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Genetic counselling in the era of genomics medicine

Overview

Genomics england

- Genetic Counselling –
  Europe and Internationally
- Impact of Genomics -
  100,000 Genomes Project and developments in the NHS
- Genetic Counselling in Genomic Health Care

Genetic counselling is a communication process that deals with the occurrence, or risk of occurrence, of a (possibly) genetic disorder in the family. The process involves an attempt by appropriately trained person(s) to help the individual or the family to

(1)understand the medical facts of the disorder;

(2) appreciate how heredity contributes to the disorder and the risk of recurrence in specified relatives;

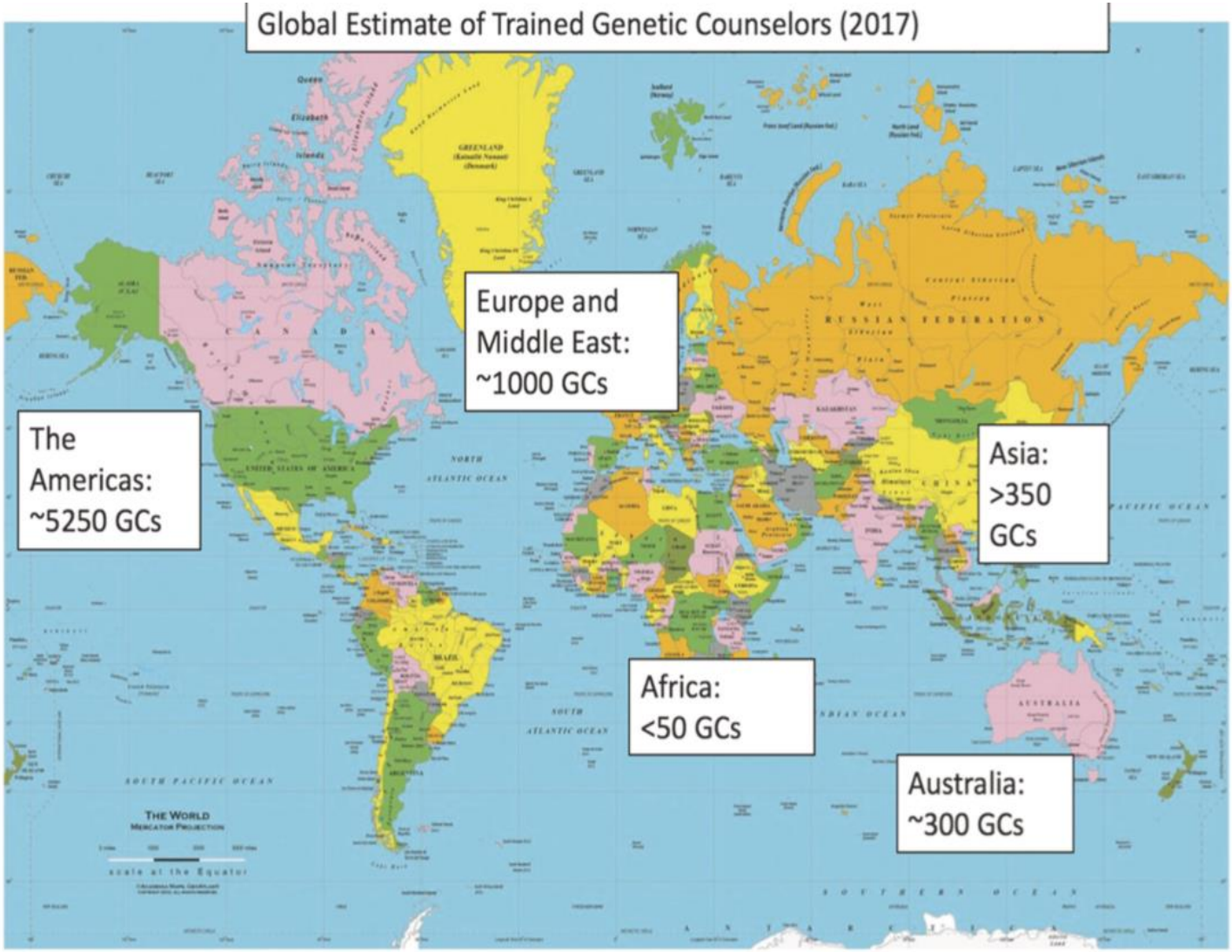
(3) understand the options for dealing with the risk of recurrence;

(4) use this genetic information in a personally meaningful way that promotes health, minimizes psychological distress and increases personal control;

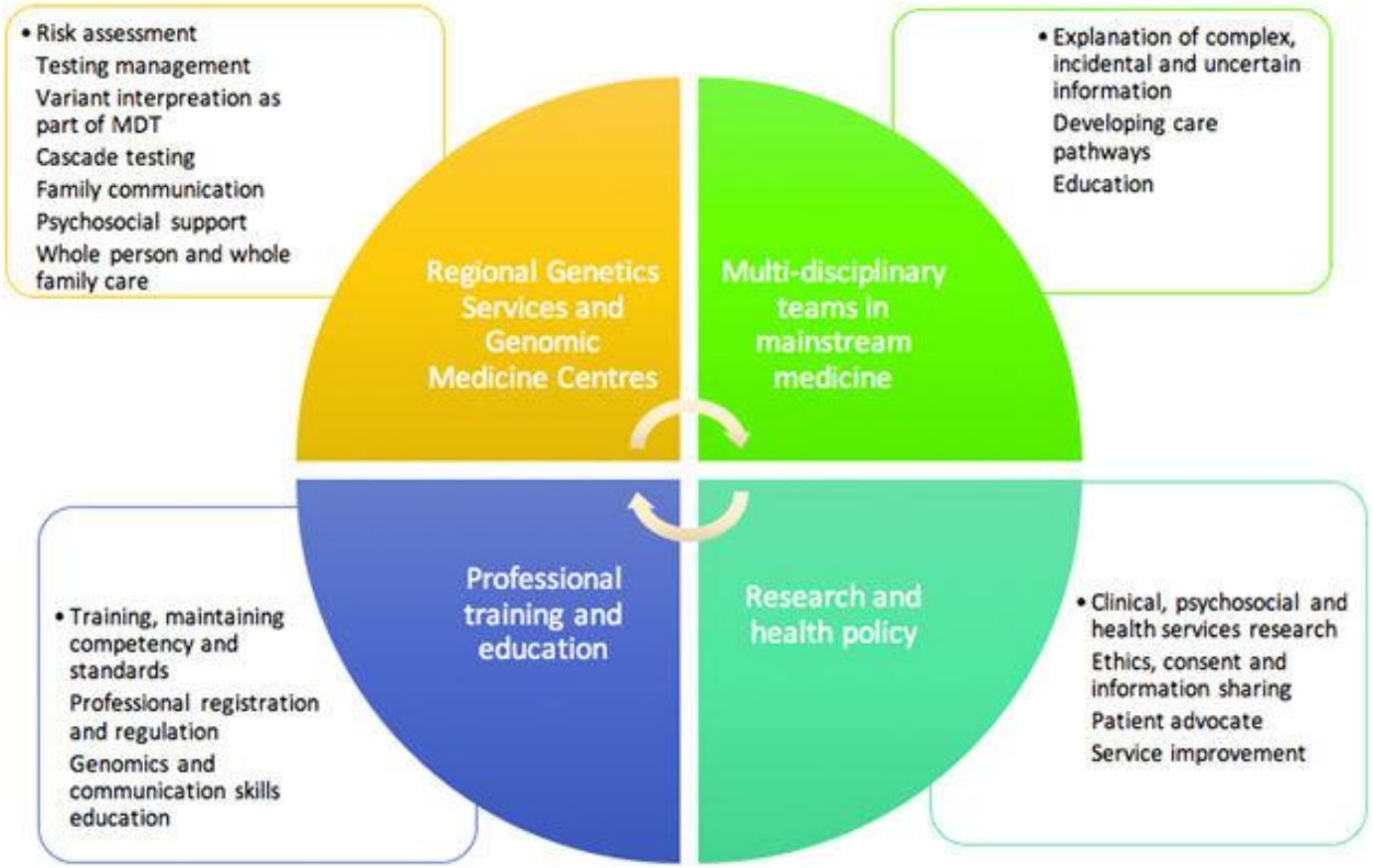
(5) choose the course of action which seems appropriate to them in the view of their risk and their family goals, and act in accordance with that decision;

(6) make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder.

(modified from Frazer FC: Genetic counselling. Am J Hum Genet 1974:26:636-661, Biesecker and Peters:Process Studies in Genetic counseling: peering into the black box. Am J Med Genetics 2001:106:191-198, , Resta, R. G. (2006), Defining and redefining the scope and goals of genetic counseling. Am. J. Med. Genet.)
http://www.eurogentest.org/professionals/info/public/unit3/final_recommendations_genetic_counselling.xhtml



Ormond et al European Journal of Human Genetics (2019) 27:183–197 https://doi.org/10.1038/s41431-018-0252-x



Middleton A, et al . The role of genetic counsellors in genomic healthcare in the United Kingdom: a statement by the Association of Genetic Nurses and Counsellors. European Journal of Human Genetics 2017; 25: 659-661

“We estimate that over 60 million patients will have their genome sequenced in a healthcare context by 2025”

Birney, E., Vamathevan, J., & Goodhand, P. (2017). Genomics in healthcare: GA4GH looks to 2022. doi:10.1101/203554

- More tests
- More and speedier diagnoses
- More choice for patients and their families
- More decisions about management and treatment
- More decisions about research participation

- More standardisation
- More information
- More complexity
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Genetic counseling is a rapidly growing profession with the overarching goal to add value to the care of patients with genetic conditions and their families.

There are many global similarities in the educational process, mechanisms of credentialing, and the scope of practice, but the profession has evolved in unique ways in different countries due to varying healthcare systems, legal restrictions, and cultural issues.

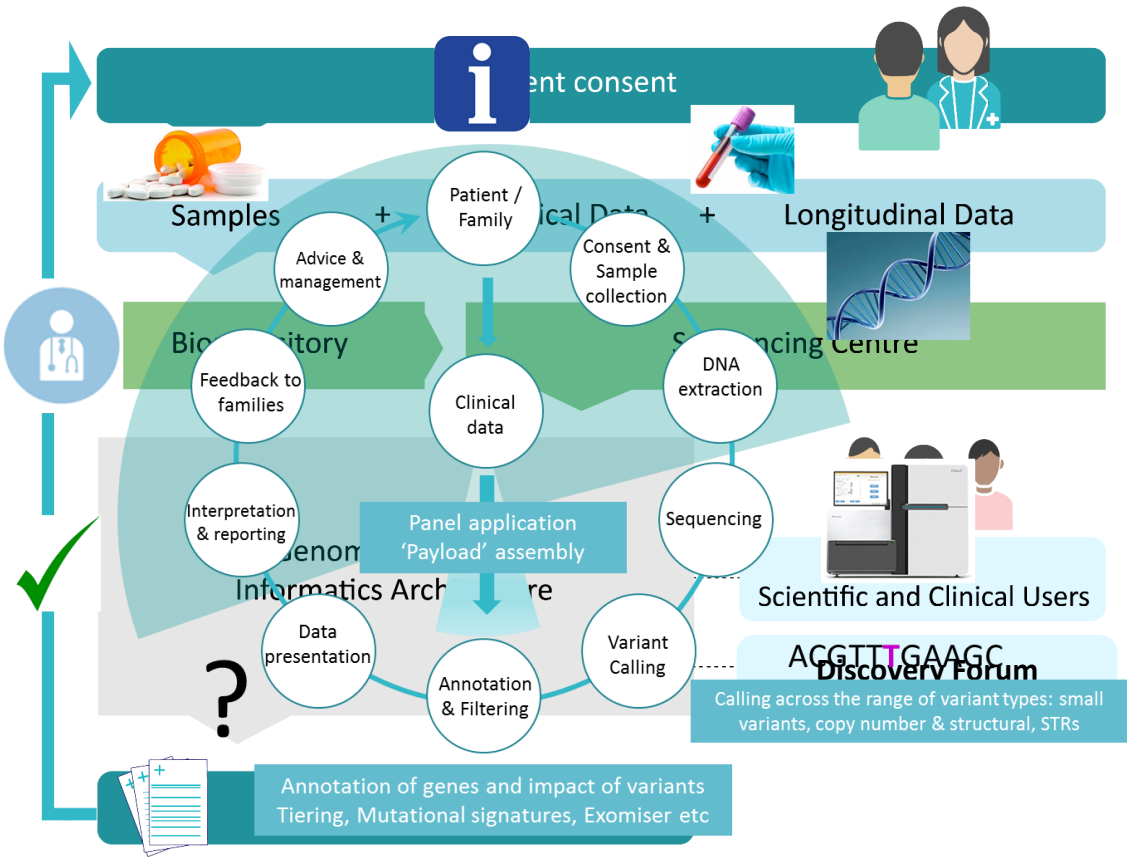
The era of precision medicine is further challenging the way that genetic testing is offered, and the roles that genetic counselors play;

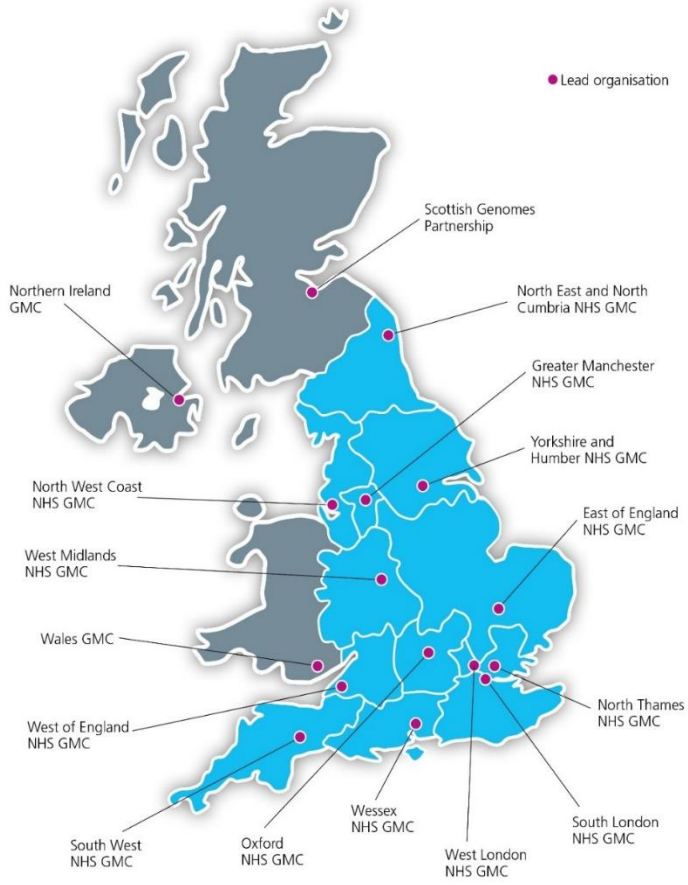
thus far a “one size fits all ” definition of the job title “genetic counselor” does not exist.

Ormond et al Am J MedGenet. 2018;178C:98–107

How the 100,000 Genomes Project worked

- 13 NHS Genomic Medicine Centres covering England, over 90 hospitals
- Responsible for identifying & recruiting participants and for reporting of results
- Northern Ireland, Scotland and Wales have now joined





100,000 Genomes Project Status

Figures as at 06/09/2019

Samples

122,941
Samples collected and received at the UK Biocentre

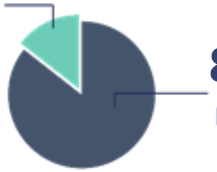
36,782 cancer

86,073
rare disease

Genomes

118,112
Genomes sequenced

32,324 cancer



83,538
rare disease

+2,782
genomes
since last month

Analysis and Results

Analyses for **101,038** genomes sent to NHS GMCs

Equivalent to **29,448** cancer genomes and **71,546** rare disease

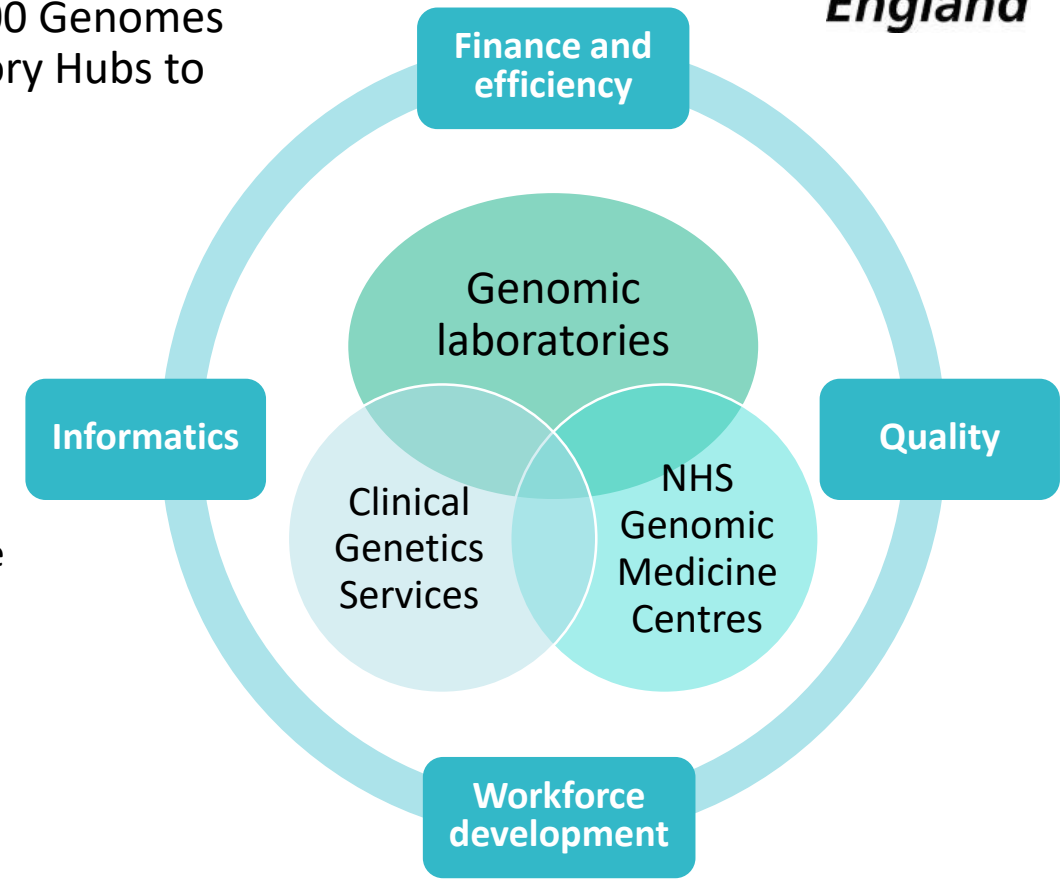
- 20-25% actionable findings for Rare Disease
- ~ 50% cancer cases contain potential for a therapy or a trial in our report

From 100,000 Genomes Project to NHS Genomic Medicine Service

The NHS Genomic Medicine Service brings together existing clinical genetics services with NHS Genomic Medicine Centres (set up for the 100,000 Genomes Project) and the new Genomic Laboratory Hubs to provide seamless delivery of service.

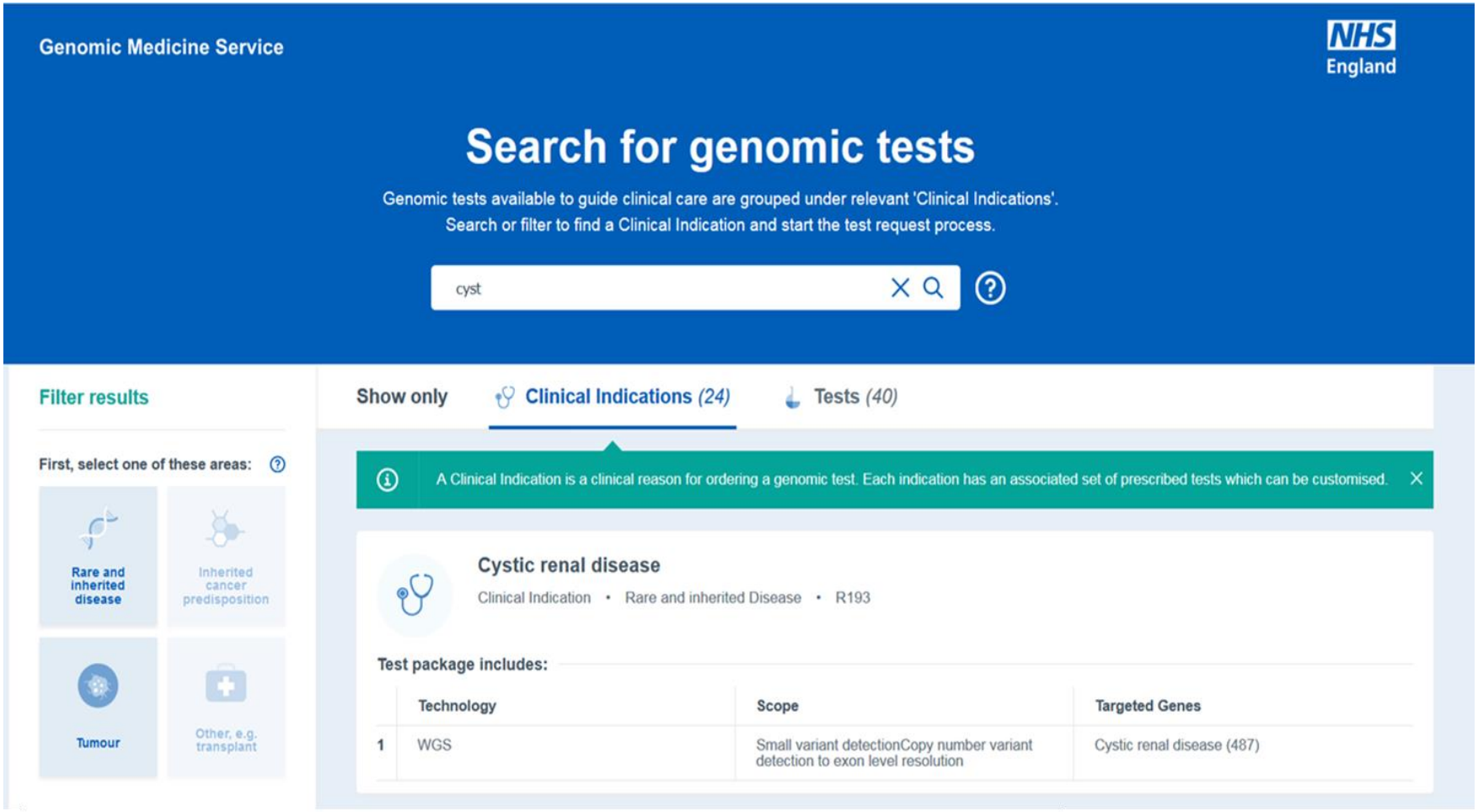
This will mean:

- Ensuring **comprehensive coverage** and access across their geography, including all hospitals, specialist providers and primary care
- Enabling access to an **approved genomic test directory**
- **Integrating clinical genetics service** to provide specialist advice
- **Further mainstreaming** & embedding genomics within other clinical specialities
- **Driving medicines optimisation**/ appropriate prescribing and **personalisation of interventions**

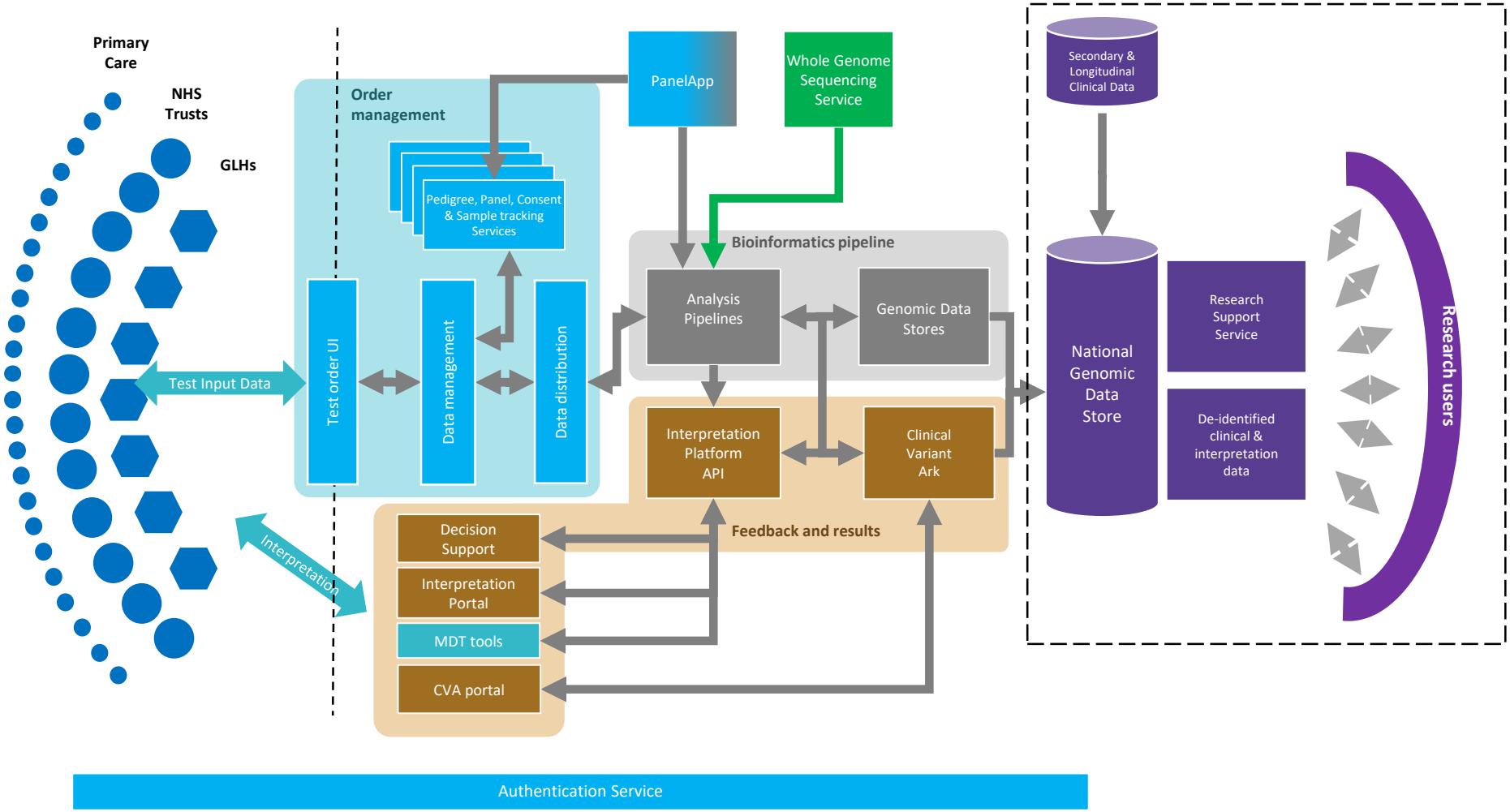


National Genomics Informatics Service

A platform for digital genomic health



NGIS Platform



5 million genomes aspiration announced in October 2018

On the 2nd October, Matt Hancock the Secretary of State for Health and Social Care, announced an ambitious vision for genomic healthcare in the UK…

> Expansion of the 100,000 Genomes Project to **one million whole genomes** sequenced by NHSE and UK Biobank in the next five years

> From 2019, the NHS will offer **whole genome analysis for all seriously ill children with a suspected genetic disorder**, including those with cancer. The NHS will also offer the same for all adults suffering from certain rare diseases or hard to treat cancers

> An **aspiration to sequence 5 million genomes** in the UK within the next five-year years

Proposed Genomics England focus areas to support the 5 million genomes ambition

5 priority areas linked to the priorities identified in the NHS Long Term Plan and Prevention green paper

Inherited and rare disease

- Focus on areas of unmet needs and NHS long term plan priority areas
- Rare Mendelian and non-Mendelian disorders

Cancer and clinical trials

- Provide detailed molecular stratification and drive clinical trials in the UK
- Support research into new cancer biomarkers

Newborns and population genomics

- Prospective cohort to assess benefits of WGS for newborn screening
- Expand coverage of under-represented ethic groups

Pharmacogenomics

- Expand knowledge of gene-drug interactions to improve safety, efficiency and effectiveness of prescribing

New technologies and analytics

- Advanced analytics, artificial intelligence, multi-omics and therapeutic innovation

Towards 5M

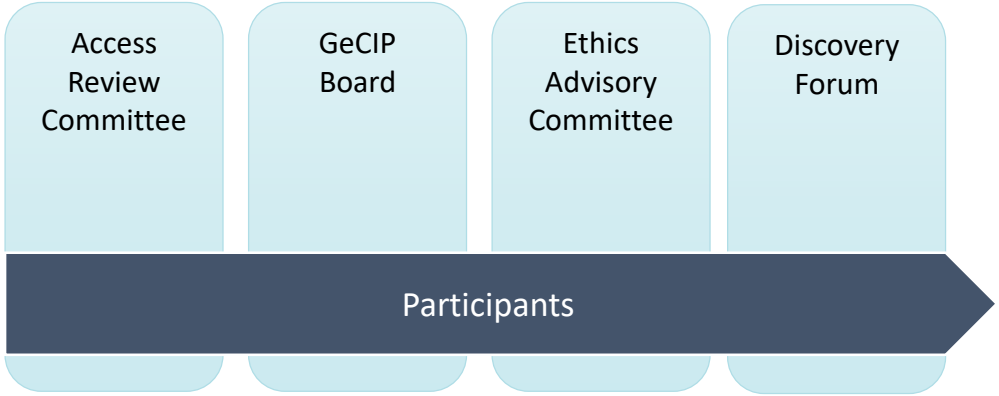
NHS Genomic Medicine Service

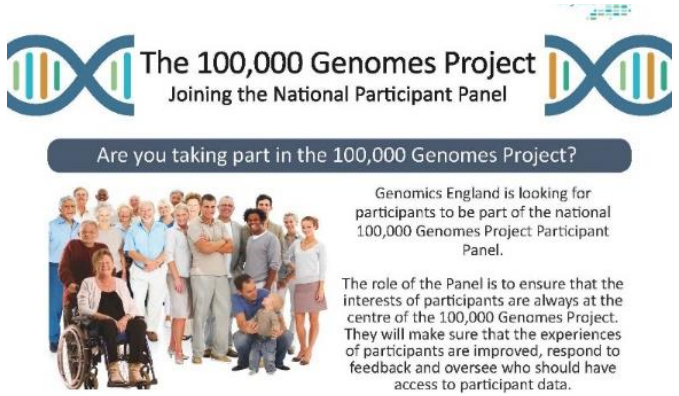
- Support the transformation of UK healthcare and enable the move to a prevention focussed model
- Provide evidence base and support to expand the use of genomics within the health service e.g. through the annual genomic test directory reviews

Patient involvement - the National Participant Panel

Role of the Panel is to ensure the interests of participants are always at the centre of the 100,000 Genomes Project. They do this by:

- Making sure experiences of participants are at the heart of the project
- Responding to feedback
- Overseeing who should have access to participant data





https://www.geneticalliance.org.uk/news-event/participants-experiences-of-the-100000-genomes-project/

The Genomic Medicine Service promises a cutting edge approach to diagnosis, so it is appropriate to sound a note of caution about the post-diagnostic service available to patients, and its potential impact on patient satisfaction with health services.

The NHS is resource-constrained.

Results from WGS might not be clinically significant at the time they are reported back to patients, but could become actionable in the future

Research will be needed to deliver a meaningful finding.

There must be a clear message to patients and carers as to what to expect, and who has responsibility for renewing contact when appropriate.

It is clear that whether the Genomic Medicine Service ultimately delivers on its promise will depend on factors outside pure progressing genomic technology.

The Clinical Utility of Genomic Sequencing for Patients and Families:
Improving the Communication of Genomic Diagnosis Results Using Experience Based Co-Design (EBCD)

Professor Alison Metcalfe (PI, Sheffield University); Dr Christine Patch; Prof Glenn Robert (both King's College London); Prof. Jill Clayton-Smith (Manchester University Hospital); Ms Elizabeth Alexander (Manchester University Hospital); Dr Milan Macek (RD Connect; Charles University, Prague); Dr Vera Frankova (Charles University, Prague)); Dr Radka Pourova (Charles University, Prague); Dr Alessia Costa (King's College, London); Ms Virginie Bros-Facer (Eurordis); Dr Amy Hunter (Genetic Alliance UK) ; MsLauren Roberts (SWAN; Genetic Alliance UK)

Preliminary findings from focus groups

- Pre-test preparation: better expectation management and clear information about path ahead
- Support and advice after results are shared (e.g. follow up appointments, support groups)
- Named point of contact (e.g. GC; specialist nurse)
- Communication about results availability
- Better coordination between clinical geneticists and other medical specialists

SolveRD
Solving the Unsolved Rare Diseases

OUTLOOK · 30 MAY 2018

Living under the shadow of Huntington's disease

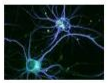
With a family history of the condition, Mark Newnham makes a tough decision to find out what his future could hold.

Simon Roach

PDF version

RELATED ARTICLES

Piecing together the puzzle of Huntington's disease



How the gene behind Huntington's disease could be neutralized

- Despite the prospect of a life without children, Newnham does not regret his decision to get tested. At least, he explains, he is moving forward with his eyes open. And advances in research fill him with “immense hope” that some form of treatment will be available in his lifetime
- https://www.theguardian.com/news/audio/2019/apr/22/hope-for-those-with-huntingtons-podcast

Huntington's Disease Association

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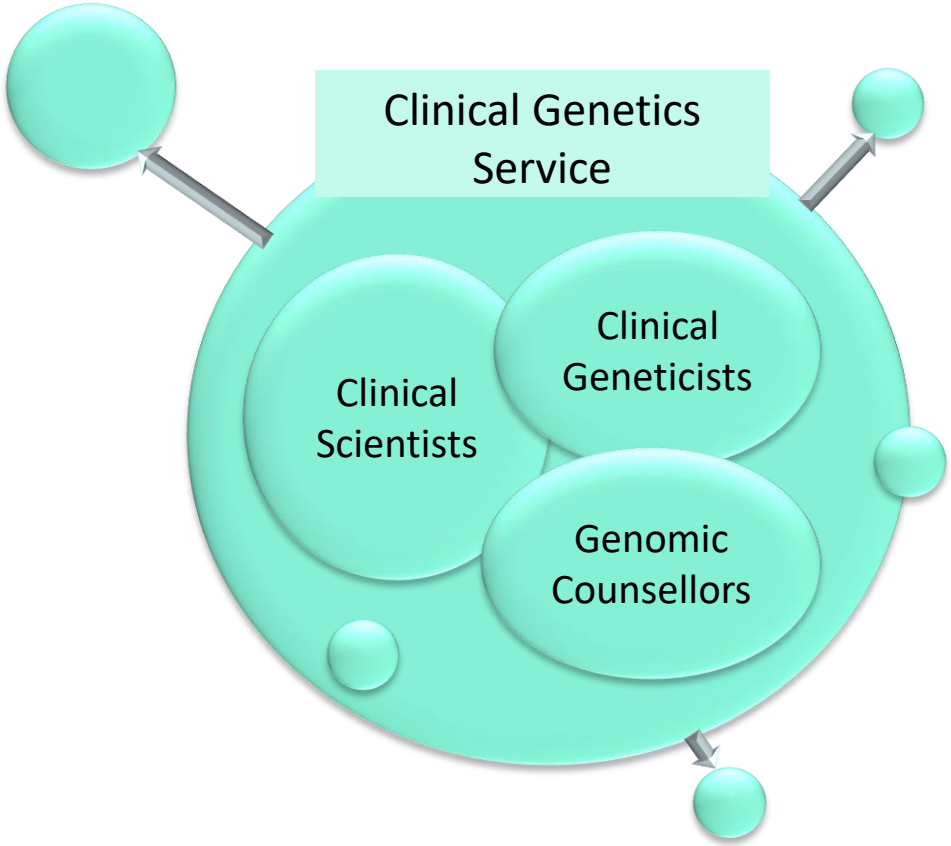
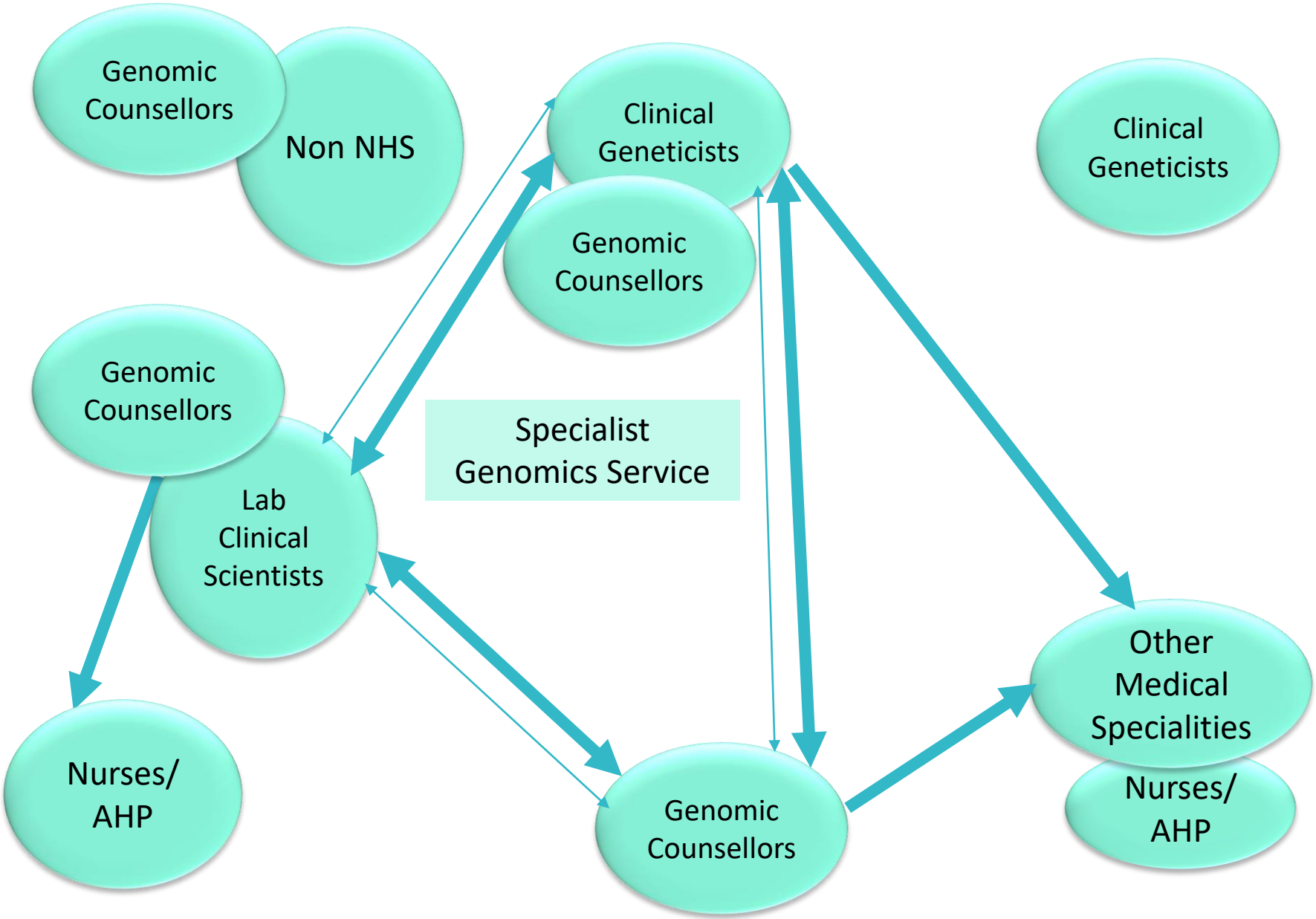
Not just genetic counsellors

Health Education Genomics Education Programme
https://www.genomicseducation.hee.nhs.uk/genomics-in-healthcare

Future Service delivery models?

- Historically tightly linked clinical geneticist, Clinical scientists, genetic (genomic) counsellors





12 December 2019

Genetic counsellors are involved at all stages in the pathway of the patient journey through genomic medicine.

At the beginning, with genetic risk estimation, decision making about testing and conversations about consent;

In the management and interpretation of results;

At the point at which the result enters into the health care system and is returned to the clinician and the patient,

At the follow up of patients in mainstream and in helping the patient to seek out and communicate the results within their family.

Genetic counsellors are also involved in supporting the patient to adapt to the genetic information and manage the psychosocial consequences of this as it is relevant to the wider family and future generations.

Patch and Middleton British Medical Bulletin, 2018, 1–10 doi: 10.1093/bmb/ldy008

Genomics england

Thank you to everyone who has taken part in the 100,000 Genomes Project

NHS

ESHG
EUROPEAN SOCIETY OF HUMAN GENETICS

WELLCOME GENOME CAMPUS