Genetic Nurse to Genetic Counsellor to Genomics: Reflection on 30 years

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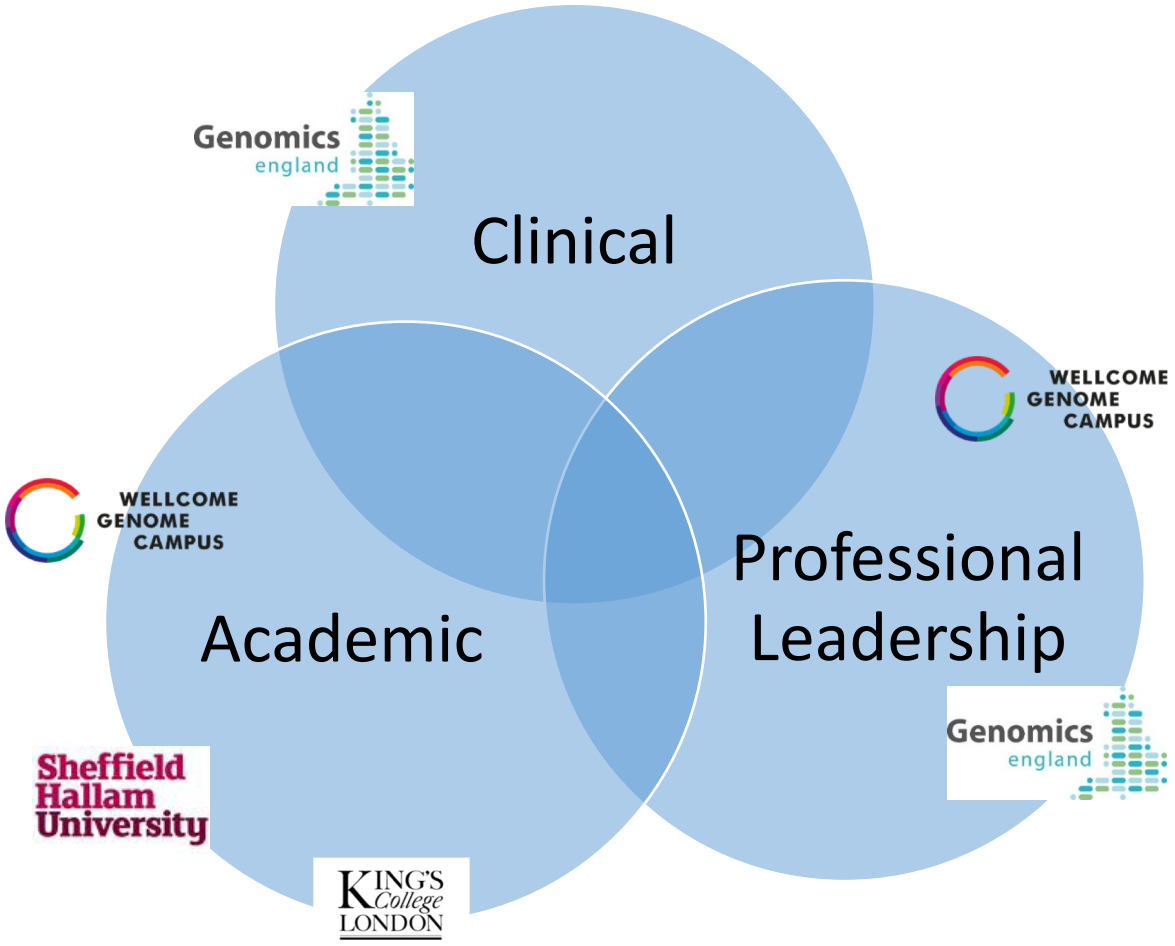
Visiting Professor Faculty of Health and Wellbeing, Sheffield Hallam University
Reader in Genomic Health Care KCL London

Genetic counselling: the early days

- Nurses (like me) were given jobs in genetics

- In the beginning was a nurse,
  - with a psychology degree (BSc),
  - experience as a volunteer for a suicide crisis line,
- two small children
- no plan: just needed a job.
- Choice between a permanent job as a night sister in a private hospital or a part time post as a genetic nurse in a genetics department.



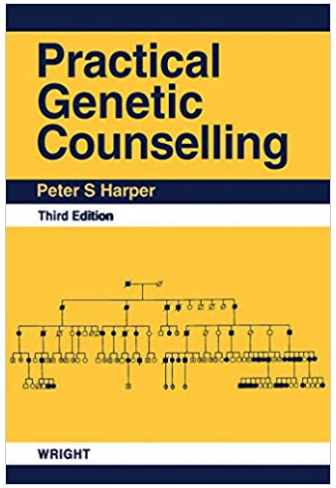
30 years later
three paid jobs,
one honorary position

Jack of all trades:
Master of some

….. life was simple

- You needed only a few books
- Unless you were being clever you worked out dominant, recessive or X linked and divided by 2 and then multiplied by …
- If you were clever you did a Bayes calculation
- There were only a few patients
- There were no genetic tests apart from karyotypes and linkage anyway
- Cancer genetics hadn’t been invented

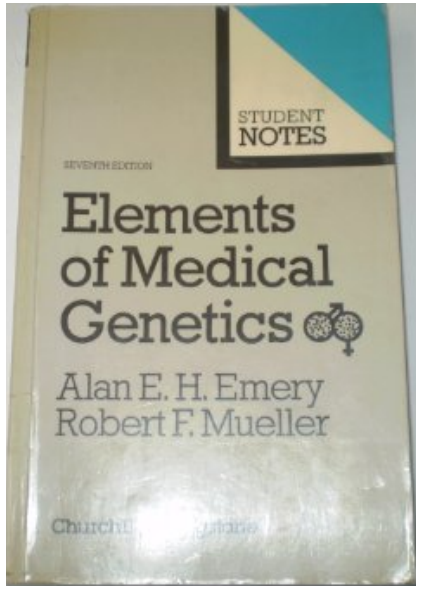
My Training



1966

1998

1989

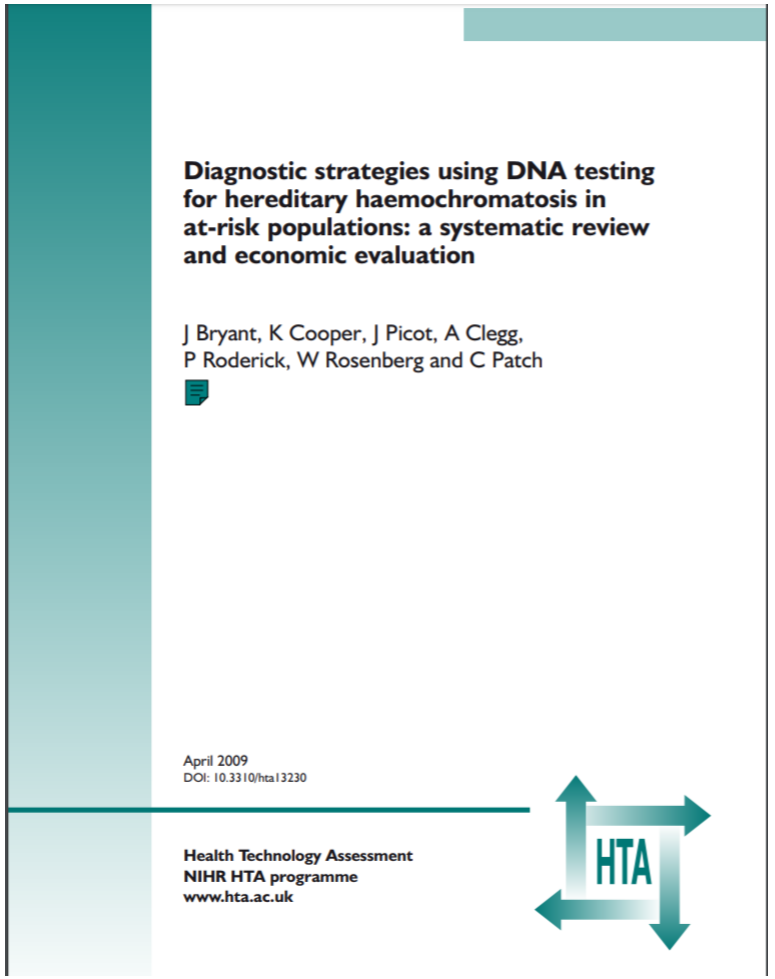


PhD – Public Health

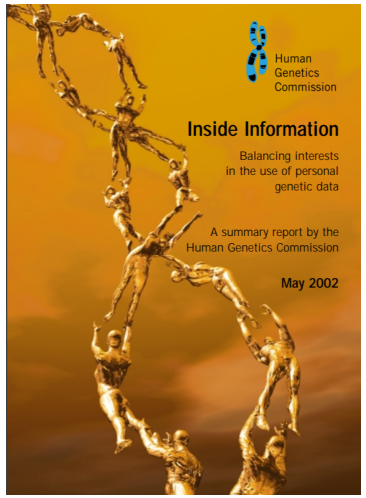
Patch C , Roderick P, Rosenberg W

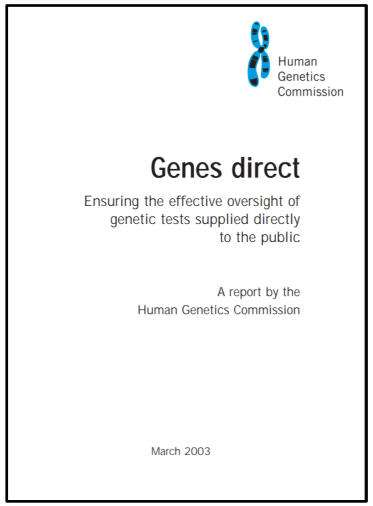
Factors affecting the uptake of screening: A randomised controlled non-inferiority trial comparing a genotypic and a phenotypic strategy for screening for haemochromatosis

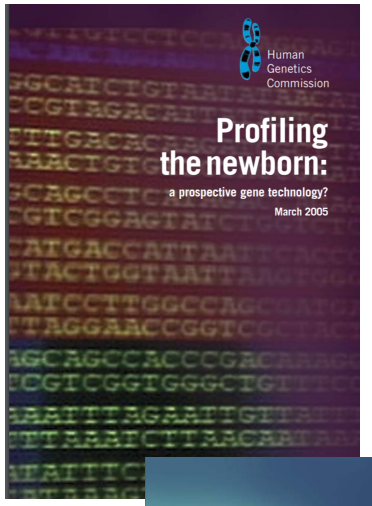
https://doi.org/10.1016/j.jhep.2005.02.018

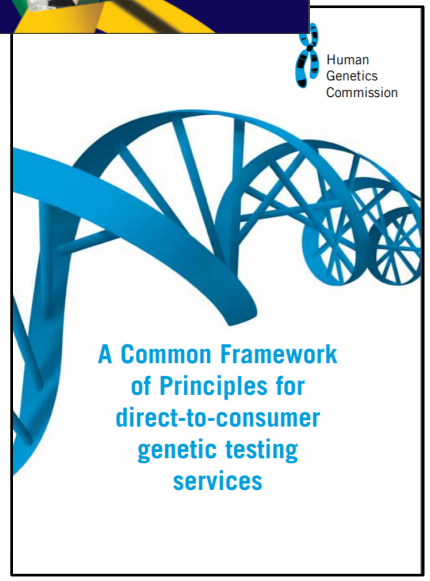


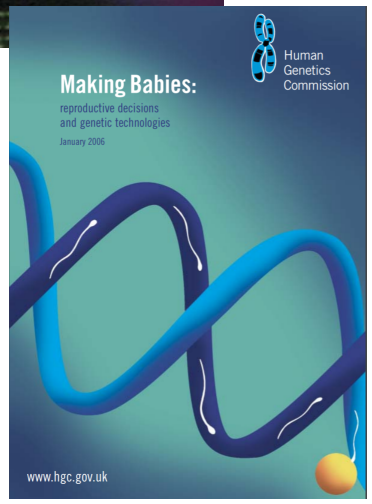
UK Human Genetics Commission 2003 - 2008











Baroness Helena Kennedy
Chair, Human Genetics Commission

Executive

Board

Professional and Public Policy Committee

Genetic Nurses and Counsellors

Genetic Counselling course - Bertinoro, Bologna, Oman

Journey towards professional registration UK and Europe

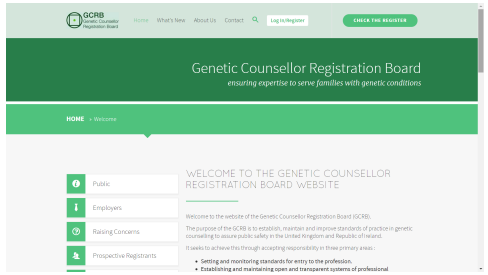
1980's Association of Genetic Nurses and Counsellors

Master programmes in Genetic Counselling

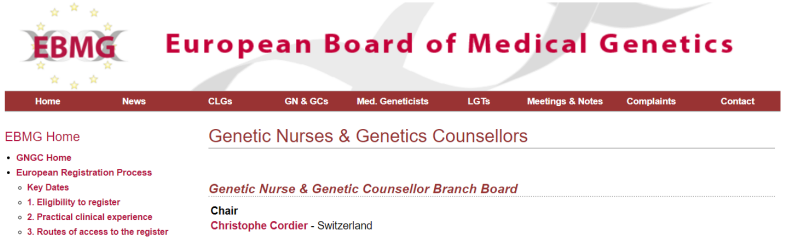
- 1992 Manchester, 2000 Cardiff

2000 Genetic Counsellor Registration Board

- National Register
- Graduate nurses or midwives or graduates of Master's degree in genetic counselling
- plus 2 years of full time supervised experience in a genetic centre
- Demonstration of achievement of competencies



2015 European Board of Medical Genetics - Genetic Nurses & Genetic Counsellors branch



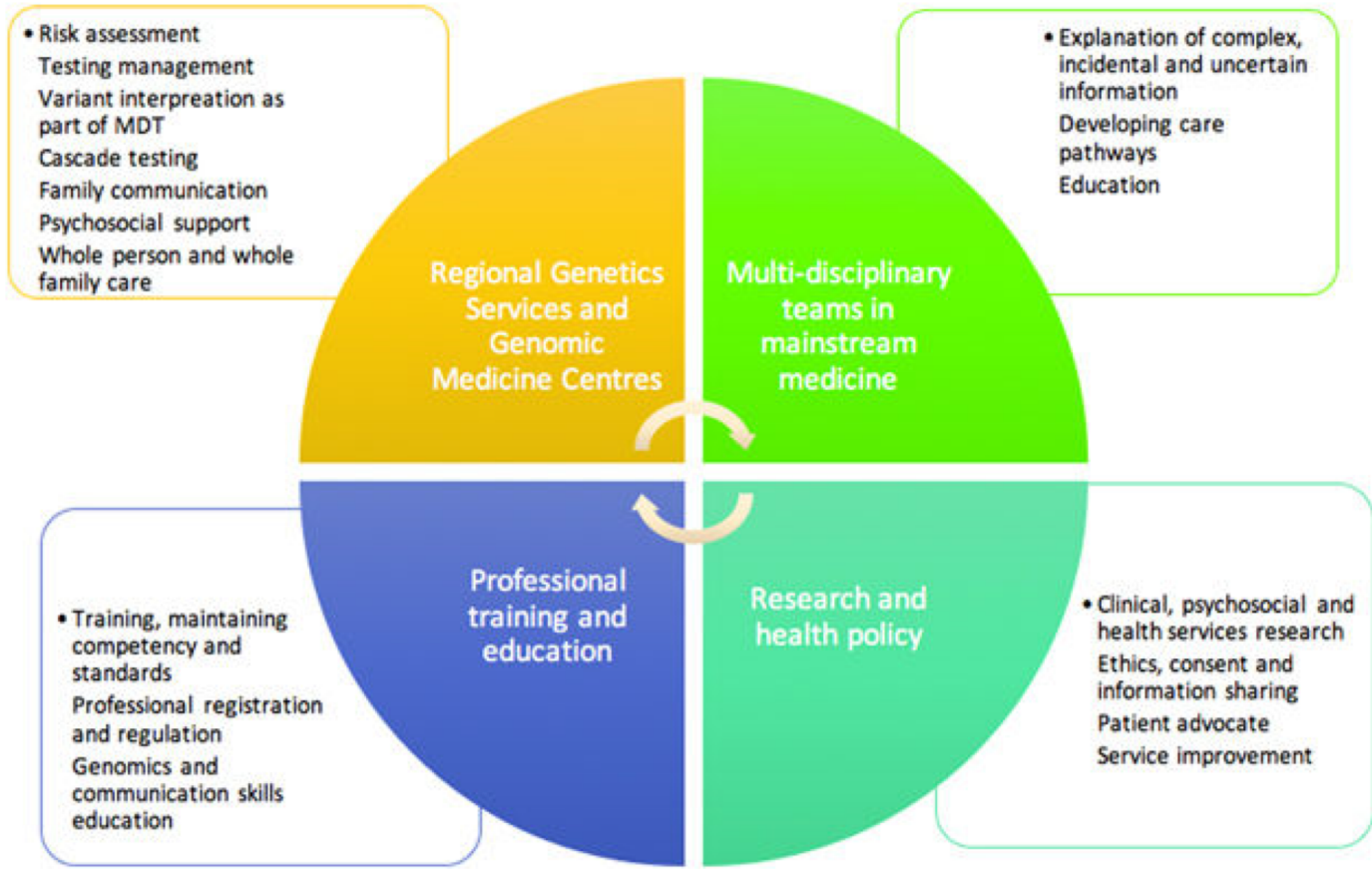
Genetic counseling is a rapidly growing profession with the overarching goal to add value to the care of patients with genetic conditions and their families.

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



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

Point of View: An evolution from genetic counselling to genomic counselling.

Patch C, Middleton A.

Eur J Med Genet. 2019 Apr 13. pii: S1769-7212(19)30251-4. doi: 10.1016/j.ejmg.2019.04.010

Genetic counselling in the era of genomic medicine.

Patch C, Middleton A.

Br Med Bull. 2018 Jun 1;126(1):27-36. doi: 10.1093/bmb/ldy008

Genomics england

The 100,000 Genomes Project
Transforming Healthcare

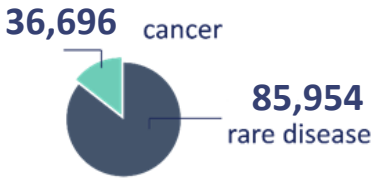
Global Alliance for Genomics & Health

Collaborate. Innovate. Accelerate.

100,000 Genomes Project Progress

Samples

122,650
Samples collected and received at the UK Biocentre



Genomes

112,198
Genomes sequenced

+3,751 genomes since last month

Analysis and Results

Results for **86,944** genomes sent to NHS GMCs

Equivalent to **21,551** cancer genomes and **65,393** rare disease

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

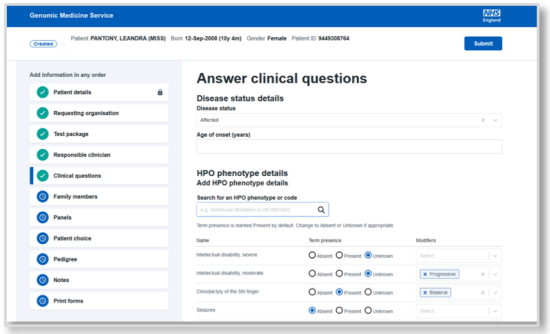
Figures as at 01/06/2019

National Genomics Informatics Service (NGIS) benefits

Shared platform for diagnostics and research

- Provides a framework to identify and recruit cohorts and trial-eligible patients
- Ongoing record of research and diagnostic genomic analyses of each individual
- Longitudinal life course data from national data sources held alongside data provided by researchers, clinicians & patients
- Integration with external systems including hospital LIMS via APIs and standardized message formats e.g. FHIR

- Readily configurable clinical data capture for rapid adaptation for new cohorts or new data
- Genomic data generated initially for research can be used for clinical care, where indicated
- Creates a shared national genomic knowledgebase where new data from research can be rapidly translated into the clinic



Genomics england

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

NHS

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

Not just genetic counsellors

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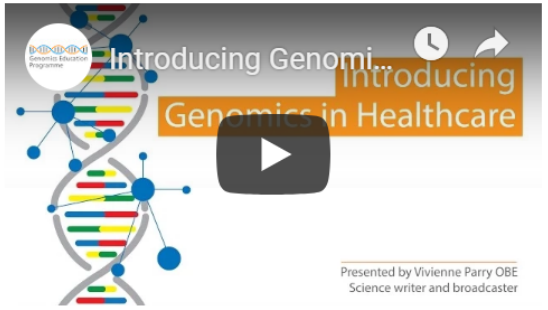
Nursing in the Genomic Era

Moving into the Mainstream

Introducing Genomics in Healthcare



Rare Disease: A Family's Journey

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Written on 21 Mar 2019

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Written on 06 Mar 2019

G2NA an international Global Genomics Nursing Alliance (founded 2017)

The G2NA vision is to serve as the unified international voice for advancing and integrating genomics into nursing practice.

The G2NA mission is in supporting nurses to realise their full potential through integrating genomics across nursing practice to improve healthcare for all.

Co-leads include Kathleen Calzone NIH/NCI

www.G2NA.org

WELLCOME GENOME CAMPUS

NEW

Nursing, Genomics and Healthcare
27 - 29 April 2020
Opens soon – Email for updates

- What is the impact of genetic information
  - Individuals, families, health systems, society
- How do we support individuals and families

With thanks to Prof Alison Metcalfe

"… I think the parents have to really really understand a lot of psychology. Its not just the physical aspects of feeding and going through a major operation…. All that is secondary I feel to how a mother has to understand and its a lot of psychological understanding cos if its not there, the the children are damaged…

Mum of a boy affected by DMD, with one daughter.

Parents and children's experiences revealed:

- Parents encouraged and advised to talk to children
- Lack or limited support from genetic counsellors (and other HPs) about living with a genetic condition.
- Parents only given information when they were young – did not realise the implications better until they had their own children
- Children unable to access or talk to health professionals
- Secrecy was detrimental to parents' and children's mental health and well-being
- The importance of using appropriate terminology

- Genetic Counsellors and other Health professionals involved in exploring applying and testing the socio-psychological theories that might inform how we use genomics in society to maximise benefit and minimise potential harms.

Black and Minority Ethnic women's decision-making for risk reduction strategies after BRCA testing: Use of context and knowledge.

Machirori M, Patch C, Metcalfe A.

Eur J Med Genet. 2018 Dec 12. pii: S1769-7212(18)30405-1. doi: 10.1016/j.ejmg.2018.12.006. [Epub ahead of print]

Study of the relationship between Black men, culture and prostate cancer beliefs

Machirori M, Patch C, Metcalfe A.

Cogent Medicine 2018, 5: 1442636 https://doi.org/10.1080/2331205X.2018.1442636 A

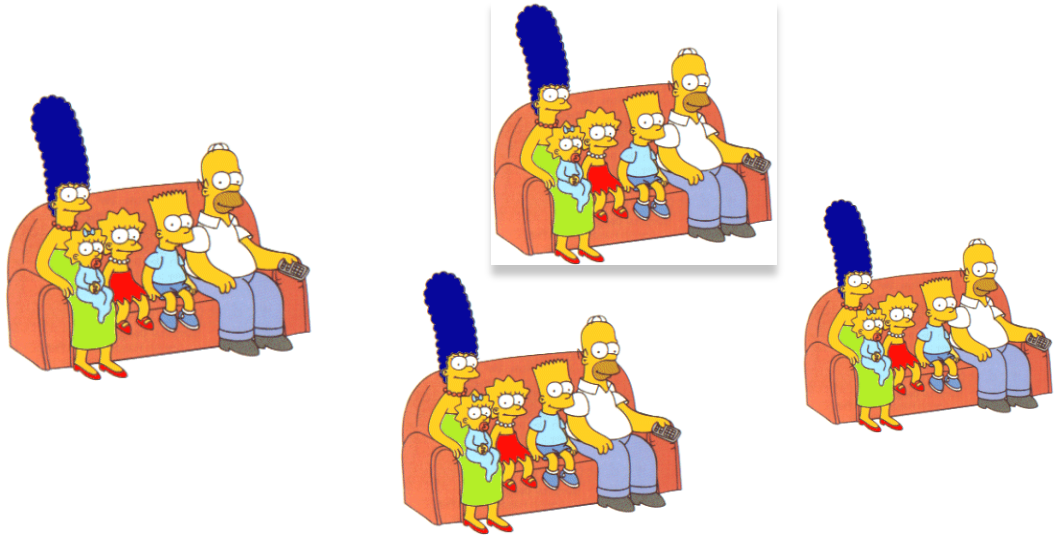
Living a normal life in an extraordinary way: A systematic review investigating experiences of families of young people's transition into adulthood when affected by a genetic and chronic childhood condition.

Waldboth V, Patch C, Mahrer-Imhof R, Metcalfe A.

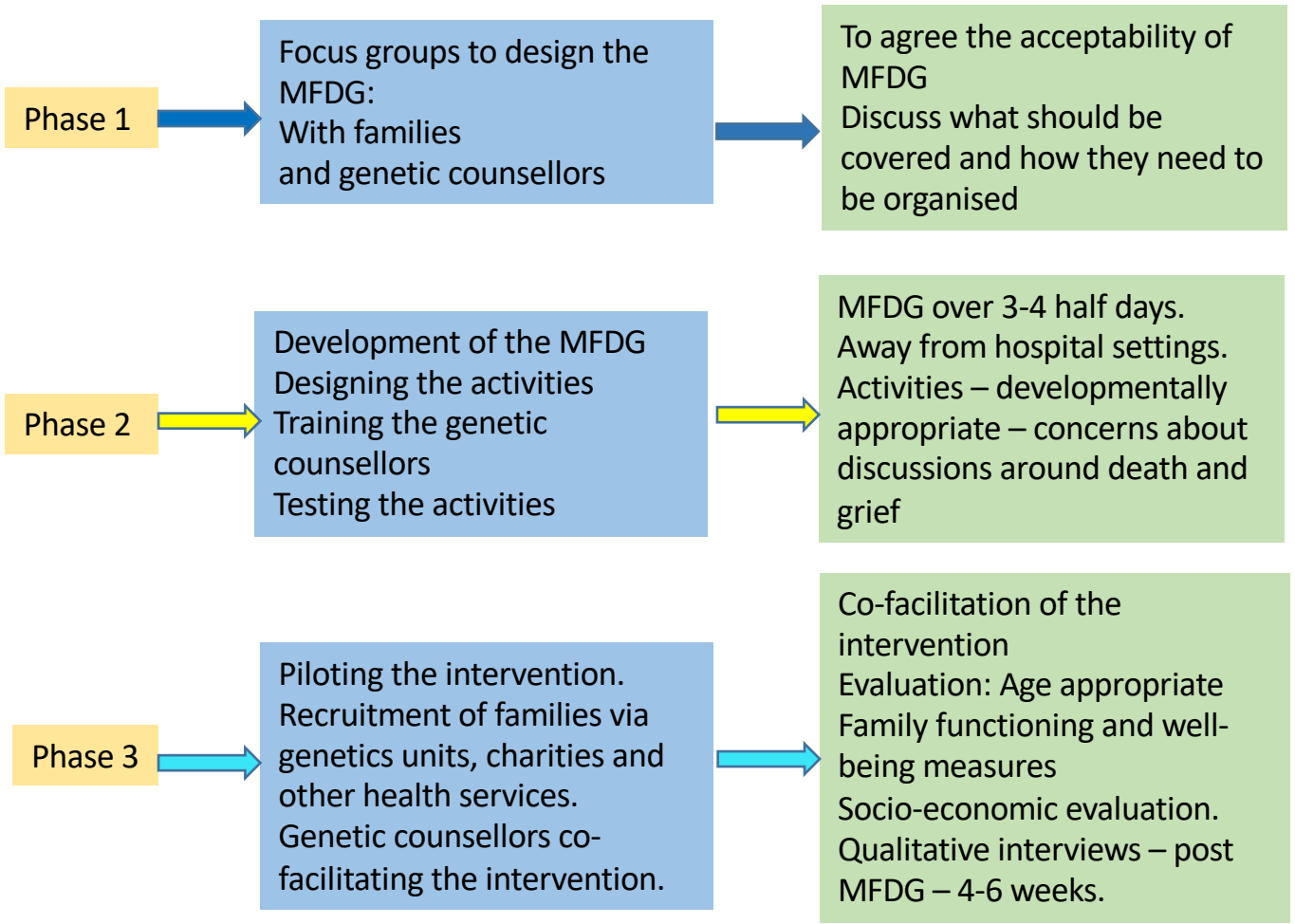
Int J Nurs Stud. 2016 Oct;62:44-59. doi: 10.1016/j.ijnurstu.2016.07.007. Epub 2016 Jul 12. Review.

Co-design of family centered interventions

Introducing Multi-Family Discussion Groups (MFDG)



Overview of the MFDG intervention's design



Findings from the Focus Groups

2 key themes emerged showing

i) the need for the intervention

ii) the intervention's design:

- Parents and their children perceived a lack of guidance and support from health professionals.
- Low levels of communication left families feeling isolated and vulnerable.
- Parents anxious about finding the right words and being able to deal with their own and children's emotions.
- Children and young people reported feeling anxious about initiating conversations.
- MFDG seen as exciting and valuable opportunity for families but concern about when and where they were run – fear of stigma.
- Genetic counsellors were surprised by the families' strength of feeling.

- Therapeutic value of focus groups!

A picture showing the child's family before and after the intervention. In the before picture, the child has written by an image of his home ‘we are so sad' and in the after picture the child has written ‘we are so happy'.

A picture showing a family before and after attending the intervention. In the before image the child is sad and confused asking their parent “why can't you tell me?” In the after picture the child is happier because he understands what is happening in his family because his parents have been ‘able to tell his son a bit more about the genetic disease'.

Eisler I et al. Developing an intervention to facilitate family communication about inherited genetic conditions, and training genetic counsellors in its delivery. Eur J Hum Genet 2016; 24: 794-802

Discussion and Conclusion

- MFDG - highly beneficial from a qualitative perspective.
- Can MFDG for genetic conditions be introduced into health services for families?
- Issues about where this fits – should genetic counsellors being undertaking this type of activity?
- Could other health professionals/support groups be involved
- Can we continue to leave families ‘isolated’ when there are options to facilitate better coping and adaptation to living with the genetic condition?
- The need for a full evaluation.

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)

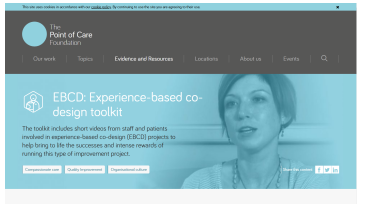
Aim: To facilitate the development of patient and family-centred interventions or services co-designed with staff for the sharing of genomic sequencing information in European Reference Networks (ERNs).

Objectives:

• Train and support researchers to work with patients, families and healthcare staff (Manchester; Prague) to deliver the EBCD method to co-design interventions or services to support the sharing of genomic sequencing information.

• Perform a process evaluation of the EBCD process and the implementation of the intervention or service using interviews and surveys (translated into member countries languages) in the initial two participating ERNs.

• Publish the EBCD method used within the initial two ERNs in order to provide the opportunity for the other 22 ERNs to adopt the findings or follow a similar process to design their own intervention or services.



How do patients and their families respond to being offered genome sequencing?

With thanks to Celine Lewis, Saskia Sanderson, Lyn Chitty and all of the project team

C22.1 To find out if it's genetic or not. Motivations concerns and perceived impact of Genomic sequencing among young people

North East Thames Regional Genetics Service

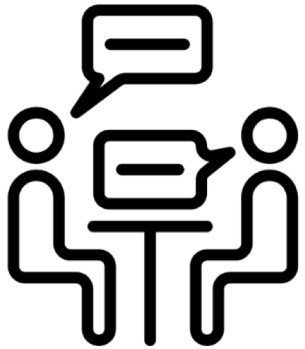
FUNDED BY NIHR | National Institute for Health Research

Great Ormond Street Hospital for Children NHS Foundation Trust

Background

- Aim: Gather **empirical evidence** on patients' **decision-making** including **knowledge, attitudes and experience** of genome sequencing in the 100,000 Genomes Project

- Sub-studies:
  1. Survey of >500 participants in the 100,000 Genomes Project
  2. Interviews with 20 parents taking part in the Project
  3. Reasons given for declining to take part in the Project
  4. Observations of 20 consent appointments
  5. Interviews with 20 health professionals referring or consenting patients into the Project



SUMMARY OF SURVEY FINDINGS

Taking part: Most participants (97%) felt this had been an informed decision

Secondary findings: Most (88%) decided to receive secondary findings, felt this was an informed decision

Motivations: Participants were motivated to take part by desire both for diagnosis and to help others (desire to help others > desire for diagnosis)

Concerns: Concerns were low (concern about potential psychological impact of high risk results > concerns about data sharing/access)

Understanding: Understanding generally high (lower about limitations/uncertainties, risks, and commercial companies having data access)

Limitations : few questions about secondary findings, lack of decliners;
shortcomings partially addressed in our qualitative studies

ARTICLE | Genetics inMedicine

Open

Opening the "black box" of informed consent appointments for genome sequencing: a multisite observational study

Saskia C. Sanderson, PhD[1,2], Celine Lewis, PhD[1,2], Christine Patch, PhD[3,4], Melissa Hill, PhD[1,2], Maria Bitner-Glindzicz, MBBS, PhD[1,2] and Lyn S. Chitty, PhD MRCOG[1,2]

IMPLICATIONS OF OBSERVATION STUDY FINDINGS FOR GENOME SEQUENCING POLICY

1. Strong evidence of **biomedical information** giving, but, little in terms of actively engaging participant in discussion and **psychological support**

2. Discussion about SFs brief and **little focus on limitations and uncertainties**

3. Importance of (online?) professional training to support genome sequencing consent processes, particularly **communication** and **listening techniques**, **eliciting participants views**, **concerns, questions** etc

4. Particularly important if **non-genetic specialists** are offering genome sequencing in new Genomic Medicine Service who may not have had ‘genetic counselling’ training

European Journal of Medical Genetics
Volume 62, Issue 5, May 2019, Page 287

Editorial

World congress on genetic counselling

Anna Middleton, Christine Patch, Barbara Biesecker

Show more

https://doi.org/10.1016/j.ejmg.2019.04.011

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Evidence-Based Genetic Counselling

Edited by Anna Middleton, Barbara Biesecker, Christine Patch

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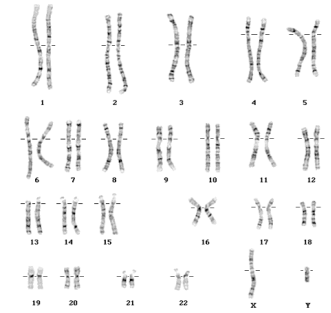
World Congress on Genetic Counselling

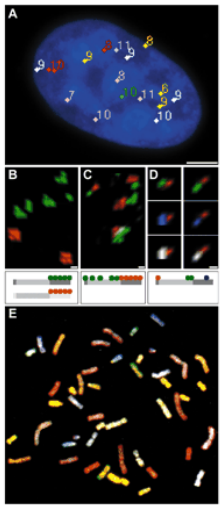
02 - 04 October 2019

Wellcome Genome Campus, UK

Genetic technologies evolution

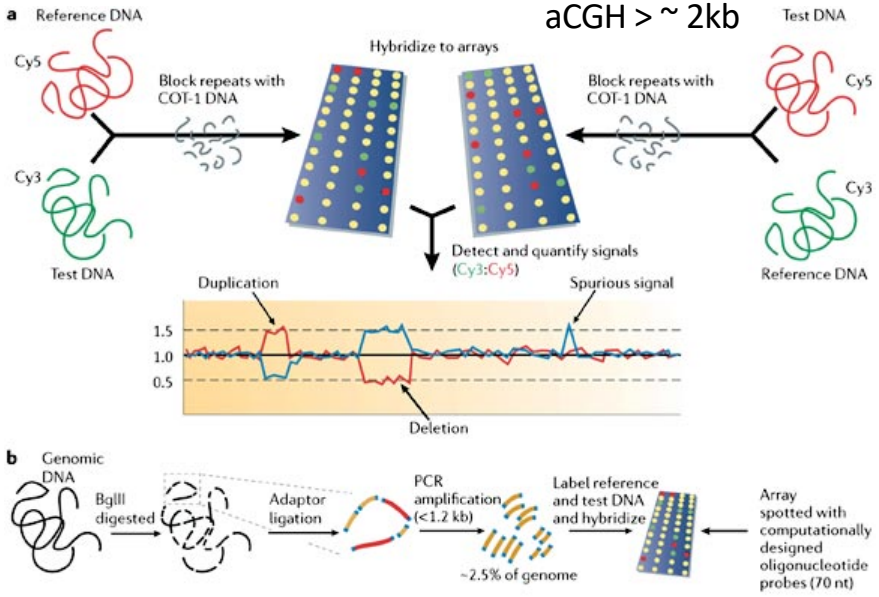
Karyotype >3-5Mb



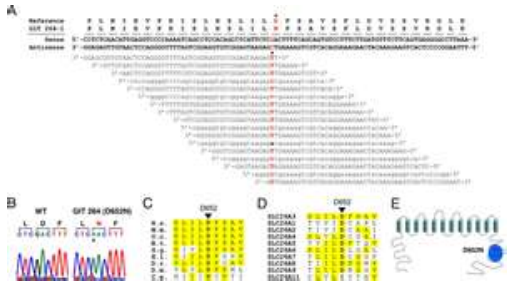


Fluorescent in situ hybridisation

aCGH > ~ 2kb



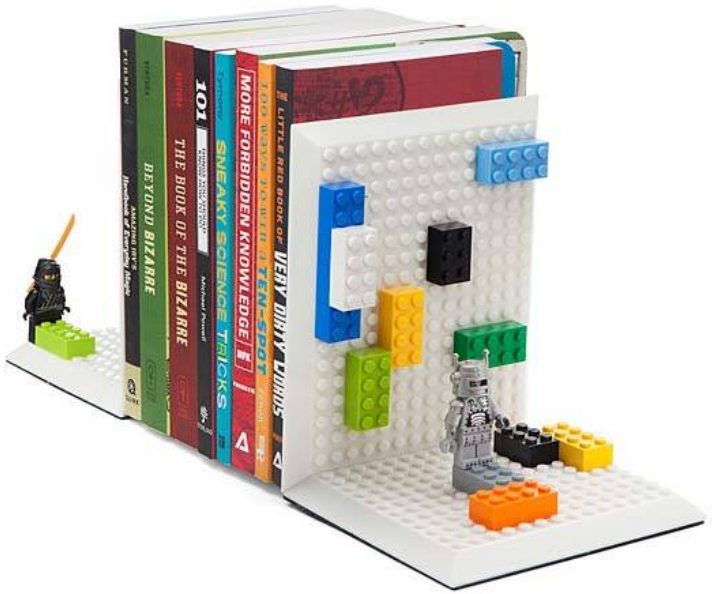
Sequencing



1bp

Abundant class of human DNA polymorphisms which can be typed using the polymerase chain reaction.

JL Weber, PE May - American journal of human genetics, 1989



Genome Sequencing

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

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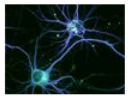
Piecing together the puzzle of Huntington's disease



How the gene behind Huntington's disease could be neutralized

Huntington's Disease Association

given his and his partner's desire to have children, Newnham concluded that he needed to know his status with respect to Huntington's disease. "We didn't want to have a child without that certainty," he says. The result was not what he had hoped for. Like his father, he carries the mutated gene.

He still works as an actor and musician, but says that his priorities have changed. "The test results made me realize that what really drove me as a person before, and what ambitions I had, they're not as important now," he says.

The dream of performing at Glastonbury will never be gone, but spending time with family and friends seems more important. This shift in perspective has given him a quiet contentment, he adds.

Nature Outlook 10th May 2018

- 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

- Huge expansion of genomic information
- Need to consider and influence policy, practice and delivery of genomic health care
- Practice of genetic counselling is focused on testing but also helping patients and families manage that information in their lives
- If this is successful the benefits of the genomic information will be maximised and potential harm minimised
- Greater focus on post test care and development of the evidence base to inform practice and implementation.

Thank you

- Colleagues
- All the organisations that have given me a chance
- All my mentors and collaborators
- The many students who attended the genetic counselling course
- My PhD students past and present
- Charities and support groups
- Patients and their families from whom I have learnt the most

And of course my family.

Acknowledge the past

Build for the future