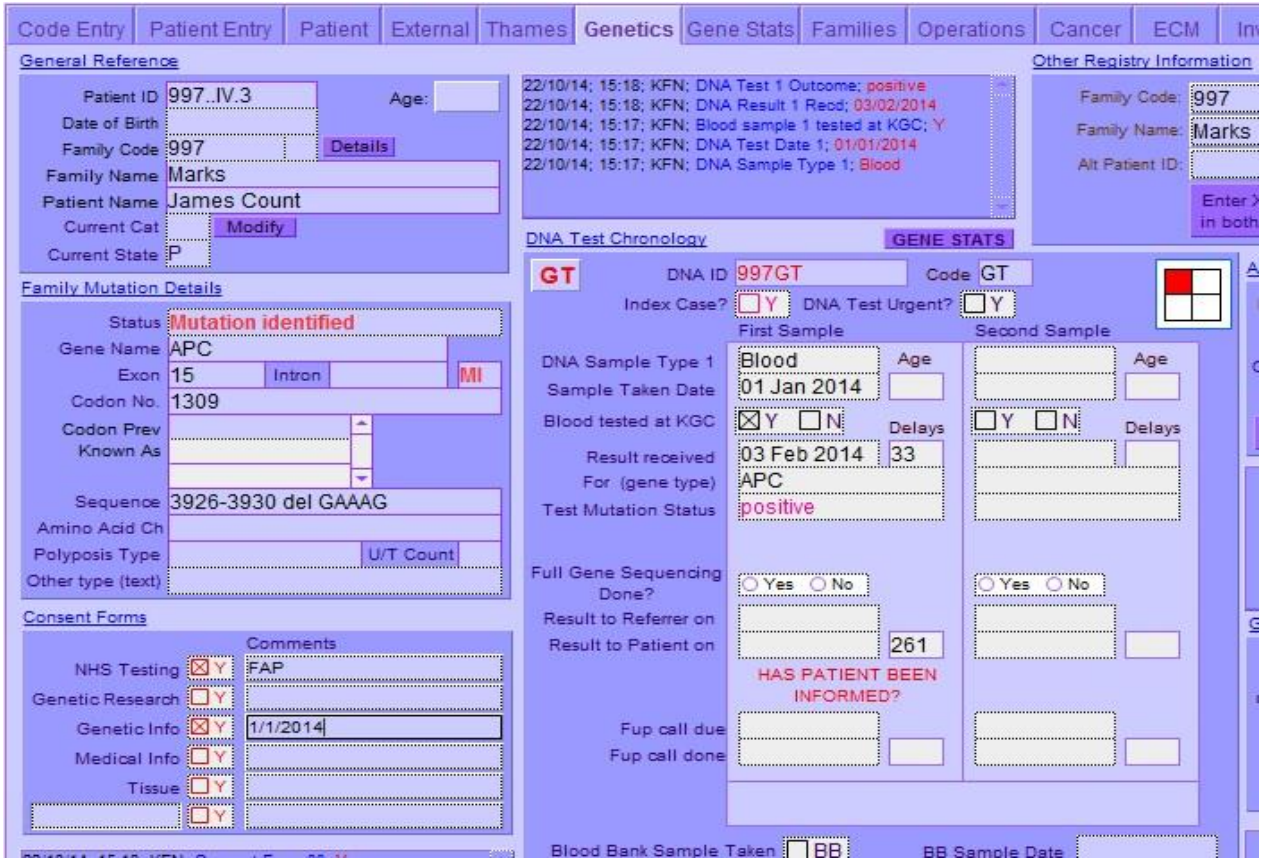
Cancer Prevention by Cascade Testing

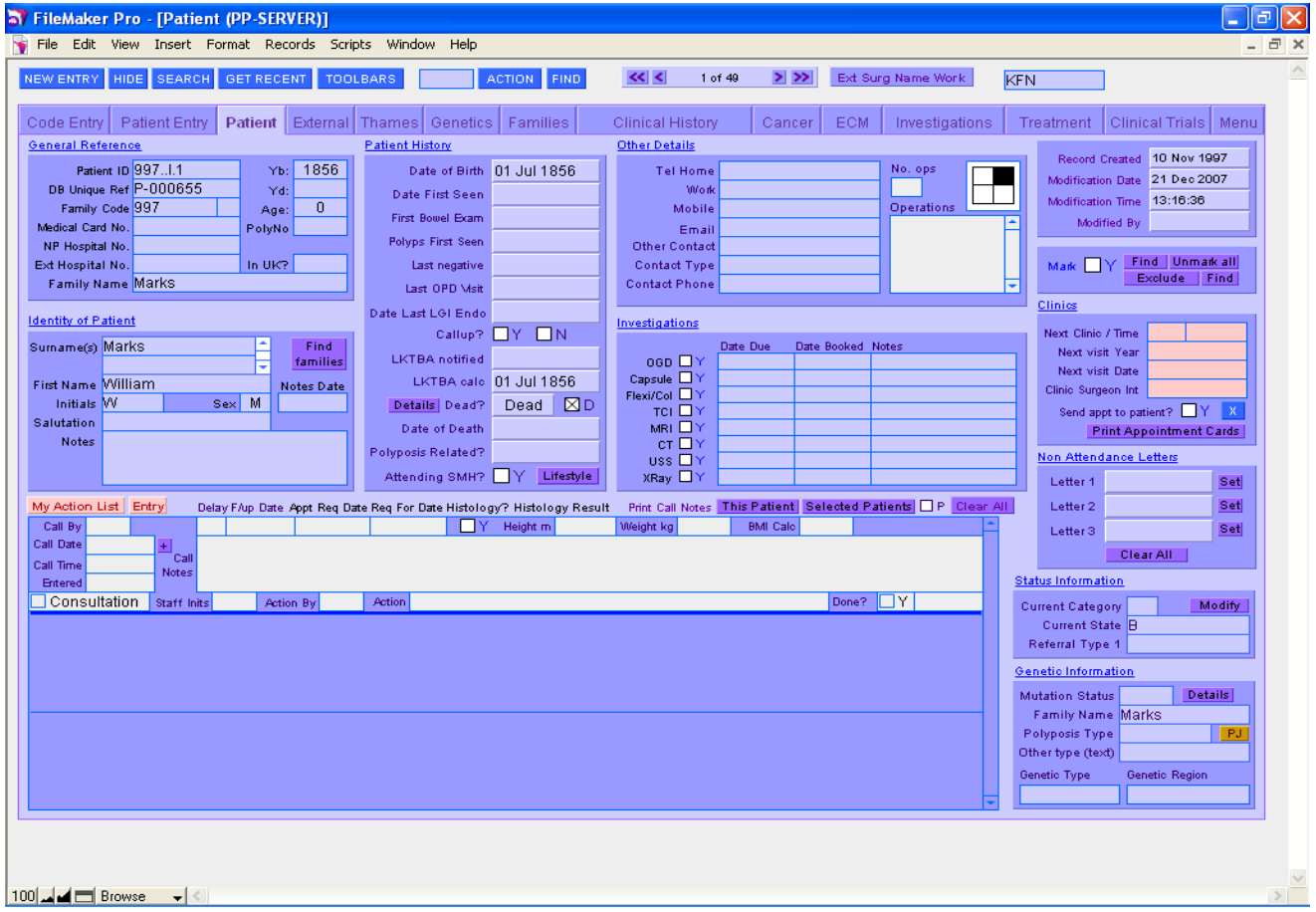


Cancer prevention by monitoring attendance

- 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

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


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 ID 997..I.1
DB Unique Ref P-000655
Family Code 997
Medical Card No.
NP Hospital No.
Ext Hospital No.
Family Name Marks
Yb: 1856
Yd:
Age: 0
PolyNo
In UK?
Identity of Patient
Surname(s) Marks
First Name William
Initials W
Sex M
Salutation
Notes
Find families
Notes Date
Patient History
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 calc 01 Jul 1856
Details Dead? Dead D
Date of Death
Polyposis Related?
Attending SMH? Y Lifestyle
Other Details
Tel Home
Work
Mobile
Email
Other Contact
Contact Type
Contact Phone
No. ops
Operations
Investigations
Date Due Date Booked Notes
OGD Y
Capsule Y
Flexi/Col Y
TCI Y
MRI Y
CT Y
USS Y
XRay Y
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 Modify
Current State B
Referral Type 1
Genetic Information
Mutation Status Details
Family Name Marks
Polyposis Type PJ
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
Y Height m Weight kg BMI Calc
Consultation
Staff Inits Action By Action Done? Y
100 Browse

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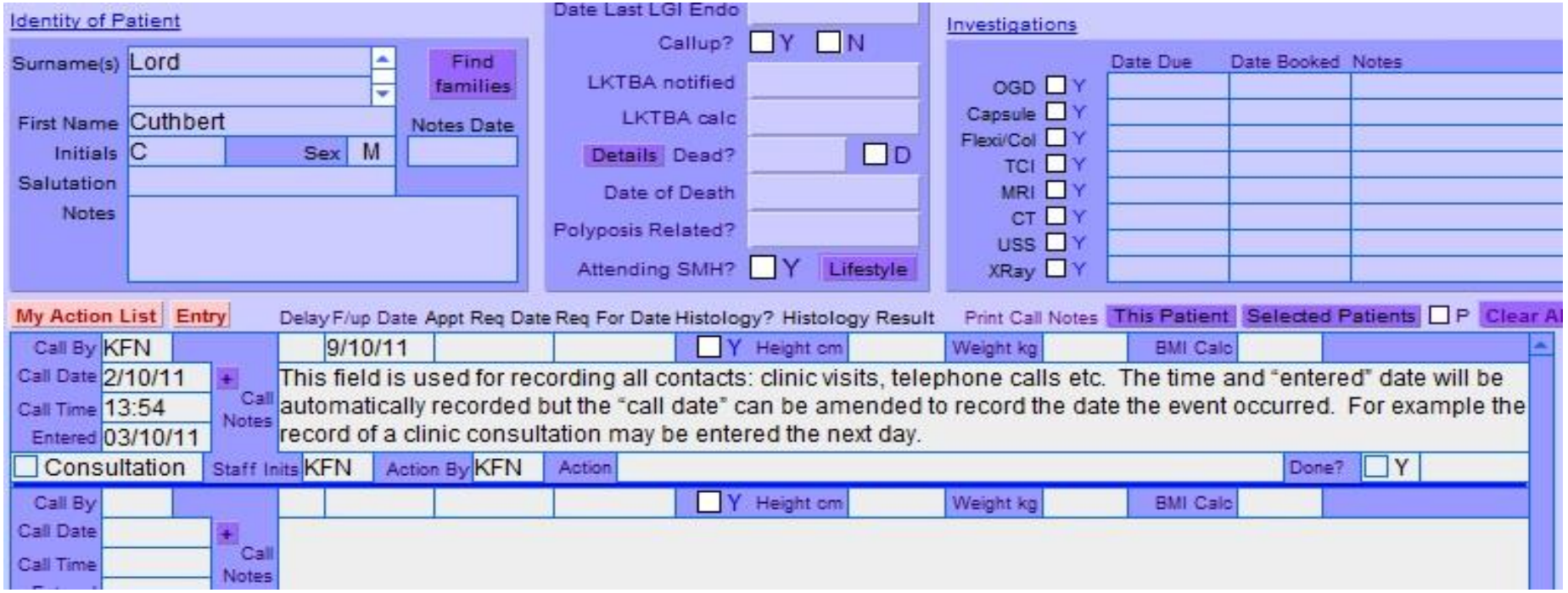
Cancer Prevention by monitoring surveillance



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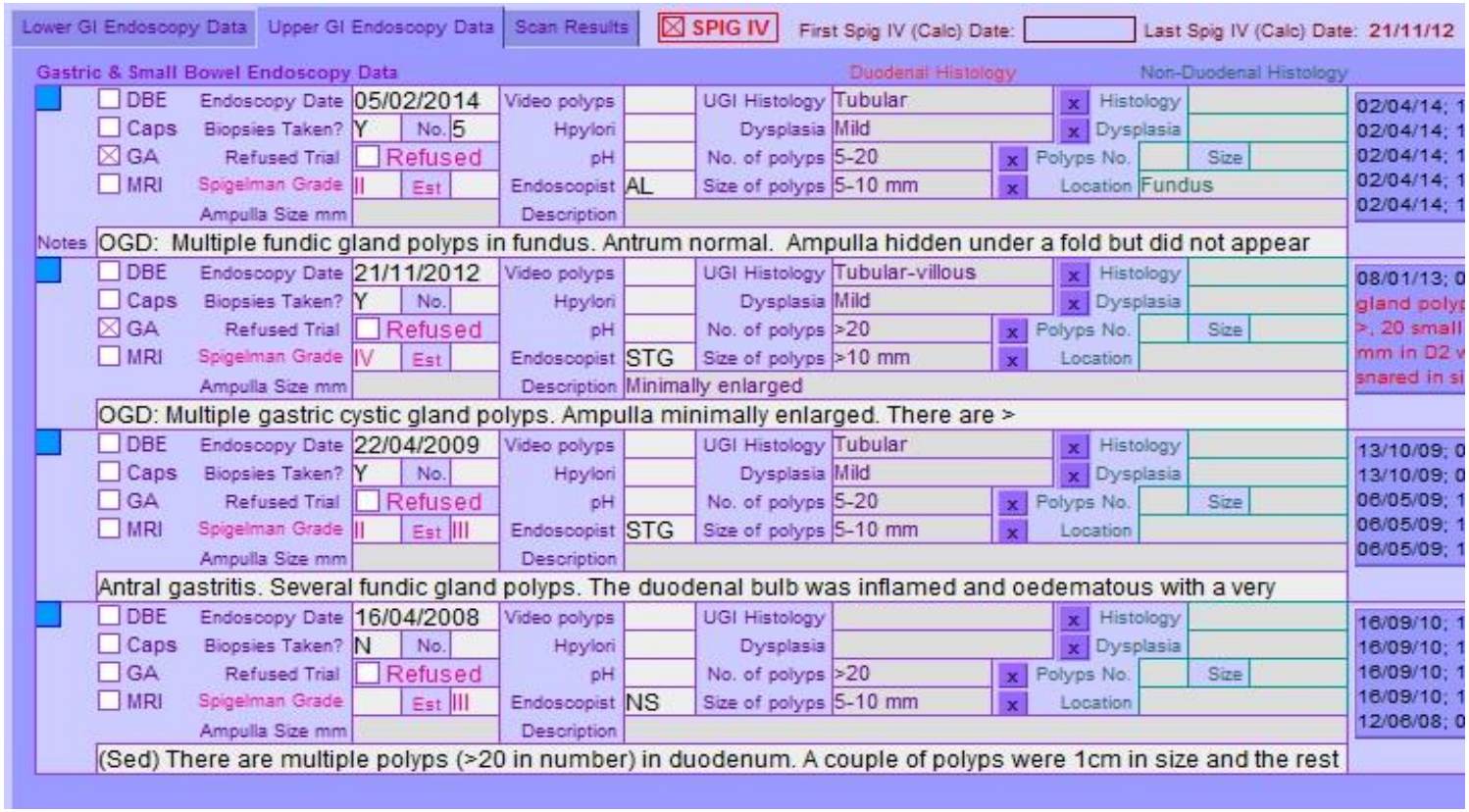
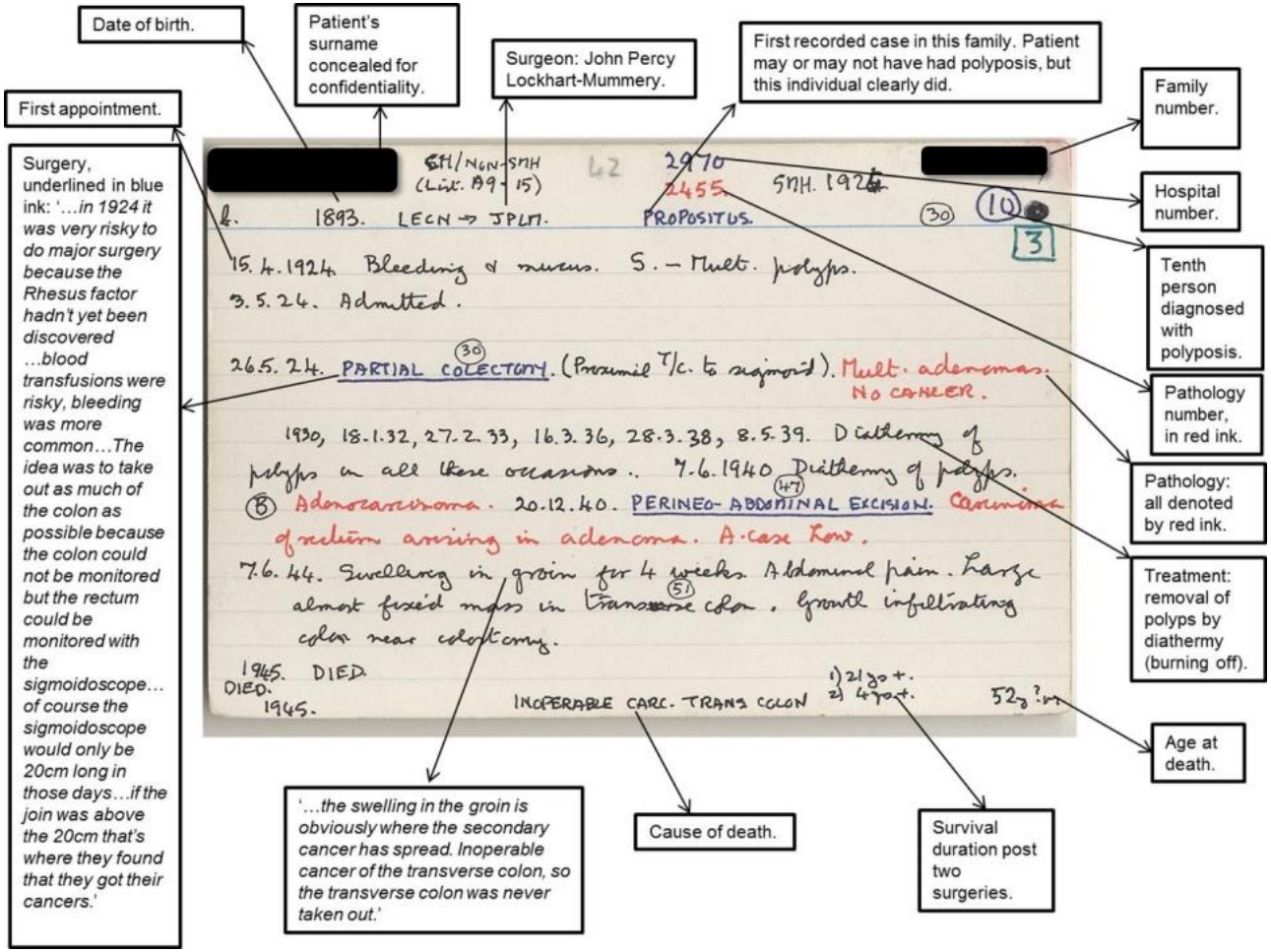
Cancer Prevention by monitoring surveillance



Lower GI Endoscopy Data | Upper GI Endoscopy Data | Scan Results | ☒ SPIG IV | First Spig IV (Calc) Date: | Last Spig IV (Calc) Date: 21/11/12

Gastric & Small Bowel Endoscopy Data — Duodenal Histology — Non-Duodenal Histology

☐ DBE	Endoscopy Date	05/02/2014	Video polyps		UGI Histology	Tubular	x	Histology				02/04/14; 1
☐ Caps	Biopsies Taken?	Y No. 5	Hpylori		Dysplasia	Mild	x	Dysplasia				02/04/14; 1
☒ GA	Refused Trial	☐ Refused	pH		No. of polyps	5-20	x	Polyps No.		Size		02/04/14; 1
☐ MRI	Spigelman Grade	II Est	Endoscopist	AL	Size of polyps	5-10 mm	x	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											
☐ DBE	Endoscopy Date	21/11/2012	Video polyps		UGI Histology	Tubular-villous	x	Histology				08/01/13; 0
☐ Caps	Biopsies Taken?	Y No.	Hpylori		Dysplasia	Mild	x	Dysplasia				gland polyp
☒ GA	Refused Trial	☐ Refused	pH		No. of polyps	>20	x	Polyps No.		Size		>, 20 small
☐ MRI	Spigelman Grade	IV Est	Endoscopist	STG	Size of polyps	>10 mm	x	Location				mm in D2 v
	Ampulla Size mm		Description	Minimally enlarged								snared in si
	OGD: Multiple gastric cystic gland polyps. Ampulla minimally enlarged. There are >											
☐ DBE	Endoscopy Date	22/04/2009	Video polyps		UGI Histology	Tubular	x	Histology				13/10/09; 0
☐ Caps	Biopsies Taken?	Y No.	Hpylori		Dysplasia	Mild	x	Dysplasia				13/10/09; 0
☐ GA	Refused Trial	☐ Refused	pH		No. of polyps	5-20	x	Polyps No.		Size		06/05/09; 1
☐ MRI	Spigelman Grade	II Est III	Endoscopist	STG	Size of polyps	5-10 mm	x	Location				06/05/09; 1
	Ampulla Size mm		Description									06/05/09; 1
	Antral gastritis. Several fundic gland polyps. The duodenal bulb was inflamed and oedematous with a very											
☐ DBE	Endoscopy Date	16/04/2008	Video polyps		UGI Histology		x	Histology				16/09/10; 1
☐ Caps	Biopsies Taken?	N No.	Hpylori		Dysplasia		x	Dysplasia				16/09/10; 1
☐ GA	Refused Trial	☐ Refused	pH		No. of polyps	>20	x	Polyps No.		Size		16/09/10; 1
☐ MRI	Spigelman Grade	Est III	Endoscopist	NS	Size of polyps	5-10 mm	x	Location				16/09/10; 1
	Ampulla Size mm		Description									12/06/08; 0
	(Sed) There are multiple polyps (>20 in number) in duodenum. A couple of polyps were 1cm in size and the rest											


Date of birth.
Patient's surname concealed for confidentiality.
Surgeon: John Percy Lockhart-Mummery.
First recorded case in this family. Patient may or may not have had polyposis, but this individual clearly did.
Family number.
First appointment.
Hospital number.
Surgery, underlined in blue ink: '…in 1924 it was very risky to do major surgery because the Rhesus factor hadn't yet been discovered …blood transfusions were risky, bleeding was more common…The idea was to take out as much of the colon as possible because the colon could not be monitored but the rectum could be monitored with the sigmoidoscope… of course the sigmoidoscope would only be 20cm long in those days…if the join was above the 20cm that's where they found that they got their cancers.'
Tenth person diagnosed with polyposis.
Pathology number, in red ink.
Pathology: all denoted by red ink.
Treatment: removal of polyps by diathermy (burning off).
Age at death.
'…the swelling in the groin is obviously where the secondary cancer has spread. Inoperable cancer of the transverse colon, so the transverse colon was never taken out.'
Cause of death.
Survival duration post two surgeries.

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

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

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

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

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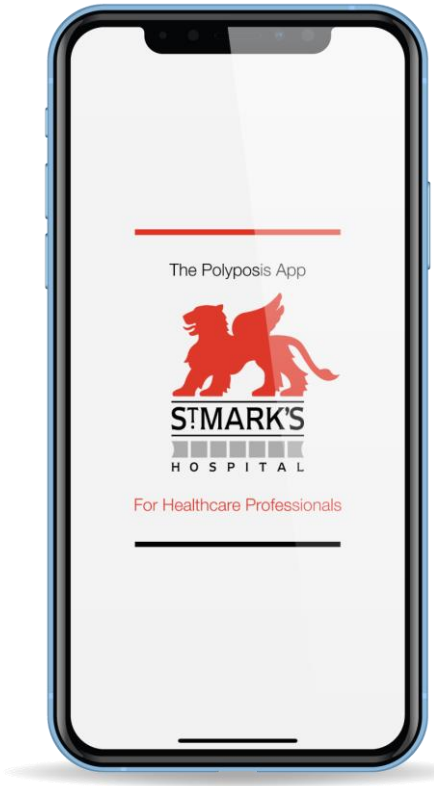
The Polyposis Registry supports people who have a polyposis condition, and their relatives.

Website:polyposisregistry.org.uk

Twitter: @PolyposisRegUK
@VickyChas3

International scientific group: insight-group.org



New version launched March 2019!

Genomics Education Programme

NHS Health Education England

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.

Genomics Education Programme

NHS Health Education England

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.

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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.

Genomics Education Programme

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Summary

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

He summed up by saying 21st 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/

Genomics Education Programme

NHS Health Education England

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 Cheryll Adams	Institute of Health Visiting
Dr Christine Patch	Genomics England
Dany Bell	Macmillan Cancer Support
Dawne Garrett	Royal College of Nursing
Donna Kirwan	NHS England and Improvement
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

Genomics Education Programme

NHS Health Education England

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

Genomics Education Programme

NHS
Health Education England

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