ADVANCED
COURSES+
SCIENTIFIC
CONFERENCES

WELLCOME GENOME CAMPUS

WORLD CONGRESS ON GENETIC COUNSELLING

2-4 October 2019

ABSTRACT BOOK

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SCIENCE

ADVANCED COURSES+ SCIENTIFIC CONFERENCES 2019/20

WELLCOME GENOME CAMPUS

CONFERENCES

Evolution and Ecology of Cancer NEW
17-19 July

CRISPR and Beyond: perturbations at scale to understand genomes NEW
2-4 September

RNA Informatics
9-11 September

Optimising Multistudy Integrative Research NEW
18-20 September

Mechanisms and Evolution of Intergenerational Change NEW
24-26 September

World Congress on Genetic Counselling
2-4 October

Plant Genomes in a Changing Environment
16-18 October

Exploring Human Host-Microbiome Interactions in Health and Disease
23-25 October

Human Evolution
30 October-1 November

Epigenomics of Common Diseases
6-8 November

Mitochondrial Medicine
11-13 December

Evolutionary Systems Biology
12-14 February

Optimmunize: Improving the beneficial effects of vaccines NEW
19-21 February

Single Cell Biology
11-13 March

Genomics of Brain Disorders
18-20 March

Genomics of Rare Diseases
25-27 March

Proteomics in Cell Biology and Disease Mechanisms
30 March-1 April

Longitudinal Studies
20-22 April

Nursing, Genomics and Healthcare NEW
27-29 April

Antimicrobial Resistance – Genomes, Big Data and Emerging Technologies
6-8 May

Curating the Clinical Genome
20-22 May

Healthy Ageing
27-29 May

COURSES

LABORATORY COURSES

Train the Trainer: Capacity building for genomic surveillance of AMR in low- and middle-income countries NEW
6-11 October

Molecular Pathology and Diagnosis of Cancer
17-22 November

Derivation and Culture of Human Induced Pluripotent Stem Cells
9-13 December

Genomics and Clinical Microbiology
19-24 January

Genomics and Clinical Virology
23-28 February

Genetic Engineering of Mammalian Stem Cells
15-27 March

Next Generation Sequencing
20-27 April

Low Input Epigenomics NEW
8-15 May

COMPUTATIONAL COURSES

Genetic Analysis of Population-based Association Studies
23-27 September

Next Generation Sequencing Bioinformatics
1-7 December

Mathematical Models for Infectious Disease Dynamics
24 February-6 March

Fungal Pathogen Genomics
10-15 May

LECTURE/DISCUSSION COURSES

Science Policy: Improving the Uptake of Research into UK Policy
19-21 August

Molecular Neurodegeneration
2-6 December

Clinical Genomics: Scientific Fundamentals and Future Directions
29-31 January

Genomic Practice for Genetic Counselling
3-5 February

Practical Aspects of Small Molecule Drug Discovery
21-26 June

OVERSEAS COURSES

NGS Analysis for Genetic Diseases
5-6 November (Philippines)

Working with Protozoan Parasite Database Resources
10-15 November (Uruguay)

Next Generation Sequencing Bioinformatics
19-24 January (Chile)

Next Generation Sequencing Bioinformatics
9-14 February (Malaysia)

Molecular Approaches to Clinical Microbiology in Africa
21-27 March (The Gambia)

ONLINE COURSES

Bacterial Genomes: Disease Outbreaks and Antimicrobial Resistance

Bacterial Genomes: From DNA to Protein Function Using Bioinformatics

Bacterial Genomes: Accessing and Analysing Microbial Genome Data

Bacterial Genomes: Comparative Genomics using Artemis Comparison Tool (ACT) NEW

What is Genetic Counselling? NEW

Please see our website for more details and scheduling of online courses

CONNECTING SCIENCE

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World Congress on Genetic Counselling

Wellcome Genome Campus Conference Centre, Hinxton, Cambridge, UK
2 - 4 October 2019

Scientific Programme Committee:

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The University of British Columbia, Canada

Barbara Biesecker
RTI International, USA

Clara Gaff
University of Melbourne, Australia

Anna Middleton
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Dear colleague,

I would like to offer you a warm welcome to the Wellcome Genome Campus Advanced Courses and Scientific Conferences: World Congress on Genetic Counselling. I hope you will find the talks interesting and stimulating, and find opportunities for networking throughout the schedule. This meeting is run in partnership with the Society and Ethics Research Group at Wellcome Genome Campus and the Association of Genetic Nurses and Counsellors (UK and Republic of Ireland).

The Wellcome Genome Campus Advanced Courses and Scientific Conferences programme is run on a not-for-profit basis, heavily subsidised by the Wellcome Trust.

We organise around 50 events a year on the latest biomedical science for research, diagnostics and therapeutic applications for human and animal health, with world-renowned scientists and clinicians involved as scientific programme committees, speakers and instructors.

We offer a range of conferences and laboratory-, IT- and discussion-based courses, which enable the dissemination of knowledge and discussion in an intimate setting. We also organise invitation-only retreats for high-level discussion on emerging science, technologies and strategic direction for select groups and policy makers. If you have any suggestions for events, please contact me at the email address below.

The Wellcome Genome Campus Scientific Conferences team are here to help this meeting run smoothly, and at least one member will be at the registration desk between sessions, so please do come and ask us if you have any queries. We also appreciate your feedback and look forward to your comments to continually improve the programme.

Best wishes,

Dr Rebecca Twells
Head of Advanced Courses and Scientific Conferences
rebecca.twells@wellcomegenomecampus.org

General Information

Conference Badges
Please wear your name badge at all times to promote networking and to assist staff in identifying you.

Scientific Session Protocol
Photography, audio or video recording of the scientific sessions, including poster session is not permitted.

Social Media Policy
To encourage the open communication of science, we would like to support the use of social media at this year's conference. Please use the conference hashtag **#WCGC19**. You will be notified at the start of a talk if a speaker does not wish their talk to be open. For posters, please check with the presenter to obtain permission.

Internet Access
Wifi access instructions:

- Join the 'ConferenceGuest' network
- Enter your name and email address to register
- Click 'continue' to send an email to the registered email address
- Open the registration email, follow the link 'click here' and confirm the address is valid
- Enjoy seven days' free internet access!
- Repeat these steps on up to 5 devices to link them to your registered email address

Presentations
Please provide an electronic copy of your talk to a member of the AV team who will be based in the meeting room.

Poster Sessions
Posters will be displayed throughout the conference. Please display your poster in the Conference Centre on arrival. There will be two poster sessions during the conference.

Odd number poster assignments will be presenting in poster session 1, which takes place on Wednesday, 2 October at 18:30 – 19:30.

Even number poster assignments will be presenting in poster session 2, which takes place on Thursday, 3 October, at 18:00 – 19:00.

The abstract page number indicates your assigned poster board number. An index of poster numbers appears in the back of this book.

Conference Meals and Social Events
Lunch and dinner will be served in the Hall, apart from lunch on Wednesday, 2 October when it will be served in the Conference Centre. Please refer to the conference programme in this book as times will vary based on the daily scientific presentations. Please note there are no lunch or dinner facilities available outside of the conference times.

All conference meals and social events are for registered delegates. Please inform the conference organiser if you are unable to attend the conference dinner.

The Hall Bar (cash bar) will be open from 19:00 – 23:00 each day.

Dietary Requirements
If you have advised us of any dietary requirements, you will find a coloured dot on your badge. Please make yourself known to the catering team and they will assist you with your meal request.

If you have a gluten allergy, we are unable to guarantee the non-presence of gluten in dishes even if they are not used as a direct ingredient. This is due to gluten ingredients being used in the kitchen.

For Wellcome Genome Campus Conference Centre Guests
Check in
If you are staying on site at the Wellcome Genome Campus Conference Centre, you may check into your room from 14:00. The Conference Centre reception is open 24 hours.

Breakfast
Your breakfast will be served in the Hall restaurant from 07:30 – 09:00

Telephone
If you are staying on-site and would like to use the telephone in your room, you will need to contact the Reception desk (Ext. 5000) to have your phone line activated - they will require your credit card number and expiry date to do so.

Departures
You must vacate your room by 10:00 on the day of your departure. Please ask at reception for assistance with luggage storage in the Conference Centre.

For Holiday Inn Express & Red Lion, Whittlesford Bridge Hotel Guests
Check in
If you are staying on site at the Holiday Inn Express you may check into your room from 14:00. Hotel staff are on hand 24 hours a day.

Breakfast and Dining
Your breakfast will be served in the hotel, Great Room from 06:30 – 09:30.

The hotel also offers a relaxed licensed bar and lounge area.

Telephone and Internet
A telephone and free wireless internet access is available in your room, wireless is complimentary.

Departures
You must vacate your room by 12:00 on the day of your departure. A luggage store is available at the Conference Centre.

Wellcome Genome Campus Scientific Conferences guests receive a 15% discount on food at the Red Lion, Whittlesford Bridge Hotel. Please note there is a charge of £5 per night for car parking.

Taxis
Please find a list of local taxi numbers on our website. The conference centre reception will also be happy to book a taxi on your behalf.

Transfers
If you are staying off site, a complimentary shuttle service has been organised with Richmond's Coaches. The shuttle service is as follows:

Wednesday,2 October
Holiday Inn Express & Red Lion, Whittlesford – Conference Centre 12:30
Conference Centre – Holiday Inn Express & Red Lion, Whittlesford 21:00

Thursday, 3 October
Holiday Inn Express & Red Lion, Whittlesford – Conference Centre 08:30

Conference Centre – Holiday Inn Express & Red Lion, Whittlesford 21:00

Friday, 4 October
Holiday Inn Express & Red Lion, Whittlesford – Conference Centre 08:30

Return Ground Transport
Complimentary return transport has been arranged for 14:20 on Friday, 4 October to Cambridge station and city centre (Downing Street), and Stansted and Heathrow airports.

A sign-up sheet will be available at the conference registration desk from 15:45 on Wednesday, 2 October. Places are limited so you are advised to book early.

Please allow a 30 minute journey time to both Cambridge and Stansted Airport, and two and a half hours to Heathrow.

Messages and Miscellaneous
Lockers are located outside the Conference Centre toilets and are free of charge.

All messages will be posted on the registration desk in the Conference Centre.

A number of toiletry and stationery items are available for purchase at the Conference Centre reception. Cards for our self-service laundry are also available.

Certificate of Attendance
A certificate of attendance can be provided. Please request one from the conference organiser based at the registration desk.

Contact numbers
Wellcome Genome Campus Conference Centre – 01223 495000 (or Ext. 5000)
Wellcome Genome Campus Conference Organiser (Laura) – 07733 338878

If you have any queries or comments, please do not hesitate to contact a member of staff who will be pleased to help you.

Conference Summary

Wednesday, 2 October

12:30-13:50	Registration with lunch
13:50-14:00	Welcome and Introduction
14:00-14:45	Keynote lecture by *Laura Hercher, Sarah Lawrence College, USA*
14:45-15:45	Session 1: Genetic Counselling: countries with an established profession
15:45-16:15	Afternoon tea
16:15-17:30	Session 2: Genetic Counselling: countries with an emerging profession
17:30-18:30	Panel led discussion: The 'therapeutic alliance' in genetic counselling, what is it and how do we translate this to non-specialist colleagues?
18:30-19:30	Poster session 1 (odd numbers) with drinks reception
19:30	Dinner & cash bar

Thursday, 3 October

09:00-10:30	Session 3: Advocating for Genetic Counselling
10:30-11:00	Morning coffee
11:00-12:30	Session 4: What changes as a consequence of genetic counselling?
12:30-14:00	Lunch
14:00-15:15	Session 5: Achieving valued outcomes for genetic counselling patients
15:15-16:00	Afternoon tea
16:00-16:45	Keynote lecture by *Shivani Nazareth, Clear Genetics, USA*
16:45-18:00	Panel led discussion: Technology and Genetic Counselling: how do we combine the two?
18:00-19:00	Poster session 2 (even numbers) with drinks reception
19:00	Conference dinner & cash bar

Friday 4 October

09:00-10:30	Session 6: Research into the Counselling Process
10:30-11:00	Morning coffee
11:00-12:15	Session 7: Leadership in genetic counselling
12:15-13:15	Panel led discussion: The evolution of genetic counselling practice
13:15-13:20	Closing remarks and conference summary
13:20-14:20	Lunch
14:20	Coaches depart to Cambridge city centre and train station & Heathrow airport via Stansted airport

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World Congress on Genetic Counselling

Wellcome Genome Campus Conference Centre,
Hinxton, Cambridge

2 – 4 October 2019

Lectures to be held in the Francis Crick Auditorium
Lunch and dinner to be held in the Hall Restaurant
Poster sessions to be held in the Conference Centre

Spoken presentations - If you are an invited speaker, or your abstract has been selected for a spoken presentation, please give an electronic version of your talk to the AV technician.

Poster presentations – If your abstract has been selected for a poster, please display this in the Conference Centre on arrival.

Conference programme

Wednesday, 2 October

12:30-13:50	Registration with lunch
13:50-14:00	Welcome and Introduction *Anna Middleton, Wellcome Genome Campus, UK*
14:00-14:45	**Keynote lecture** *Chair: Anna Middleton, Wellcome Genome Campus, UK* Genetic Counsellors: imagining the next 10 years *Laura Hercher* *Sarah Lawrence College, USA*
14:45-15:45	**Session 1: Genetic Counselling: countries with an established profession** *Chair: Jehannine Austin, University of British Columbia, Canada* 14:45 Client-Centred genetic counselling in Japan *Chieko Tamara* *FMC Tokyo Clinic, Japan* 15:15 Models for the provision of additional genomic findings *Clara Gaff* *Melbourne Genomics Health Alliance, Australia*
15:45-16:15	Afternoon tea

16:15-17:30 **Session 2: Genetic Counselling: countries with an emerging profession**
Chair: Michelle Bishop, Health Education England, UK

16:15 The delivery of genetic counselling in Hong Kong from a counsellor's perspective
Olga Zayts
The University of Hong Kong, Hong Kong

16:45 Genetic counselling in Portugal: enduring challenges, emerging research
Alvaro Mendes
i3S, University of Porto, Portugal

17:15 LEADERS: A culturally tailored approach to genetic counseling for Arab communities in northern Israel
Naama Nahama Cohen Kfir
Bar Ilan University, Israel

17:30-18:30 **Panel led discussion: The 'therapeutic alliance' in genetic counselling, what is it and how do we translate this to non-specialist colleagues?**
Chair: Anna Middleton, Wellcome Genome Campus, UK
1. Barbara Biesecker, RTI International, USA
2. Jehannine Austin, University of British Columbia, Canada
3. Clara Gaff, Melbourne Genomics Health Alliance, Australia

18:30-19:30 **Poster session 1 (odd numbers) with drinks reception**

19:30 Dinner & cash bar

Thursday, 3 October

09:00-10:30 **Session 3: Advocating for Genetic Counselling**
Chair: Chris Patch, Genomics England, UK

09:00 Selling what genetic counsellors do to policy makers
Michelle Bishop
Health Education England, UK

09:30 Embedding genomics into clinical care – Meeting the needs of policy makers
Elly Lynch
Melbourne Genomics Health Alliance, Australia

09:45 Specific considerations in genetic counselling of transgender patients: altered risk factors and cultural competencies
Reubs Walsh
Gender Identity Research and Education Society (GIRES)

10.15 High risk – what's next? decisional conflict, regret and satisfaction among pregnant women making choices about further prenatal testing
Charlotta Ingvoldstad Malmgren
Karolinska University Hospital, Sweden

10:30-11:00 Morning coffee

11:00-12:30 **Session 4: What changes as a consequence of genetic counselling?**
Chair: Galen Joseph, University of California, San Francisco, USA

11:00 Behaviour change as an outcome of psychiatric genetic counseling
Jehannine Austin
University of British Columbia, Canada

11:30 Experience as knowledge: Perceptions of Screening amongst Families Living with Genetic Disease
Felicity Boardman
Warwick Medical School, UK

12:00 Improving the communication of genomic results to patients with rare diseases and their families using Experience-based Co-design (EBCD)
Alessia Costa
King's College London, UK

12:15 Parent experiences with ultra-rapid genomic sequencing in paediatric acute care
Samantha Ayres
Victorian Clinical Genetics Services, Australia

12:30-14:00 Lunch

14:00-15:15 **Session 5: Achieving valued outcomes for genetic counselling patients**
Chair: Barbara Biesecker, RTI International, USA

14:00 The Genomics Outcome Scale: A short form of the Genetic Counselling Outcome Scale
Marion McAllister
Cardiff University, UK

14:30 Communicating Polygenic Disease Risk for Coronary Artery Disease: Design and Optimization of a Polygenic Score Report
Deanna Brockman
Massachusetts General Hospital, USA

14:45 Pretest Chatbots: Is Information Value Neutral?
Kelly C Donahue
Genetic Support Foundation, USA

15:00 Effect of Providing Education about Carrier Results via Web versus Genetic Counselor on the Subsequent Therapeutic Relationship
Lori Erby
National Human Genome Research Institute, USA

15:15-16:00 Afternoon tea

16:00-16:45 **Keynote lecture**
Chair: Christine Patch, Genomics England, UK

Futuristic models of genetic counselling
Shivani Nazareth
Clear Genetics, USA

16:45-18:00 **Panel led discussion: Technology and Genetic Counselling: how do we combine the two?**
1. Shivani Nazareth, Clear Genetics, USA
2. Patricia Birch, University of British Columbia, Canada
3. Laura Hercher, Sarah Lawrence College, USA
4. Jon Roberts, Wellcome Genome Campus/Addenbrooke's, UK

18:00-19:00 **Poster session 2 (even numbers) with drinks reception**

19:00 Conference dinner & cash bar

Friday 4 October

09:00-10:30 **Session 6: Research into the Counselling Process**
Chair: Alvaro Mendes, i3S, University of Porto, Portugal

09:00 A family systems approach to genetic counselling; development of interventions
Rhona MacLeod
Manchester Centre for Genomic Medicine, UK

09:30 Communication of clinical uncertainties: A systematic literature review
Barbara Biesecker
RTI International, USA

10:00 An anthropological view of genetic counselling
Galen Joseph
University of California, San Francisco, USA

10:30-11:00 Morning coffee

11:00-12:15 **Session 7: Leadership in genetic counselling**
Chair: Shivani Nazareth, Clear Genetics, USA

11:00 Preparing a genetic counselling workforce of the future
Alison McEwen
University of Technology Sydney, Australia

11:30 International Genetic Counseling: What do Genetic Counselors do?
Jon Weil
California St Univ. Stanislaus, USA

	11:45	Developing a nationally agreed cross-professional competency framework to facilitate consent for genomic testing *Amanda Pichini* *University Hospitals Bristol NHS Foundation Trust, UK*
	12:00	Patient and counselor perceptions of a telehealth genetic counseling model. Can high efficiency co-exist with high satisfaction? *Kiley Johnson* *GeneMatters, USA*
12:15-13:15	**Panel led discussion: The evolution of genetic counselling practice** *1. Chieko Tamara, FMC Tokyo Clinic, Japan* *2. Marion McAllister, Cardiff University, UK* *3. Gemma Chandratillake, Eastern Genomic Lab Hub, UK*	
13:15-13:20	Closing remarks and conference summary *Programme Committee*	
13:20-14:20	Lunch	
14:20	Coaches depart to Cambridge city centre and train station & Heathrow airport via Stansted airport	

These abstracts should not be cited in bibliographies. Materials contained herein should be treated as personal communication and should be cited as such only with consent of the author.

Spoken Presentations

Genetic Counsellors: imagining the next 10 years

Laura Hercher

Sarah Lawrence College, USA

Abstract not available.

Notes

Client-Centred genetic counselling in Japan

Chieko Tamara

FMC Tokyo Clinic, Japan

Abstract not available at time of printing.

Notes

Models for the provision of additional findings

Clara Gaff

University of Melbourne, Australia

There is growing recognition internationally that provision of secondary findings to patients having1 genomic testing must be addressed. In the USA, this is well established with secondary analysis for additional findings being offered at the same time as diagnostic sequencing. In public health systems elsewhere, diagnostic laboratories generally do not offer secondary findings. Concerns about this 'opportunistic screening' include the diversion of finite clinical and laboratory resources away from the pressing needs of those with existing medical conditions, particularly as genomic testing is incorporated into the practice of a wide range of medical specialists.

How can the preferences of patients to be offered secondary findings, limitations on health care resources and the importance of informed decision making be reconciled? Cost-effective models of care are needed whereby patients can choose to receive additional findings at a point in their life that best suits them and genetic counselling services can focus on those whose needs cannot be met through, for example, the education or decision support approaches employed by screening programs.

As a first step towards identifying such models, we offered reanalysis for additional findings to two cohorts: (1) 106 newborns with congenital deafness at the time of diagnostic testing and (2) 200 adults *after* they received diagnostic sequencing results. Process evaluations have been conducted using data from patient surveys, health professional interviews and recorded consultations. Data has also been used to develop a trainee chatbot for adults. Implications for future service delivery will be discussed.

Notes

The delivery of genetic counselling in Hong Kong from an interdisciplinary perspective

Olga Zayts and Brian HY Chung, The University of Hong Kong, Hong Kong

In this talk we present the ongoing interdisciplinary collaboration between medical professionals and social scientist to research the delivery of genetic counselling in Hong Kong that spans over the last 14 years. In Hong Kong genetic counselling is largely practiced within clinical genetic services that are integrated in the public health sector. There is currently no in-country certification/ licensure available, and genetic counselling training is obtained either overseas or on the job. While many of the professional practices and principles are transferrable from the contexts where genetic counselling has been long established (e.g. the USA, Canada, the UK, Australia), there are unique historical, cultural and socioeconomic factors (Resta, 2017) that affect the local practices of genetic counselling in Hong Kong. To that end, in our collaborative projects we have focused on the context-specific practices of genetic counselling, in particular the practices that evolve in the process of counselling clients with diverse sociocultural and linguistic backgrounds.
While genetic counselling field is not short of insights and practical guidelines derived from commentaries on sociocultural and language issues, and increasingly, research that employs interviews and surveys, empirical research that draws on real-life interactional data is scarce. Over the years, we have developed an *innovative empirical approach* to examine the sociocultural dimensions of genetic counselling practices that: (a) draws on large corpora of real-life clinical interactions between genetic counselling professionals and clients; (b) employs an integrated discourse analytic approach supplemented by other (quantitative) methods; (c) incorporates secondary data sources, such as interviews and focus groups with clients and family members; (d) draws on multidisciplinary insights and expertise of the research team. Such approach to analyzing large data corpora allows for the identification of trends within the data, and yields clear practical insights for enhancing context-specific cultural training in genetic counselling (Zayts et al., 2019).
In our talk we outline several major themes related to genetic counselling in the specific sociocultural contexts of Hong Kong that are consistent across the data corpora from different genetic counselling settings, namely: (1) familial dimensions of counselling, including negotiation of roles - responsibilities within a family, familial piety, children's assent/ consent; (2) the understanding of 'culture' and its impact on genetic counselling; (3) the language of counselling, including lingua franca interactions. Drawing on examples from three specific contexts of investigation, prenatal screening for Down syndrome, counselling for G6PD deficiency, and counselling for SADS, we illustrate how genetic counsellors and clients negotiate these sociocultural issues in the ongoing talk-in-interaction, and discuss some suggestions aimed at enhancing counsellors' sociocultural awareness, including self-awareness, in their professional practice.

References

Resta, R. (2017). What have genetic counselors been doing and have they been any good at it? Keynote lecture. 1st World Congress on Genetic Counselling, Hinxton, UK.
Zayts, O., Shipman, H., Fung, J. L.-H., Liu, A. P.-Y., Kwok, S.-Y., Tsai A. C.-H., Yung, T.-C., Chung, B. H.-Y. (2019). The different facets of "culture" in genetic counseling: A situated analysis of genetic counseling in Hong Kong. American Journal of Medical Genetics Part C, 181C: 187– 195. https://doi.org/10.1002/ajmg.c.31699

Notes

Genetic counselling in Portugal: enduring challenges, emerging research

Álvaro Mendes, i3S, University of Porto, Portugal
UnIGENe and Centre for Predictive and Preventive Genetics, IBMC – Institute for Molecular and Cell Biology, i3S – Instituto de Investigação e Inovação em Saúde, Univ. Porto, Portugal

Portugal is one of the European countries where genetic counselling is emerging as an independent field. In the last decade important steps have been made towards establishing safe and ethical practice, delivered by adequately trained professionals. Some of these included the establishment of a master level training programme at the University of Porto, and an increased awareness to the need to include genetic counsellors as part of multidisciplinary genetics healthcare teams. Whilst the recognition of genetic counsellors as healthcare professionals and their full integration in the NHS is yet to be seen, a growing body of research, however, has been consistently produced.
This talk intends to briefly report some of the developments of genetic counselling in Portugal. It also aims to provide examples of research that has been emerging (particularly in the context of pre-symptomatic testing for late onset neurological diseases) and discuss how this research may be relevant to the provision of genetic counselling and psychosocial support to those living with genetic diseases and genetic risks.
I will reflect on the relevance of delivering appropriate genetic counselling and ongoing support to patients and families, and of developing research that may enhance the value of genetic counselling for those who use it. This will set the stage to affirm the need to integrate genetic counsellors in the multidisciplinary teams of all medical genetics services, and to maintain effective multidisciplinary team-working and communication among health professionals at a community level.

Notes

LEADERS: A culturally tailored approach to genetic counseling for Arab communities in northern Israel.

Nehama Cohen Kfir, Mary Rudolf, Miriam Bentwich, Nomy Dickman, and Tzipora C. Falik-Zaccai

Institute of Human Genetics, Galilee Medical Center, Nahariya, Israel. Azrieli Faculty of Medicine Bar-Ilan, Safed, Israel

Buckground : Many couples of Arab ethnic origin in North Israel, have doubts regarding genetic counseling, in part, due to misconceptions of the messages that are relayed.
Aims: To explore attitudes regarding genetic counseling in local Arab ethnic groups; develop "tailored" genetic counseling appropriate to needs and evaluate the outcome.
Methods: Semi-structured interviews were conducted with potential Arab counselees and focus groups were held with "Mother and Child" clinic nurses and genetic counselors. Content analysis revealed key themes regarding lack of knowledge and negative attitudes towards genetic counselling and gaps in expectations of the outcome of genetic counseling between counselees who emphasized their need for empathy, and counselors who focused on genetic data to foster informed decisions. Based on the findings, learning theory, self-regulation theory and cultural competency models, a new approach for practical guidance was designed, using the mnemonic LEADERS.
LEADERS represents: (a) Launch - Explaining what is expected to happen to relieve stress and adjust expectations; (b) Expose fears, misconceptions, and gaps- Disclosure of misconceptions enabling learning new concepts; (c) Acknowledge differences in attitudes, knowledge, and beliefs - to establish trust and empathy; (d) Discuss genetic information, using visuals and analogies from the counselee's environment; (e) Encourage counselees to respond and to ask questions; (f) Recommendations; (g) Shared decision-making - Achieving an agreed decision incorporating counselee's preferences.
The LEADERS model was introduced at the Galilee Medical Center following training of the counselling team. Using the validated Genetic Counseling Outcome Scale (GCOS-24), evaluation of the new counselling model was conducted among counselees in a 6-month period following the workshop, and compared to 'historic' controls from 6 months prior to the LEADERS training.
Results: 106 'historic' controls and 112 counselees completed GCOS (84.8%, 88.8% response rate). There was no difference in GCOS-24 score (5.1±0.57 vs. 5.18±0.61). However, there was significant improvement in the emotional domain (4.47±0.85 vs. 4.84±1.00, p=0.002). The greatest improvement was in less educated (4.34±0.85 vs. 4.96±1.00, p=0.001), more secular (4.46±0.78 vs. 4.94±1.06, p=0.004), Muslim (4.37±0.89 .vs 4.85±1.01, p=0.015) and Druze (4.55±0.78 vs. 4.93±0.99, p=0.05) counselees. Counselors reported high satisfaction using the model.
Conclusions: The LEADERS Model is a promising approach which was positively received by counselors, and was found to have a benefit on counselees' emotions, a domain that can potentially influence counselees' ability to make informed decisions. This is of high relevance to Arab communities in Israel and beyond, where genetic counselling has poor uptake.

Notes

Selling what genetic counsellors do to policy makers

Michelle Bishop

Education Development Lead (Genomics), Health Education England

Genomics has been a focus within England's National Health Service (NHS) for the last six years, ever since the government announced its ambitious aim to sequence 100,000 genomes from NHS patients. During this period, there has been a spotlight on the specialist clinical teams delivering genomic services, with a focus on education needs, training pathways and required workforce numbers.

Early engagement with Health Education England (HEE), the organisation responsible for the education and training of the future and existing NHS workforce, indicated that genetic counselling was an 'invisible' workforce in the NHS that was not being considered as part of either the wider workforce planning or commissioning processes. The NHS is a large organisation employing 1.2 million people, of whom 700,000 have a clinical role. The clinical workforce is dominated by medics and nurses, and as such these professions receive more attention from those making decisions around educational planning and workforce numbers. How do you champion a workforce of fewer than 300 people, working in a niche area, within this landscape?

As a trained genetic counsellor, now working in education and policy, I will present a personal reflection on the work that I have done over the last five years to: (1) raise the profile of genetic counsellors throughout HEE; (2) navigate the transition to a new nationally commissioned training programme; and (3) ensure that the voice of genetic counsellors is heard during key policy discussions. I will outline the obstacles that I have overcome and provide insights into how I prepared for these challenges. While this presentation will provide a personal narrative, many of the strategies I have employed could be used by other genetic counsellors when advocating for the profession in different settings.

Notes

Embedding genomics into clinical care – Meeting the needs of policy makers

Elly Lynch 1,2,3, Melissa Martyn 1,3,4, Clara Gaff 1,3,4

1. Melbourne Genomics Health Alliance
2. Victorian Clinical Genetics Service
3. Murdoch Children's Research Institute
4. The University of Melbourne

For genomics to be embedded into everyday healthcare, policy makers need evidence to make decisions around funding of services and tests. Melbourne Genomics Health Alliance is a clinically driven research program designed to provide evidence to inform and meet the needs of policy makers.
Evidence generated from Melbourne Genomics has already resulted in changes at government and service levels. This includes the Medical Services Advisory Council of Australia making a recommendation to the federal government that funding be made available for whole exome sequencing for childhood syndromes through a medicare item number. In addition, the state government allocated AU$8.3million of funding for Clinical Genetic Services in Victoria for genomic testing. Results continue to be generated as genomic testing is compared to standard care across sixteen different disease areas through clinical design projects (flagships), creating evidence for when genomics is clinically useful and cost effective.
In order for the evidence produced to be useful for decision makers, a hybrid effectiveness-implementation model was used in the design of the clinical Flagships, whereby clinicians were involved in the design and evaluation of the model for offering testing. The scope of the Flagship was clearly defined upfront. Upon completion of each Flagship, in addition to the research publications, an evidence report for state government was generated, which included the main outcomes of the project, including health economic outcomes where available and key messages from which recommendations could be made.
Challenges included working with clinicians to ensure the evidence was presented in a way that was helpful for state government. Clinicians are often experienced in typical research methodologies and therefore considering the implications for the broader healthcare system and workforce was often a challenge. Government input was provided as evidence reports were created, ensuring that messages could be carefully drafted around state government priorities, including but not limited to questions around potential costs to the health system and impacts on service delivery if genomic testing continued in a particular disease area.
Examples and lessons learned from the conduct of the Flagships and communication with policy makers will be presented.

Notes

Specific considerations in genetic counselling of transgender patients: altered risk factors and cultural competencies.

Reubs Walsh

Gender Identity Research and Education Society (GIRES)

As society's attitudes to gender continue to evolve, it is increasingly apparent that gender is a biopsychosocial phenomenon separable from karyotype or reproductive anatomy. Estimates of the prevalence of gender incongruence (where the gender assigned at birth based on external reproductive anatomy is different from the gender identity experienced by the individual; i.e. transgender people) have been rising over the past few decades, ranging from 0.17 in 100,000 in 1996 to 0.1% in 2007, and evidence from surveying young people, up to 4% of whom identify as trans, shows this trend is set to continue. Many transgender people choose to undergo hormonal and/or surgical treatments to better align their bodies with their gender. These treatments present a source of uncertainty for genetic counsellors in assessing certain types of risk (e.g. breast, cervical or prostate cancer and cardiovascular disease), where there is currently a lack of consensus on a number of areas where trans-specific healthcare may alter the risk profile for trans patients in other areas of their medical care. Furthermore, trans people often report negative experiences in healthcare stemming from a lack of cultural competencies among clinicians, which contribute to risk via differences in screening access and compliance. This presentation aims to cover the basic information that genetic counsellors need to provide their usual high standard of care to transgender patients, including an overview of current opinion on the influence of hormonal and surgical treatment on relevant cancer risks, and a discussion of cultural competency and sensitivity in the transgender context.

Notes

High risk – what's next? decisional conflict, regret and satisfaction among pregnant women making choices about further prenatal testing.

Charlotta Ingvoldstad Malmgren3 Tanja Schlaikjær Hartwig, Caroline Borregaard Miltoft2, , Ann Tabor2, Finn Stener Jørgensen1

1. Fetal Medicine Unit, Department of Obstetrics and Gynecology, Copenhagen University Hospital Hvidovre, Denmark
2. Fetal Medicine Center, Department of Obstetrics, Copenhagen University Hospital Rigshospitalet, Denmark
3. Center for Fetal Medicine, Department of Obstetrics and Gynecology, Karolinska University Hospital, Sweden

In Denmark, Non-invasive prenatal testing, NIPT, is offered as an alternative to ivasive testing to women with a high-risk result from the combined first trimester screening test. The aim of this study was to investigate decision-making among Danish high-risk pregnant women when choosing between non-invasive prenatal testing (NIPT), invasive testing or no further testing. Women with a high-risk result from the combined first trimester screening were invited to fill in two online questionnaires at GA 12-14(Q1) and GA 24(Q2). The scales used were Decisional Conflict and Regret Scales, Satisfaction with genetic Counselling Scale and Health-Relevant Personality Inventory.
In total, 339 women were included, and the response rates were 76 % on Q1 and 88% on Q2, respectively. Of the participants, 75.4% chose an invasive test and 23.8% chose NIPT. The median DCS score among all participants was within the level associated with implementing decisions, whereas 13.3% had a high level of decisional conflict. Choosing NIPT was associated with a high decisional conflict; receiving genetic counselling the same day was associated with a high decisional conflict; and a high satisfaction with the genetic counselling was associated with low decisional conflict. Furthermore, 'alexithymia', the personality sub-trait that describes a disinterest or inability in identifying and understanding feelings, was associated with low decisional conflict. High decisional regret was associated with high decisional conflict and low satisfaction with genetic counselling.
in this study we present evidence that satisfaction with, and timing of counselling are essential factors to limit decisional conflict. Also, the results indicate that women choosing NIPT have more decisional conflict compared to women choosing invasive testing. There was a significant association between high decisional conflict and later decisional regret.

Notes

Behaviour change as an outcome of psychiatric genetic counseling

Jehannine C. Austin PhD FCAHS CGC

UBC Departments of Psychiatry and Medical Genetics, Executive Director BC Mental Health and Substance Use Services Research Institute, Rm A3-127, 938 W28th Ave, Vancouver, BC

Background: Psychotropic medication non-adherence is a complex problem for people with serious mental illness (SMI) (schizophrenia, bipolar disorder, schizoaffective disorder). Beliefs about illness etiology may impact adherence, thus we hypothesized that psychiatric genetic counseling (PGC), improves: 1) the proportion of people who are adherent, and 2) the degree people adhere to medications.

Methods: We recruited people with SMI to complete the Brief Adherence Rating Scale (BARS,scored 0-100% adherent per medication), twice Pre-PGC (T1:1month Pre-PGC and T2:immediately Pre-PGC) and Post-PGC (T3:1month and T4:2months Post-PGC). **Hypothesis 1**: We used a mixed effects logistic regression to model "adherent" (defined as >70% adherent to *all* medications)proportions over the four timepoints. **Hypothesis 2:** We used paired Wilcoxon signed-rank test to compare change in BARS scores between T1 to T2 (no intervention), and T2 to T4 (after PGC).

Results: Hypothesis 1: Using N=31 participants, proportion (%) of adherent individuals was greatest at T4 (48%) and lowest at T2 (29 %), but though the effect size was moderate, the relationship between the % adherent and timepoint was not statistically significant (p=0.40). **Hypothesis 2**: On average adherence improved after PGC (mean change in BARS= 1.3), compared to an average decrease after no intervention (mean change in BARS= -2.7), but the difference was not statistically significant.

Conclusion: PGC may improve adherence to psychotropic medications, however larger studies purposively sampled for those who are non-adherent are needed.

Notes

Experience as knowledge: Perceptions of Screening amongst Families Living with Genetic Disease

Dr Felicity Kate Boardman

Associate Professor, Warwick Medical School

As genomic medicine continues to expand, and pre-conception genetic screening panels begin to emerge around the globe (e.g. Australia, The Netherlands), questions of which conditions are of relevance to couples for reproductive planning are now increasingly urgent.

Disease severity is often used as a proxy for the significance of the disease and consequently its inclusion on screening panels. However, 'disease severity' is notoriously difficult to define, and often relies entirely on medical interpretations of the condition's symptoms and their perceived impact on quality of life, rather than the accounts of people actually living with those conditions that are candidates for genetic screening programmes.

In order to explore the experiences and views of people living with a range of different genetic conditions, including both their attitude towards their condition and their views of population screening for it, this project involved 160 qualitative interviews with people living with genetic disease, their family members and genomic sequencing volunteers, as well as over 1,500 quantitative surveys with these groups. The conditions included in the study were selected based on their prevalence in the UK population, and the range and contrast of impacts associated with them: Cystic Fibrosis, Spinal Muscular Atrophy, Fragile X Syndrome, Thalassaemia and Haemophilia.

A mixed methods analysis of the resulting qualitative and quantitative data highlighted the way in which 'experiential knowledge', or 'lived experience' of a genetic condition is an important site of expertise that is of great relevance in defining, and determining, the boundaries of disease severity. For example, data from this study disrupts the assumed correlation between poor quality of life and increased disease severity, instead suggesting that particular types of impairment experience (for example, deterioration, pain) are more closely associated with poor quality of life than the overall degree of disability experienced by that person. Similarly, the social, cultural and environmental contexts in which genetic disease is experienced were illuminated through the data, demonstrating the range of factors that determine experiences of genetic disease, and yet are not accounted for in medical definitions of severity.

This talk presents verbatim data from this study from participants, and concludes by demarcating 'experiential knowledge' as a site of expertise on genetic disease, ultimately calling for its greater inclusion in policy decisions that concern population-level genetic screening programmes, as well as a re-evaluation of commonly accepted ideas about both disease severity and quality of life as they relate to genetically disabled people.

Notes

Improving the communication of genomic results to patients with rare diseases and their families using Experience-based Co-design (EBCD).

Alessia Costa, Christine Patch, Alison Metcalfe, Glenn Robert,

Genomics England, Queen Mary University of London, London. UK King's College London, London. UK
Sheffield Hallam University, Sheffield. UK

Genome sequencing is rapidly moving from research into clinical practice with the aim of finding a diagnosis for previously undiagnosed conditions. For families, receiving genomic results (incl. diagnosis, VUS, null results) can have a significant socio-psychological impact. This is putting services under pressure to identify optimal ways to feed back results to patients and families and support them in the early stages of coming to terms with the results.

As part of the European Union's Horizon 2020 Solve-RD project, we have worked with genetic health professionals and families of patients with developmental delays and rare syndromes to co-design an intervention to improve the communication of genomic diagnostic results in two health services, one in the UK and one in Czech Republic. Using Experience-based Co-design (EBCD), we conducted qualitative interviews and non-participant observations to explore families' and genetic professionals' experiences of receiving and communicating results. Through a series of participatory research activities, including workshops and co-design processes, we have facilitated participants to discuss and reflect on their experiences, identify shared priorities for change and work together to develop and test interventions to improve selected aspects of current services.

In the proposed paper, we will introduce the EBCD process used and outline the outcomes at the two services. These include the priorities for change that were jointly identified by families and health care staff and the interventions that were developed to improve these specific aspects of current service. We will discuss implications for the role of genetic counsellors and clinicians in genomic health care and current challenges. The presentation will include the opportunity to view sections of a short film about families' experiences of receiving genomic results, which was made as part of the study.

Notes

Parent experiences with ultra-rapid genomic sequencing in paediatric acute care

Samantha Ayres1,3,5, Gemma R Brett 1,2,3, Melissa Martyn2,3,4, Michelle de Silva1,2,5, Kirsten Boggs5,6,7, Anne Baxendale5,8, Sarah Borrie5,8, Sarah King-Smith5,9, Lyndon Gallacher1,2, Jason Pinner6, Sarah Sandaradura7, Meredith Wilson7, Christopher Barnett8, Chirag Patel10, Anand Vasudevan11, Emma Krzesinski12,13, Sebastian Lunke1,2,5, Zornitza Stark1,2,5

1 Victorian Clinical Genetics Services, Murdoch Children's Research Institute, Melbourne, Australia
2 University of Melbourne, Melbourne, Australia
3 Melbourne Genomics Health Alliance, Melbourne, Australia
4 Murdoch Children's Research Institute, Melbourne, Australia
5 Australian Genomics Health Alliance, Australia
6 Sydney Children's Hospitals Network - Randwick, Sydney, Australia
7 Sydney Children's Hospitals Network - Westmead, Sydney, Australia
8 Paediatric and Reproductive Genetics Unit, South Australian Clinical Genetics Service, Adelaide, Australia
9 Centre for Cancer Biology, SA Pathology, University of South Australia, Adelaide, Australia
10 Genetic Health Queensland, Royal Brisbane and Women's Hospital, Brisbane, Australia
11 Royal Women's Hospital, Melbourne, Australia
12 Monash Children's Hospital, Melbourne, Australia
13 Department of Paediatrics, Monash University, Melbourne, Australia

Background: Emerging evidence that rapid turnaround times impact the clinical utility of genomic testing in acute paediatrics is driving widespread adoption. However, little is known about the experience that parents of critically unwell infants and children have during the testing process and beyond.
Methods: Participants were recruited as part of the Australian Genomics Acute Care study, a national rapid genomic diagnosis program for infants and children admitted to intensive care with suspected genetic conditions. Pre- and post-test counselling was provided by genetic health professionals. Over 95% of parents offered testing gave consent. Results were available within five days of sample receipt. Parents were surveyed >12 weeks after results return. We explored parental experiences with consent processes, perceived impact of testing on child health, relationships and reproductive decisions. This questionnaire included the Decision Regret, Short Form Genetic Counselling Outcomes and PedsQL Family Impact Module scales.
Results: From 42 respondents in the first ten months (RR=63%), most felt they received enough information during pre-test (n=40, 100%) and post-test (n=34, 85%) counselling. Few respondents (n=6, 15%) reported decisional regret regarding testing. Perceptions varied about the benefits of rapid genomic sequencing for the child. The majority of respondents (n=22, 55%) were extremely concerned about the condition occurring in future children, regardless of their actual or self-perceived recurrence risk. Fourteen respondents (35%) reported the test impacted their reproductive plans.
Importance: Understanding parental experiences, opinions, and the short and long term impacts on families will guide the design and delivery of rapid genomic diagnosis programs.

Notes

The Genomics Outcome Scale: A short form of the Genetic Counselling Outcome Scale

Marion McAllister, 1. Peter E. Grant, 2. Maria Pampaka, 3. Katherine Payne, 4. Angus Clarke

1,4: Division of Cancer & Genetics, School of Medicine, Cardiff University, Cardiff, UK; 2: Departments of Social Statistics (School of Social Science) and Education (School of Environment, Education and Development), The University of Manchester, Manchester, UK; 3: Division of Population Health, Health Services Research and Primary Care, The University of Manchester, Manchester, UK

The Genetic Counselling Outcome Scale (GCOS-24) is a 24-item patient-reported outcome measure developed for use in evaluating genetic counselling and testing interventions. A short form of GCOS-24 would be less burdensome for patients and could be used where genetic testing is done outside clinical genetics services e.g. oncology, paediatrics. The aim in this study was to create to create a short 6-item form. Cognitive interviews were used to explore interpretability of GCOS-24 items and identify items highly valued by the target population. The Graded Response Model (GRM) was then used to examine item discrimination in an existing set of GCOS-24 responses (n=395). Three principles guided item selection for the short form: (i) Items with poor discriminative properties were excluded; (ii) Items capturing a similar concept were not selected together; (iii) item information curves and cognitive interview findings were used to identify superior items. Rasch analysis was then applied to establish the optimal scale. Ten cognitive interviews were conducted with members of families affected by a genetic condition, recruited through Genetic Alliance UK. Interview transcripts were analysed using qualitative methods to identify twelve GCOS-24 items highly valued by participants. GRM item characteristic curves and item information curves were generated, and combined with the qualitative findings to select ten items that were both highly valued and perform well. Finally, items were iteratively removed and permutated to establish optimal fit statistics using the Rasch model. A six-item questionnaire with a five-point Likert Scale was created (The Genomics Outcome Scale (GOS)). Correlation between GCOS-24 and GOS is high (r=.838, 99% confidence), demonstrating that GOS maintains the ability of GCOS-24 to capture empowerment, whilst providing a less burdensome scale for respondents. GOS will benefit from further psychometric assessment of reliability and responsiveness.

Notes

Communicating Polygenic Disease Risk for Coronary Artery Disease: Design and Optimization of a Polygenic Score Report

Deanna Brockman, Bang Wong, Lia Petronio, Andrew Tang, Tera Bowers, Alyssa Macbeth, Renee Pelletier, Candace Patterson, Trish Vosburg, Akl Fahed, Niall Lennon, Amit Khera

Center for Genomic Medicine, Massachusetts General Hospital, Boston, MA; Broad Institute of MIT and Harvard, Boston, MA.

BACKGROUND: The traditional approach to identifying individuals at increased risk for common diseases has focused on finding rare, monogenic variants. However, the majority of common diseases occur in individuals without such variants and are recognized as 'polygenic' in nature. In recent years, 'Genome-wide polygenic scores' (GPS), which integrate information from millions of common variants, have demonstrated improved predictive capacity to identify individuals with increased risk of diseases, including coronary artery disease (CAD), breast cancer, and obesity. As the technology for generating and interpreting GPS improves, a thoughtful approach to reporting complex risk information is critical. OBJECTIVE: In parallel to refining GPS risk prediction algorithms, our team is creating GPS reports that can be returned to various stakeholders, including patients, consumers, and clinicians. Here, we describe a two-phase approach for designing and optimizing a GPS report and describe key findings from phase one: designing a GPS report for CAD. METHODS: In phase one, we reviewed 5 polygenic score reports from commercial laboratories and research studies to identify recurring themes in both text and visual communication of polygenic scores in terms of: numeracy communication, polygenic risk definition, disease definition, and technical specifications. An expert advisory committee consisting of clinicians, genetic counselors, and data visualization developers used these recurring themes as a guide to create a GPS report for CAD. Phase 2 will utilize qualitative and quantitative user testing strategies to (1) assess user comprehension and (2) improve personalization of polygenic score information. Phase two will allow us to evaluate the efficacy of the overall report and refine language and visuals used to describe risk for common diseases. RESULTS: In phase one, two categories emerged as fundamental components to include in a GPS report for common complex disease. At the highest level these components include: (1) The Polygenic Score - (1a) What is a polygenic score? (1b) What does it mean for me? and (2) The Disease of Interest - (2a) What is the disease? (2b) How can I modify my disease risk? While visuals describing polygenic scores tended to adopt a similar approach, specific language used around risk communication varied substantially. CONCLUSIONS:The findings from phase one demonstrate the need for ongoing research that utilize systematic methods for creating and optimizing GPS reports that can be adapted for multiple diseases. This study will have implications for academic and commercial genetic testing laboratories with intentions of returning polygenic score information to patients and consumers.

Notes

Assessment of Current Pretest Chatbots: Is Information Value Neutral?

Kelly C. Donahue, MS, CGC, Jennifer Rietzler, MS, CGC
Katie Stoll, MS, CGC

Genetic Support Foundation, Olympia, WA, USA

Individuals considering genetic testing often have a broad range of questions: What information does the test tell me? How reliable is it? What are the features of the condition(s) included? What will the results mean for my health or the health of my family?

Formal pre-test genetic counseling has been the longstanding framework for helping people find answers to such questions and to help them ponder other points they may not have considered such as: What might your next steps be if your results are positive or negative? Have you thought about other potential risks of genetic testing such as impact on future insurability or employability? Post-test genetic counseling has focused on assisting patients to make the best adaptation to the implications of their specific test result, irrespective of whether it is positive, negative or uncertain.

Given the surge in volume of available genetic tests and the relative small numbers of genetic counseling providers, it has been suggested that pre-test genetic counseling has become a barrier to, rather than a facilitator of, genetic testing. Mass commercialization has led to a large volume of pre-test counseling to fall under the purview of other types of providers that have little knowledge or training regarding the nuances of genetic testing.

Chatbots have been newly launched as a way to fill gaps in pre-test counseling and to increase overall access to genetic testing. They are promoted as a way to enable a larger number of people to make informed decisions that cannot be served by the current genetic counseling workforce. Their use is advocated for as a way to allow genetic counseling providers to focus their efforts on assisting the minority of people who receive positive test results as well as a way to allow non-genetic trained providers to focus on their own field of expertise.

The functionality of chatbots for this purpose has not been studied. American Board of Genetic Counseling website lists a set of questions that are suggested for patients to ask when considering genetic testing. We presented the question set to the genetic pre-test chatbots currently available in the United States and culled the AI simulated communication for word neutrality. Our findings illustrate that, just as in clinical practice, it is important to engineer chatbots to be mindful of the power of word choice and how it may influence the core values of patient autonomy and informed consent.

Notes

Effect of Providing Education about Carrier Results via Web versus Genetic Counselor on the Subsequent Therapeutic Relationship

Lori Erby, Lori Erby, Tyler Wisniewski, Katie Lewis, Les Biesecker, Barbara Biesecker

Lori Erby, Tyler Wisniewski, Katie Lewis, Les Biesecker - National Human Genome Research Institute, National Institutes of Health; Barbara Biesecker - RTI International

The expertise of genetic counselors should be targeted to contexts in which clients are most likely to benefit. One possible delivery model involves education via a web platform with follow-up genetic counseling to assist with adaptation to the information. A prior study of carrier results delivery to healthy adults beyond childbearing years examined post-session outcomes and demonstrated noninferiority of web-based results delivery when compared to delivery by a genetic counselor (GC). The counseling tasks in genetic counseling rely on the development of a therapeutic relationship. Psychotherapy research demonstrates that a therapeutic relationship grows stronger when the counselor and client meet multiple times. We examined whether the therapeutic relationship as assessed by an observer was higher in post-carrier results follow-up genetic counseling sessions when results were previously delivered by the same genetic counselor than when results were delivered via the web. Participants were part of the NIH ClinSeq study. They were first randomized to receive education about their results via a web platform or via a GC and were then further randomized to receive follow-up genetic counseling or not. We rated audio recordings of 73 follow-up genetic counseling sessions using the observer version of the Working Alliance Inventory (WAI-O). Eleven sessions were rated by a second coder, with an inter-rater reliability of 87.8%. T-tests were used to consider differences in WAI-O scores between the two groups who received follow-up counseling. Participants had a mean age of 64 years and were primarily white (96%) and well-educated. The mean therapeutic alliance scores did not differ significantly between the two study arms (education by GC 5.26/7; education by web 5.22/7; t=0.48, p=0.63). Results suggest that the use of a web platform in this specific context did not adversely affect the subsequent therapeutic relationship, but it would be important to consider this in future studies with higher impact genetic test results.

Notes

Futuristic Models of Genetic Counselling

Shivani Nazareth, MS, CGC

Head of Clinical Development, Clear Genetics, 322 Greenfield Ave, CA, USA

Implicit in the pursuit of personalized medicine is the lofty goal of anticipating and preventing disease. As more individuals are sequenced, perhaps even before birth, the notion of genetic counselling as a one-time, isolated interaction will appear shortsighted. Instead, the clinical and psychosocial guidance of a genetics expert will prove to be critical at various stages throughout a person's life. In order to do this effectively, genetic counsellors will have to lend their expertise to the development of centralized management tools, data-driven care plans, and novel approaches to patient and provider education. In other words, the future— including the complete integration of genetics into healthcare— must be built now.

Notes

A family systems approach to genetic counselling; development of interventions"

Rhona MacLeod

"*The psychotherapeutic models most pertinent to the genetic counselling situation are those of brief therapy, crises intervention, and/or family therapy…..Many psychotherapists would view the problems with which counselees come to genetic counselling as life crisis, understandable only in the context of their overall life histories, as individuals, as members of a family, and as members of a specific social group."*
(Kessler 1979)

To what extent are these models relevant in the genomics era? What difference does it make to remember the wider social context within which 'problems' associated with a genetic diagnosis reside? How does this influence the conversations we have with our patients? These questions will be considered in relation to systemic approaches to practice.

Systemic approaches such as narrative therapy (White and Epston 1990), with an emphasis on people's strengths, wishes and ways of resisting the effects of a problem, may be a particularly useful framework for genetic counsellors. Narrative practice views people as multi-storied and is concerned with the question of how we encourage people to tell their stories in ways that make them feel stronger.

Increase in the uptake of genomic testing and the number of people seeking genetic counselling present opportunities to consider new ways of working, particularly around support following a new genetic diagnosis. One option is to realise the potential of group interventions. It is known that families may struggle to communicate genetic risk information leading to partial or non-disclosure of information and the subsequent longer term distress has been well documented. Family therapy and narrative practices have the potential to encourage communication and for families to learn from each other (Mendes et al 2013, Eisler et al, 2016, Stopford et al, 2019).

In a novel programme of work Prof Alison Metcalfe and colleagues conducted a series of focus groups with families impacted by a genetic condition to look at the acceptability of family therapy interventions. The findings were used to develop the Multi Family Discussion Group (MFDG) and in an extension of this work, it was shown that GCs could be trained to deliver the MFDG intervention alongside a family therapist (Eisler et al 2016, 2017).

It seems likely that a plurality of research approaches will be required in the development of systemic approaches to genetic counselling. This could extend to looking at pre and post counselling training programmes for genetic counsellors and effects of counselling supervision on practice.

Notes

Communication of Clinical Uncertainties: A Systematic Literature Review

Biesecker BB, Boyea B, Kulkarni N, Peay HL, Paquin RS, Wheeler AC, Lewis MA

Uncertainties pervade medicine, and genomics is no exception. How patients perceive uncertainty is related to how providers communicate it. Thus, theories on managing uncertain health information include the role of provider messaging. To assess the state of the science, we conducted a systematic literature review on communication of uncertain health risk information. Our aims were to determine the breath and quality of the evidence, identify research gaps and posit evidence-based hypotheses. The search identified 1020 abstracts from PubMed, Web of Science, PsycINFO, Communication Source and Cochrane Reviews published between 1/1/1990 and 6/1/2018. Forty-one abstracts met inclusion criteria: 16 quantitative and 25 qualitative studies. A total of 3656 providers and 4530 patients are represented in the data. Our initial data synthesis focused on provider communication, deferring patient perceptions for subsequent analysis. Among the quantitative studies, one was of high quality-assessing an intervention to enhance provider communication of uncertainty. Nine were observational studies of medium quality. Seven were self-report and of lower quality. Among 25 qualitative studies, 12 were analyses of recorded clinical sessions of high quality, ten were self-report interviews, three were focus groups and one was an ethnographic study. Communication and management of clinical uncertainties were observed or reported between providers and patients in primary care, advanced cancer care, genetics, obstetrics, oncology, cardiology, and emergency medicine. Research gaps exist in intervention research. Evidence revealed providers' avoidance of communicating clinical uncertainties, recognition of the challenges in communicating uncertainties, decisions to provide information regardless of uncertainties, and relational factors that led to expressions of uncertainty. These studies helped to generate hypotheses for future testing. High-quality research is needed to inform best practices in clinical medicine, particularly in genomics. In the meantime, this evidence can inform clinical practice in communicating uncertainty as further evidence is generated.

Notes

Communicating Effectively about Cancer Genetics with Patients of Low Health Literacy

Galen Joseph

Introduction: Research conducted with patients of limited health literacy suggests a mismatch between information provided by genetic counselors and information desired and needed by patients. Such information gaps have the potential to exacerbate existing health disparities as access to genomic medicine expands. As part of the NHGRI Clinical Sequencing Evidence-Generating Research (CSER2) consortium, the Cancer Health Risk Assessments Reaching Many (CHARM) study is enrolling ~800 adults at risk for hereditary cancer into a randomized controlled trial. The trial compares exome sequencing results disclosure via usual care genetic counseling and a literacy-focused genetic counseling approach.
Methods: Counselors in the literacy-focused arm received training (six 1-hour sessions) and ongoing support through weekly case reviews to use evidence-based techniques for effective communication with individuals of limited health literacy (e.g. plain language and teach back); counselors in the usual arm use traditional genetic counseling methodology and did not participate in any specialized training for the study. Audio recordings of the counseling sessions are assessed to ensure fidelity to the two counseling approaches. We hypothesize that the literacy-focused approach will be non-inferior to usual care genetic counseling and may be more effective in terms of participant satisfaction and engagement with counseling, perception of the communication, and understanding of and adherence to recommended care, particularly for those of limited health literacy. These outcomes will be measured through patient surveys and qualitative interviews, evaluation of the session recordings, and medical record review.
Results: Preliminary results, based on the completion of 224 results disclosure sessions to date, review of 50 audio recorded sessions and counselor case conference discussions demonstrate: 1) counselors' ability to implement the modified counseling with fidelity to the protocol; 2) the utility of teach-back for identifying opportunities for clarification and further education; and 3) the utility of direct questions and recommendations over non-directive counseling for counselor's ability to tailor the communication and engage the participant.
Discussion: As genomic medicine expands to include population health, it is reaching an increasingly diverse patient population. Communication of cancer risk and prevention recommendations needs to be accessible and appropriate to individuals of all literacy levels. The results of this trial will inform results disclosure for diverse patients in the precision population health setting, and have implications for the training and practice of clinical genetics professionals.

Notes

Preparing a genetic counselling workforce for the future in Australasia

Alison McEwen

Head of Genetic Counselling, Graduate School of Health, University of Technology Sydney, NSW 2007 Australia

Advances in technology, including genomic sequencing, robotics, big data, precision medicine and artificial intelligence are dramatically changing the field of genetics, bringing enormous opportunities for healthcare and genetic counsellors. Current genetic counselling students will graduate into a world of work more diverse and uncertain than any previous generation. Preparing the future genetic counselling workforce is an exciting challenge, both for the profession and for educators.

Australia is a large country with a diverse population. Health care services are state funded. The dominance of the medical model creates an imbalance between doctors and other health professionals including in genetic services. In this environment, the voices of the genetic counsellors struggle to be heard. The Human Genetics Society of Australasia is addressing this problem through the introduction of regulation to enhance the professional standing of genetic counsellors. Such significant change disrupts the status quo, creating uncertainty, fatigue and the perception of threat.

Against this backdrop, we at UTS are building a Discipline of Genetic Counselling, set within a graduate school of health, alongside other allied health professionals. We are delivering our program using a three-pronged approach to learning - online, live and online and in person - reaching across the country and beyond. Without the constraints of a medical school, we are able to foreground the inherent knowledge, skills and values of genetic counselling, to move away from a focus on testing, and to educate competent, person-centred, research enabled genetic counsellors who are ready to enter the workforce in roles as yet unknown.

We ask our students to be courageous, to step into a deeper exploration of their own identity, beliefs, understanding and experiences of oppression, power and privilege. We are creating ‘brave spaces’ - that foreground the need for courage to create genuine dialogue - in which our students can interact authentically with us and with one another to facilitate the development of effective client-centred genetic counselling practice.

As educators, we have a responsibility to prepare students to embrace the uncertainties, challenges and potential of the genomic era, to seize the many possibilities that lie ahead, and to avoid limiting their thinking and vision. We are pushing boundaries in an already tumultuous environment, challenging ourselves and our students to remain always open to possibilities. Equipping students with open eyes and listening ears may be the single most important thing we can do to prepare the genetic counselling workforce of the future.

Notes

International Genetic Counseling: What do Genetic Counselors do?

Jon Weil[1], Laura Hayward[2], Tina-Marié Wessels[3], Christine Patch[4], Kelly E. Ormond[2]

[1]Department of Biological Sciences, California State University, Stanislaus
[2]Department of Genetics, Stanford University School of Medicine, Stanford, California
[3]Division Human Genetics, University of Cape Town, Cape Town, South Africa
[4]Clinical Lead for Genetic Counselling, Genomics England, Queen Mary University of London, London

The purpose of this study was to investigate similarities and differences in reported genetic counseling practice internationally. A survey was developed using literature, the U.S. American Board of Genetic Counseling practice analysis (2012) and the Accreditation Council for Genetic Counseling practice based competencies (2015), and recommendations from international colleagues. 5,600 genetic counselors were invited by email: 200 responses from 28 countries met inclusion criteria. Five countries had more than 10 responses, (82% of total): USA 78, Canada 29, Japan 26, Australia 16, France 15. There were similarities across all five countries, with >90% respondents from each reporting that many components of a genetic counseling session are part of their role including: evaluating referral information (90%), reviewing medical records (97%), pedigree analysis (95%), identifying family members at risk (94%), educating patients about basic genetic concepts (92%), providing details about testing choices (90%), providing details about testing results (90%) and recognizing psychosocial factors that may affect the counseling interaction (90%). Less than 40% of respondents from each country reported their role included components of medical history (range 2%-37%) and ordering tests in counselor's own name (27%). We also found differences among countries: Respondents from Japan reported a lower rate of documenting ethnicity and consanguinity; Australian respondents reported a higher rate of arranging pre-session tests; and respondents from Japan and France reported lower rates of agenda setting, cross-cultural assessment and documentation, and interdisciplinary collaboration. There were 42 responses from 19 other countries in Europe, Asia, Africa, Middle East and South America (1-4/country). Two items reached >80% endorsement: evaluating referral information and explaining genetic testing outcomes. Twelve had >70% endorsement: reviewing medical records, eliciting patient concerns, pedigree analysis, integrating medical-genetic information, identifying at risk individuals, genetic education, explaining testing options, facilitating decision making, discussing risks and benefits and emotional anticipatory guidance. The most highly endorsed components in the five higher-response countries all received agregate 64-83% endorsement in these countries. Generalizations about international practice patterns are limited by the low response rate. However, our study, which is to our knowledge the first to systematically compare the clinical practice of genetic counselors in different countries, identified a common core of practice defined by highly endorsed components as well as those of limited frequency. We also found differences among countries that can be related in part to differences in health care systems, cultural norms and the historical development of genetic counseling in each.

Notes

Developing a nationally agreed cross-professional competency framework to facilitate consent for genomic testing

Amanda Pichini [1,2], Anneke Seller [1], Michelle Bishop [1]

[1] Genomics Education Programme, Health Education England, Birmingham, United Kingdom
[2] Bristol Clinical Genetics Service, St Michael's Hospital, Bristol, United Kingdom.

Building on current medical practice, genomic medicine highlights unique considerations with regards to consent including addressing the needs of different family members, data sharing protocols and feedback of results. England is implementing a national Genomic Medicine Service leading to the utilisation of genomic testing across a growing number of specialties. This has resulted in a requirement for workforce development around the consent conversation, which has significant implications for genetic counselling practice. Genetic counsellors are central to the effective implementation of genomic medicine, with skills in communicating genomic information. As testing is increasingly requested by non-genetic healthcare professionals, genetic counsellors will also play a crucial role as educators based on their expertise and as part of multi-disciplinary teams. To support this, the Genomics Education Programme has developed a nationally agreed cross-professional competency framework which outlines the knowledge, skills and behaviours required to facilitate consent for any genomic test.

The methodology for reaching consensus on this framework is founded on the nominal group technique. An initial framework was developed based on existing literature and experience of the authors. A one-day expert consensus meeting reviewed clinical scenarios in iterative rounds, mapped themes to the framework and voted on areas of inconsistency. A revised framework was open for consultation with individual healthcare professionals, professional bodies and Royal Medical Colleges before being finalised. Feedback was also gained from rare disease and cancer patient communities to ensure that the patient narrative was incorporated into discussions about genomic testing.

Evidence-based competencies are an important basis to support the responsible delivery of genomic medicine. This framework can assist clinical leaders, including genetic counsellors, in identifying the training needs of the healthcare professionals they work with. For those delivering education, the framework provides a comprehensive foundation to structure the development of training such that the consent conversations around genomic testing can be delivered in a consistent manner across specialties. In addition, these competencies can be used as a reference to evaluate how consent is being facilitated in different speciality areas to enhance the delivery of genomic medicine.

Notes

Patient and counselor perceptions of a telehealth genetic counseling model. Can high efficiency co-exist with high satisfaction?

Kiley Johnson, MS, LCGC, Jill Davies, MS, CCGC

GeneMatters, LLC

Introduction: The growth in demand for genetic counseling requires improved efficiencies, but not at the expense of patient care or genetic counselor (GC) job satisfaction. We sought to understand the impact of a highly efficient telegenetics platform and delivery model (GeneMatters) on patient and genetic counselor satisfaction.

Methods:
Genetic counselors conducted case prep and follow-up using our telegenetics platform automation tools and specialized administrative support. Direct and indirect time spent for each patient case was tracked. From 2/2018 to 7/2019, we collected satisfaction survey data from a subset of patients (N=245) who received telehealth counseling, provided an email address and responded to rate their experience. During this same timeframe, GCs completed a satisfaction survey, rating multiple facets of their role, patient interaction and work environment, using the same language and rating scale as the 2018 National Society of Genetic Counselors Professional Status Survey (NSGC survey) for an industry benchmark, where applicable. Results were compared to GCs in the NSGC survey who provided direct patient care.

Results: Patients highly rated their genetic counseling experience: 93.8% strongly agreed that there was enough time to discuss everything that needed to be covered. 90.1% strongly agreed that they understood the information being provided by the GC. 96.3% strongly agreed that the counselor listened to and supported them. Overall, 95.5% of patients would recommend GeneMatters to a friend or colleague. GC survey results also showed high satisfaction. GeneMatters GCs averaged 34 minutes of direct and <10 minutes indirect time for oncology and 22 minutes direct and <9 minutes indirect for prenatal, across pre- and post-test counseling. At this rate, they are able to counsel between 32 and 50 patients/week, compared with an average of 17 patients/week in the NSGC survey. 100% of GeneMatters' GCs were satisfied/very satisfied with the number of patients/cases, compared with 74% in the NSGC Survey. 100% were satisfied/very satisfied with the technology available to complete their work (no NSGC comparison) and 100% were satisfied/very satisfied with the administrative support available to them compared with 61% in the NSGC survey.

Conclusion: Providing administrative and technological support allows GCs to maximize time spent on direct patient care by reducing indirect time, resulting in high GC and patient satisfaction. Our results demonstrate support for a telehealth genetic counseling delivery model and platform to increase capacity for patient care.

Notes

Poster Presentations

Reflections on using Laboratory Genetic Counsellors in the Genomic Era

Irene Abreu-Rodríguez, Irina Royo, Albert Torrents, Héctor San-Nicolás, Laura González, María-José Roca, Beatriz Rey, Joana Fortuño, Cristina Gómez, Xavier Maciá, Mireia Calvo, Albert Ferrán, Cristina Camprubí-Sánchez

Reference Laboratory- Genetics (S.A.)

Knowing that we find ourselves in the Genomic Era, where researchers and clinicians have started to use the knowledge of genomics to improve health; the presence of genetic counsellors are becoming increasingly necessary within laboratories themselves. Currently, the demand in both public and private centers for exomic and genomic tests are showing up the importance of conducting genetic counselling before and after the genetic analyses itself. Laboratory genetic counsellors are well trained to understand different "languages" (medical terms and lab tests) in order to be an adequate and valid interlocutor between the laboratory and the clinician.
In the Genomic Era, which variants will be informed, combined with how we are going to report on them, must be at the core of respecting the autonomy of the patient. It must be based on guidelines and an specialized informed consent document. The role of a laboratory genetic counsellor in a patient-focussed system avoids, among other things, the application of unnecessary genetic studies, as well as facilitation of a personalised report corresponding to choices made by the patient.
Our center is a genetic/genomic test accredited laboratory, putting into practice laboratory-driven genetic counselling. For that, we model our work flow experience with the challenge on the plane of communication and interaction between the clinician and the laboratory. After receiving a requested form, we pre-analyse the clinical data and the genetic test that was ordered. The same process is followed before results gets sent out. In case of inconsistency, the inherent skills of the counsellor is applied even when contacting the clinicians by phone or email. In conclusion, a significant number of genetics tests are being changed into useful analyses. Based on feedback expressed by clinicians, contacting them directly, gives them the confidence to communicate the information/results with the patient. This model of work flow applies counselling skills, particularly in the genomic era, not only assists with improving the diagnostic process for families with a suspicious genetic condition, moreover, in the past two years, our team have had to double the number of staff who work in genomic testing.

Setting up Genetic Counsellor lead clinics for recurrent neurodevelopmental susceptibility factors – the journey.

Elizabeth Alexander, Michelle Bottomley

Manchester Genomic Medicine Centre, Manchester University Hospitals NHS Foundation Trust. Saint Mary's Hospital, Oxford Road, Manchester. M13 9WL

We present our experience of developing genetic counselling clinics for families where a recurrent copy number variant described as a neurodevelopmental susceptibility factor has been found following microarray testing carried out for congenital and/or developmental reasons. These results are associated with increased risk of developmental delay, learning difficulties and emotional/ behavioural difficulties, which range from autism to mental health problems (Kaminsky et al, 2011, Kirov et al, 2015 and Sahoo et al, 2011). These findings can be de novo but are often found to be carried by a parent, who may or may not present with associated difficulties.

Over a period of three years our department has moved from these patients being seen exclusively in consultant lead genetics clinics to the majority now being seen by genetic counsellors (GCs). Initially two GCs worked closely with medical genetic colleagues to establish GC lead clinics expanding to involve a number of GCs across the department incorporating similar referrals into their clinics. There are many interesting elements to this process which has moved the focus of the appointment from a diagnostic/medical approach to a genetic counselling one.

We describe this evolution, how we met the challenges of reassuring medical colleagues this was safe practice and the routes taken to train and support the wider team. This involved working alongside medical colleagues to develop medical guidelines and carrying out a number of audits looking at the nature and management of patients seen in these clinics.

We discuss cases to illustrate our perception of the strengths and potential hazards of a genetic counselling model versus a medical approach with these families. While this is an approach based on the fundamental principles of genetic counselling, the nature of these results (susceptibility factors with much variability, little predictive utility and some medical considerations) has meant developing a specific approach; encompassing the family story, multi-factorial genetics and incorporating discussion around mental health. We also describe our exploratory steps to broaden our approach by establishing links with a psychiatrist running social communication clinics in our trust and the experience of running two narrative groups to help parents recognise existing resilience and sources of support.

Our experience of genetic counselling in the context of susceptibility factors found following broad level genetic testing carried out in mainstream care, is not only relevant for this group of patients but provides a model for the relevance of genetic counselling in the genomic era.

Impact of adding a Clinical Genetics Team in the care of Breast Cancer Patients at a Tertiary Care Center in a Low Middle-Income Country (LMIC) setting

Ms. Fizza Akbar

MSc. Genomic Medicine

Background: Offering genetic counselling and testing in suspected hereditary breast cancer, is standard of care, and best done through a Multidisciplinary Team approach (MDT). Pakistan has the highest incidence rate of breast cancer in Asia, but little is known about the underlying genetic factors in our population. Being a Low-middle income country (LMIC) with limited accessibility to genetics professionals, most patients with breast cancer are not offered genetic counselling or testing. We added a geneticist and genetic counsellor to the Breast Cancer MDT and assessed the impact on patient care at our institution.

Objective: To compare the total number of genetic referrals and uptake for genetic testing before and after the addition of the Genetics team to the Breast Cancer MDT.

Methods: 3-year retrospective review at The Aga Khan University Hospital, Karachi from June 2016 to July 2019.

Results: A total of 2,111 patient records were reviewed. Total number of new patients presenting to the Oncology/Breast Surgery department remained same throughout the study at an average of 59 per month. During an 11-month period from June 2016 to April 2017, no genetics referrals were made. Over the next 13-months between May 2017 and June 2018, a total of 37 referrals were made, averaging 3 per month, for a total of 5.1% of breast cancer patients being referred for genetic counselling. After adding a Geneticist and Genetic Counsellor to the Breast Cancer MDT, with their presence being mandatory at the weekly breast tumour board meeting, the next 11 months (July 2018 till June 2019) saw an increase in referrals to a total of 151, averaging 14 new patients per month, an increase in referral rate (14/59) to 23.5% vs. 5.1% ($p<0.05$). Genetic testing uptake rate also improved, from 48.7% (17/37) to 68.2% (103/151), before and after this intervention. This led to the creation of a database of germline mutations causing hereditary breast cancer in our population. Current data indicate that out of the patients on whom genetic testing was sent, 20% (21/103) showed pathogenic variants, 33% (34/103) showed variants of uncertain significance and 47% (48/103) showed negative results.

Discussion: Adding a Genetics team to the Breast cancer MDT proved highly successful in increasing the referral rate and patient uptake for genetic testing, improving access to genetic counselling even in an LMIC like Pakistan. A population specific hereditary breast cancer database promises to improve care of patients and families with breast cancer.

The assessment of ethical aspects of the genetic counseling efficacy in Russian Federation

Elena Baranova1, Vera Izhevskaya 2

1 Federal State Budgetary Educational Institution of Further Professional Education Russian Medical Academy of Continuous Professional Education of the Ministry of Healthcare of the Russian Federation, Moscow, Russian Federation
2 Research Centre for Medical Genetics, Moscow, Russian Federation

Genetic counseling is getting widespread due to an increase in the technical possibilities of genetic testing. The ethical aspects of genetic counseling in Russia are poorly investigated. The present research will be dedicated to these problems. We plan to evaluate a number of ethical problems arising during the reception of a geneticist and for this, we have developed a special questionnaire. According to the results of a pilot study on 20 questionnaires, the questionnaire is refined and contains 28 questions. The questionnaire now contains a number of questions that allow you to identify age, gender, marital status, religion, and other parameters relevant to the consultant himself. Also, in the questionnaire, there are questions about the number of patients on admission per week for consultation, how many of these patients are pregnant women with a high risk of pregnancy pathology, paid admission or free. The section related to the evaluation of ethical problems begins with questions whether there are people with hereditary diseases, mental disorders and other features in the family of the counselor himself. Further, in the questionnaire are present hypothetical situations in which the doctor is invited to make a choice and explain it.
We also will ask our respondents about future perspectives in genetics and the possibilities of ethics problems due to that. In addition, we suggest clarifying their vision of the education and health care situation in genetics. This work is supported by the Russian Science Foundation under grant 19-18-00422.

FaceMatch – using facial recognition technology to help parents searching for a diagnosis for their child

Jackie Boyle 1, Brian Lovell 2, Carlos Riveros 3, John Attia 4, Anna Hackett 1, Sheridan O'Donnell 5, Anne Baxter 5, Ben Kamien 6, Tracy Dudding-Byth 1.

1. The NSW Genetics of Learning Disability (GOLD) Service, Newcastle Australia.
2. The University of Queensland, Brisbane, Australia.
3. Hunter Medical Research Institute, Newcastle, Australia.
4. University of Newcastle, Newcastle, Australia.
5. Hunter Genetics, Newcastle, Australia.
6. Genetic Services of Western Australia, Perth, Australia

FaceMatch (facematch.org.au) uses advanced computer vision technology to accurately match facial images with the aim of improving diagnostic rates and discovering novel intellectual disability (ID) genes.

Despite advances in genomic technology, around 60% of children with intellectual disability remain without a genetic diagnosis. An estimated 50% of these children have facial features which may provide a clue to their diagnosis.

As individuals with the same genetic condition often share similar facial features, matching faces can help identify people with the same rare genetic condition. Currently, the process of finding another child with similar features is inefficient, as clinicians compare photographs at conferences or enter written descriptions and DNA variant data into global phenotyping databases. Written clinical descriptions of a face, even using Human Phenotype Ontology terms, are subjective and prone to human variation.

We will present our pilot data showing that within 10 syndromes, FaceMatch accurately matched faces of individuals with the same genetic condition more than expected by chance ($P < 0.00001$), outperforming senior clinical geneticists who participated in the study. We will also present project progress, challenges and opportunities to date.

Participation in FaceMatch can be initiated by a parent or their clinician, empowering parents, in partnership with their clinician, to play an active role in finding a genetic diagnosis for their child.

FaceMatch can also assist clinicians with interpretation of variants of uncertain significance within known or candidate ID genes. As more pathogenic variants causing syndromic ID are identified and the phenotypic spectrum of each condition broadens, it will become increasingly challenging for clinicians to have a comprehensive knowledge or experience of the phenotypic spectrum across age, gender, severity and ethnicity for all monogenic causes of syndromic ID.

As with all artificial intelligence, FaceMatch has the ability to store and match more images than the human brain. It has potential application for clinicians with limited access to specialist dysmorphology services or advanced genomic technology, particularly in rural areas or developing world countries.

A psychoeducational intervention supporting patients with a new diagnosis and/or genetic carrier status for an inherited cardiac condition (PISICC)-a feasibility study protocol

Bueser, Teofila*, Patch, Christine*; Rowland, Emma*; Carr-White, Gerald**; Metcalfe, Alison***

*Kings College London, Florence Nightingale Faculty of Nursing, Midwifery and Palliative Care, London, UK; **Guy's and St Thomas' NHS Foundation Trust, London, UK; ***Sheffield Hallam University, Sheffield, UK

Background
The prevalence of inherited cardiac conditions (ICCs) is significant affecting 1:500 of the population. It is the leading cause of sudden death in those under 40 which may be the only manifestation of the disease. The disease entities within ICCs include inherited cardiomyopathies and arrhythmic syndromes; which are mainly autosomal dominant conveying a 50% risk for first-degree relatives. Cardiac screening and/or predictive genetic testing is recommended and if relatives are affected or carriers for ICCs, apart from the psychosocial impact, decisions are to be made regarding lifestyle, management and communication to other family members (Ackerman et al. 2011). To date, no psychoeducational intervention has been developed to specifically support patients who have a new diagnosis or genetic carrier status for an ICC.
The Psychoeducational Intervention Supporting patients with a new diagnosis of an Inherited Cardiac Condition (PISICC) model was developed utilising the first stage of the Medical Research Council (MRC) framework for developing and evaluating complex interventions (Craig et al. 2008). PISICC incorporates the findings of a systematic review and an in-depth analysis of patient interviews on the experiences of undergoing cardiac screening and/or predictive testing for ICCs; underpinned by self-determination theory.
Purpose
To determine the feasibility and acceptability of the PISICC model. Method
The PISICC feasibility study is the second stage of the MRC Framework and will be conducted in 2 phases using mixed methods. Phase 1 is an uncontrolled clinical trial of the PISICC model delivered in a group setting of up to 3 groups with a maximum of 10 participants/group. Patients 16 and older with an ICC diagnosis or carrier status given within 6 months are eligible. The main outcomes are key feasibility measures related to trial procedures and delivery. The secondary outcome is the suitability of clinical outcome measures assessed through validated questionnaires, given at baseline and at 3 months' follow up, pertaining to the degree of self-determination, autonomy support and competence (Perceived Competence Scale, Perceived Choice and Awareness of Self Scale, Health Care Climate Questionnaire), as well as, allaying stress and anxiety (Cardiac Anxiety Questionnaire) associated with the intervention. Phase 2 is a nested qualitative component comprised of 7 semi-structured interviews with trial participants to gain insight on their experience with study; and suggestions for improvements for a definitive psychoeducational intervention model. Interviews will be digitally recorded, transcribed and will undergo thematic analysis.

Case series: Interpretation of somatic tumour test results and the impact on genetic counselling practice

Stephanie Burcher1, Kelly Kohut1, Dr Helen Hanson 1, 2 and Dr Katie Snape 1, 2

1 South West Thames Regional Clinical Genetics Service, St George's Hospital, London, United Kingdom
2 St Georges University of London, London, United Kingdom

Somatic tumour testing is increasingly becoming a routine part of clinical management for cancer patients, with a trend for more genes being tested via panels to guide treatment options and inform clinical trial eligibility. Finding a somatic mutation in a cancer predisposition gene increases the likelihood that the variant may also be present in the germline (Turnbull et al, 2019); however, somatic testing in cancer patients usually takes place in the absence of paired germline testing. It is essential that the clinician requesting a somatic genetic test is able to accurately interpret the result, understand the difference between a somatic and germline test result and know how and when to refer a patient into inherited cancer services, whilst ensuring that they do not raise undue anxiety over results which may be confirmed as tumour-only findings.

The South West Thames Regional Genetics service has identified six referrals for patients and their relatives who have undergone somatic tumour testing and mistakenly believe they carry a germline mutation. Some clinicians/patients also informed family members who then sought testing, which is unnecessary and distressing if the mutation is acquired and not hereditary.

We will present a case series of patients who were referred due to misinterpretation of somatic tumour test results, including:

- A patient who was referred for bowel screening after a somatic APC mutation was identified in her mother's bowel tumour.
- A patient who was told her children might be at risk of Li-Fraumeni Syndrome due to a somatic TP53 variant found in her deceased husband's tumour.
- A patient who was referred for predictive BRCA2 testing following identification of a somatic BRCA2 mutation in a relative with pancreatic cancer.

Additionally, we will present other examples where inappropriate screening and familial genetic testing were recommended, due to misinterpretation of somatic results by clinicians and patients.

We will discuss the management of these cases with reference to the ESMO Precision Medicine Working Group recommendations (Turnbull et al, 2019), highlight the need for implementing clinician and patient education programmes, including an outline of the education programmes we have developed and explore the impact somatic tumour testing may have on genetic counselling practice in the future.

Reproductive preferences in parents with lived experience of caring for their children with intellectual disability.

Louise Christie2, Radhika Rajkumar1, 3, Jackie Boyle2, Kristine Barlow Stewart8, Deborah Schofield1 ,Morgan Rice1, Lucinda Murray2, Melanie Leffler2, Elle Martin2, Rupendra Shrestha1, Owen Tan1 , Luke Rynehart1, Sarah West1, Nadine Kasparian5, 6 Tony Roscioli4,7,Michael Field2

1GenIMPACT: Centre for Economic Impacts of Genomic Medicine, Department of Economics, Faculty of Business and Economics, Macquarie University, North Ryde NSW, Australia
2Genetics of Learning Disability (GOLD) Service, Department of Clinical Genetics, St Leonards NSW and Hunter Genetics, Waratah NSW, Australia 3The Garvan Institute of Medical Research, Darlinghurst NSW, Australia 4Sydney Children's Hospital Network, Sydney NSW, Australia 5Heart Centre for Children, Sydney Children's Hospital Network, Sydney NSW, Australia 6School of Women and Children's Health, UNSW Medicine, University of New South Wales, Randwick NSW, Australia 7NeuRA (Neuroscience Research Australia), Sydney NSW, Australia 8Northern Clinical School, Faculty of Medicine and Health, University of Sydney, NSW 2006, Australia

Introduction: The study of the economic and psychosocial impacts of caring for families affected by intellectual disability provides insight into the lived experience of parents caring for children with moderate to severe intellectual disability (ID). One section of the study explores the reproductive preferences in a cohort of parents previously seen by our genetic service.
Method: Counsellor delivered survey, exploring recurrence risk perception, previous reproductive experiences, and preferences regarding carrier testing and reproductive options.
Results: The majority of participants were women 78/89 (88%). The perception of recurrence risk was high with 48/89 (53%) estimating their perceived chance of having another affected child as 50-100%. The majority of participants 64/89 (72%) reported they would have had carrier testing, had it been available at the time they were having children.
Just over half of participants; 47/89 (53%) would have prenatal testing. Of these, 24/47 (51%) would consider ending an affected pregnancy whilst the remainder would continue an affected pregnancy or were undecided. Overall, 31/89 (31%) would continue an affected pregnancy. A large number of participants, 56/89 (63%) would consider having preimplantation genetic diagnosis.
Most families, 62/89 (70%) did not have a genetic diagnosis for the ID in the family. Of the 27/89 (30%) families who had a diagnosis for the ID, the majority (89%) had received the diagnosis after completing their family and/or when their children were adults. Therefore, most participants were answering questions about reproductive preferences from a hypothetical perspective. Three participants who had a genetic diagnosis prior to completing their families, elected to have prenatal testing in subsequent pregnancies. One participant ended an affected pregnancy and other two planned to end the pregnancy if affected but after receiving normal results continued their pregnancies.
There were no significant differences in reproductive preferences based on their perceived risk, the availability of a diagnosis; the severity of ID or the number of affected children.
Conclusion: The most preferred reproductive option for parents of children with ID was preimplantation genetic diagnosis. This study highlights the difficulties parents face when considering reproductive options and especially, in regards to ending an affected pregnancy. A lived experience of caring for children with ID and personal beliefs appear equally important in influencing reproductive preferences.

Managing VUS results in a mainstream setting: learning from a case example

Pooja Dasani, Alice Coulson

Guy's and St Thomas' NHS foundation trust, London, UK

Managing a variant of uncertain significance (VUS) in a clinical setting has always been a challenge. With the advances in technology and genomic testing being carried out on gene panels and whole genome sequencing, there is a higher chance of VUS results.
Interpretation of these for patient management can be complex and literature indicates that healthcare professionals may not feel confident in delivering this result.
Additionally the ACGS-ACMG variant interpretation guidelines highlight the complexity of interpreting VUS results. With increasing amounts of evidence for interpretation, VUS results may be assigned as 'hot' or 'cold' depending on emerging evidence. Variant classification may also change over time depending on current guidelines and evidence.
Case example
We present a case where two BRCA2 VUS results were identified in a woman diagnosed with breast cancer (Patient AB) at 42 years of age. The patient's mother was diagnosed with a breast cancer at 36 years of age. AB underwent risk-reducing mastectomy and complete hysterectomy after discussion with her surgeon who supported this management based on the genetic result. The patient requested genetic testing for her daughter (Patient CD) through the surgeon who was not able to provide this, but advised that they would support risk reducing mastectomy and a complete hysterectomy for her daughter if she was found to carry the same variants. There was a belief in the family that this result explained the family history. There had been an expectation in the family that CD would be offered testing at the age of 18 in order to plan her risk-reducing surgeries.
The family met with a genetic counsellor to discuss genetic testing. They were frustrated and angry that risk-reducing surgical options would not be recommended based on a VUS result. This lead to feelings of mistrust towards the clinical genetics department.
Learning points
This poster will present the complexities of VUS results in families. We will consider how VUS results are presented on a test report, as well as communicated to a patient in a clinical setting. Addressing these factors requires an assessment of training needs in mainstream healthcare professionals who action genomic test results and in particular, improving understanding of variant interpretation.

Patients' and their families' experiences with participation in whole genome sequencing research projects - A systematic review

Varisha Desai,

University of Glasgow; NHS Greater Glasgow and Clyde

Background
Genetic testing has proven its requirement in medicine as a diagnostic, prognostic and predictive tool. With high throughput and low costs, genetic testing is heading towards an era of whole genome sequencing (WGS). Numerous countries have launched individual WGS projects, with the hope to build databases for various populations and to find new personalised treatments. Most of the research, to date, is focused on challenges of interpreting new variants, informed consent and disclosure of incidental and secondary findings. There is limited literature on the perspectives of patients' and families, with undiagnosed genetic conditions, on their journey of participating in WGS projects.
Objectives
This review aims to elicit patients' perceptions, expectations and awareness of WGS by exploring participation in WGS projects. An increased knowledge of patient understanding would optimise genetic counselling conversations when offering WGS in the future.
Search - Methods and Criteria
Relevant electronic databases and reference lists of eligible articles were searched using appropriate search terms. Hand searches of pertinent journals were also carried out. By doing so, studies that reported original results about patients' and their families' experiences after receiving results from participation in a WGS project were included. Literature included were published in English language, in a peer-reviewed journal and used qualitative or mixed-methods data analysis. Studies that focused on the experiences, motivations, preferences and perspectives of participants' or the public who did not have a personal or family history of a genetic or suspected genetic condition were excluded from this review.
Results
8 studies met the criteria. Results showed that the main motivations and expectations of patients' or parents of patients' participating in WGS projects were to end the 'diagnostic odyssey', receive treatment or better management of their condition, and altruism or for the benefit of future generations.
This review emphasises understanding research participants' experiences and expectations of WGS to help inform protocols and standards for WGS implementation as part of routine NHS testing. Having clear guidelines to help patients understand the complexities of WGS and better manage their expectations will make the process more valuable to the patients and their families.
Impact of this study
Through this systematic review of literature, I found that, to date, there is no literature focusing on the Scottish population's perception and expectation of WGS. Hence, a research proposal to explore the experiences of participants of the Scottish Genomes Partnership by qualitative analysis is put forth in this review.

A new approach to Oncogenetic counselling: Triaging the patients

Laura Diskin1, Revital Bruchim1, Varda Nadler1, Tamar Wolf1, Mordechai Shohat1,2

1 Maccabi Healthcare Services, Central Laboratory, Rehovot, Israel.
2 Sackler Faculty of Medicine, Tel Aviv University, Israel.

Summary: Our aim was to improve and facilitate oncogenetic counselling services to as many patients as possible.
Background: Genetic testing of BRCA common mutations (a total of 14 mutations in BRCA 1+2) is available in Israel and is legally covered by the national healthcare services for all patients with breast, ovary or pancreatic tumors, regardless of age and ethnicity. In addition, healthy relatives may seek genetic testing for reassurance, or surveillance recommendations and advice about risk reduction measures if found at high risk. The "Angelina effect" raised awareness of genetic testing, even in the general population. Specialists in genetic counselling and specifically oncogenetics are few, and waiting times for appointments can be as long as a year.
Our approach is based on a "central genetic counselling" program whereby three certified genetic counsellors triage requests for genetic testing and classify the risks according to personal information and family medical history, in Maccabi Healthcare Services (MHS), a large state-mandated healthcare services provider in Israel, responsible for 25% of the population (2.2 million members).
Methods: We generated a simple questionnaire that included questions concerning personal and family history of cancer and demographic information that was sent via SMS to all individuals who requested genetic testing. According to the information received, we assigned either a "high risk" counseling (face to face meeting) within a short time or a "low risk" referral directly to the lab where relevant blood tests were performed at a subsidized price, without need for an initial meeting with a genetic counsellor.
Results: The program was launched in March 2019 and about 1200 requests for testing or counselling have been received to date. As of 30th June 2019, 76% of applications were classified as "low risk" and were referred directly to the lab to test for common mutations. All "low risk" patients with negative results chose not to meet with a genetic counsellor. For patients with positive results and those originally classified as "high risk", mean waiting times for appointments with a genetic counsellor were reduced from 20 months to a maximum of two months, and urgent appointments were arranged within a week.
Conclusion: This new approach has enabled MHS to prioritize appointments and dramatically cut waiting times for patients at high risk. We are very hopeful about this new system and are constantly examining all aspects of this new process in order to improve the service we provide.

AIDING PARENTS IN TALKING TO YOUNG CHILDREN REGARDING ADULT ONSET CONDITIONS: A GENETIC COUNSELLOR'S PERSPECTIVE

E. DIXON,

Great Ormond Street Hospital for Children NHS Foundation Trust The University of Manchester

Parents frequently report the need for more assistance from health professionals in aiding them to have conversations with their children about the genetic condition in the family. Studies have also argued that there needs to be a shared responsibility between professionals and patients in conveying genetic risk information and research has already been conducted into ways to aid this disclosure of information and incorporate this into their practice. However few of these studies have looked into the counsellor's views and experiences of such interventions and ultimately their success in practice is dependent on how confident and prepared counsellors themselves feel in delivering this advice. The purpose of this study was to look at how confident and prepared Genetic Counsellors feel in aiding parents in having genetic discussions with their young children regarding adult-onset conditions by gathering and evaluating their thoughts, views and experiences on this matter. Practising Genetic Counsellors and Clinical Nurse Specialists were sent an invitation via email to participant in an online questionnaire. The questionnaire was divided into three sections: their current practice, the continuing role of the Genetic Counsellor and what could be done to help meet the counsellor's own needs. Twenty-five individuals responded. Results indicated that counsellors felt confident in talking to parents about having discussion about genetic information with their children regardless of the pattern of inheritance or type of condition and that they often raise the subject in clinic even if parents do not. Genetic Counsellors felt that other healthcare professionals could be involved in this process and it should be part of their role to work with them to deliver this support. Many Genetic Counsellors did not consider their training to have fully prepared them but would find it helpful to have some additional training in this area or additional support in their current practice but there was some variation over how best to provide this. Additionally there was no clear indication that training background or years of experience influenced these views and opinions. This study has indicated the need for further investigations into how best to support Genetic Counsellors provide this service to parents in their regular practice. I have a poster prepared on this abstract which I have presented at the MAHSE STP Research Day.

Pilot study of expanded preconception carrier screening in Western Australia – Health professional training and perspectives

Samantha Edwards (1), Royston Ong (1), Georgina Hollingsworth (1), Karen Harrop (2), Denise Howting (1), Sarah Moore (3), Ben Kamien (2,4), Nick Pachter (2,,4), Mark Davis (5), Karen Carpenter (5), John Beilby (5), Nigel Laing (1,5)

(1) Centre for Medical Research, University of Western Australia, Harry Perkins Institute of Medical Research, Perth WA, Australia (2) Genetic Services WA, King Edward Memorial Hospital, WA Department of Health, Perth WA, Australia (3) Rural Clinical School, University of Western Australia, Perth WA, Australia (4) UWA Medical School, University of Western Australia, Perth WA, Australia (5) Department of Diagnostic Genomics, PathWest Laboratory Medicine, WA Department of Health, Perth WA, Australia

Expanded preconception carrier screening (EPCS) assesses the chance a couple will have a child affected with a recessive disorder. With the advent of new technologies it has become more affordable to sequence hundreds of disorders simultaneously. This pilot study aims to determine the requirements for successful implementation of a public health system EPCS program in Western Australia (WA).
In the pilot study, 250 couples planning to fall pregnant will have EPCS for more than 400 severe genetic disorders that are life limiting and/or chronic with onset in infancy or early childhood. Couples are recruited from the Perth and Busselton regions of WA through general practitioner (GP), clinical genetic, and private genetic counselling services. Results are reported as a couple rather than individual carrier status.
All recruiting health professionals (HPs) received training and supporting resources for pre-test genetic counselling and the ability to offer EPCS specific to this pilot study. GPs were observed by the study genetic counsellor during their first pre-test counselling session with a couple. Training and support resources and HP perspectives were evaluated through a series of questionnaires and follow-up interviews.
A total of 30 HPs attended the training presentations and 17 of these went on to recruit between 1 and 60 couples each. Data was collected from the completion of 17 pre-training, 18 end-of-training and 13 post-training questionnaires.
41% of respondents were GPs or GP/Obstetricians, 35% genetic counsellors (GCs) and 24% clinical geneticists. 88% of these HPs routinely provided preconception advice prior to their involvement in this study. However, only 29% had substantial experience discussing EPCS.
Recruiting HPs were predominantly GCs (47%) and GPs (41%). Of the 143 couples recruited in the first 9 months, GCs contributed 69%, followed by GPs with 25% and clinical geneticists with 6%.
Knowledge and preferences of participating couples were also evaluated through a questionnaire administered immediately following their pre-test counselling appointment. Analysis identified the knowledge, preferred methods of training, confidence in offering EPCS, and expected vs actual barriers to recruitment of the different HPs performing pre-test counselling and recruiting to the study, as well as the knowledge and preferences of participating couples following pre-test counselling. Whilst traditionally the responsibility of genetic professionals, we have demonstrated EPCS can be provided by other HPs when given sufficient training and support, and is a preferred option to patients.

Knowledge, Attitude and Practice of Consanguineous Marriage in Sudan 2018.

Fatima Abdelhakam MohamedElmugadam1, Haytham Mohammed Gorshi1, Almigdad Hayder Mohammed1, Murad Elmardi Almak1, Israa Hamza Hussain1, Mohammed Akasha Farag1, Mohammed Suliman Tawar1, Elhami Abdalmuti Ahmed3, Almegdad Sharafaldin Ahmed1, Wadah Osman Awad1, Ahmed Mudawi Musa2

1- Faculty of Medicine, University of Khartoum, Khartoum, Sudan
2- Institute of Endemic Diseases, University of Khartoum, Khartoum, Sudan.
3- UNESCO chair on Bioethics, University of Khartoum, Khartoum,, Sudan

Background. Consanguinity (intra-familial marriage) accounts for over 10% of marriages worldwide and is most common in the Middle East, where it is respected for many socio-economic and psycho-social benefits. Sudan has one of the highest rates of consanguinity, exceeding 50%. This practice increases homozygosity of recessive alleles giving higher risk of early mortality and morbidity of offspring. Our study aimed to investigate the knowledge, attitude and practice aspects on the consanguinity-associated health hazards.
Methods. We collected data from 1089 participants from eight different states in Sudan using convenience sampling and interview based questionnaires. Analysis was done using descriptive and inferential statistics.
Results. Seventy four percent of the participants (803) indicated that consanguinity inflicts health risks on offspring s, 14% (150) refuted any negative health outcomes of consanguinity, while 12% (136) did not have a view about the subject. Sixty four percent of the participants (696) showed non-preference for consanguineous marriages, of these 54% (377) attributed their "non-preference" to the fear of transmitting genetic diseases. Eighty three percent of the participants (908) were willing to undergo premarital genetic testing if affordable.
Conclusion. Most respondents to our questionnaire were aware of the risks of consanguineous marriage on descendants and were willing to take measures to reduce those risks. This indicates a general acceptance and provides a platform for launching nationwide health programs to reduce the burden of familial genetic disorders in Sudan.
Keywords. Consanguinity; awareness; attitude; Sudan

What Happens after the Disclosure of ACMG 59 Secondary Findings? Preliminary Results from an Ongoing Systematic Review

Flavia M. Facio, Julie C. Sapp, Katie L. Lewis, and Leslie G. Biesecker

Medical Genomics and Metabolic Genetics Branch, National Human Genome Research Institute, National Institutes of Health

The American College of Medical Genetics and Genomics (ACMG) released its first guidelines for the return of secondary genomic findings (SF) in 2013, with a subsequent revision in 2017. The main reasons for returning SF include (1) the opportunity to communicate to the individual specific healthcare interventions which have the potential to prolong or preserve life, and (2) the ability to extend such tailored medical care to at-risk family members through cascade testing. We conducted a scoping review to summarize the published outcome data supporting the communication of SF and the downstream actions taken by individual recipients.

We searched six major biomedical databases (CINAHL, Embase, PubMed, Scopus, Web of Science, and PsychInfo) to identify peer-reviewed publications between 01/01/2012 and 30/09/2018 describing 1) how ACMG findings are communicated and/or 2) any outcomes associated with this disclosure, including psychosocial impact, healthcare behaviors, family communication/cascade testing. Abstracts were eligible for inclusion if they reported any component of the process or method of returning an ACMG SF to an individual, if they described any actions taken by recipients after disclosure, and/or any other post-disclosure outcomes. We report here preliminary findings from one author's (JCS) scoping review. Duplication of this process is currently underway through a systematic literature review.

Of 675 non-duplicate records screened, 14 met inclusion criteria. Two studies emphasized efficiencies in the disclosure process and pipeline. Three studies investigated both process and outcomes and two reported similar rates of compliance with recommendations given during disclosure (~70%); rates of family communication were as high as 90% but specific patterns of communication were not reported. Two studies reported on returning results to biobank participants. The remaining studies reported only negative findings (n = 1), were case studies (n = 2), or reported only process data (n = 4). The psychosocial impact of receiving SF was consistently minimal across studies.

Since the release of the ACMG guidelines, few studies have examined the disclosure process or followed recipients to evaluate clinical utility and family communication. The preliminary findings of our review suggest that not all recipients of SF communicate these important results to their doctors or family members, and suggest that a substantial fraction may not be engaging in the recommended healthcare actions which could prolong or save their lives. Those interested in advancing the practice and science of "precision medicine" may look to these early experiences to shape research priorities and policies in this domain.

Cultural adaptation of a booklet for a hereditary cancer telephone genetic counseling intervention with Spanish-speaking Latinas

Katie Fiallos1, Sara Gómez-Trillos,2 Kristi D. Graves,2 Marc Schwartz,2 Beth Peshkin,2 Heidi Hamilton,3 Vanessa Sheppard,4 Susan Vadaparampil,5 Claudia Campos6, Filipa Lynce,2 Andrés Gronda,2 Daniela Morales,2 Federico Palacardo,2 Alejandra Hurtado-de-Mendoza.2

1 Sidney Kimmel Comprehensive Cancer Center, Johns Hopkins University, 2 Lombardi Comprehensive Cancer Center, Fisher Center for Hereditary Cancer and Clinical Genomic Research,3 Georgetown University, 4 Virginia Commonwealth University, 5 Moffitt Cancer Center, 6Nueva Vida.

Background: Evidence-based practices are the gold-standard for health interventions. However, studies show that cultural adaptations can be more effective than unadapted interventions when working with minorities. Latina women have as high a rate of BRCA1/2 pathogenic variants as non-Hispanic white women but have lower rates of uptake of genetic counseling (GC) for hereditary breast cancer. Telephone genetic counseling (TGC) has been shown to be non-inferior to in-person GC. A culturally adapted TGC intervention with a Spanish-speaking genetic counselor could help reduce barriers to GC for Latinas with breast cancer.
Methods: We culturally adapted a booklet previously developed by two of the authors (MS, BP) in a non-inferiority trial for use in TGC with Latina breast-cancer survivors. This booklet contained visual educational material and a guide for the TGC intervention. Using the Learner Verification and Revision Framework, we iteratively adapted the booklet and conducted interviews with 10 Latinas after presenting and explaining the booklet. We then piloted the TGC intervention using the booklet with four Latinas and interviewed them post-TGC. Interviews were coded using Dedoose qualitative software, and themes were recorded.
Results: Fourteen foreign-born, Spanish-speaking Latinas were interviewed. Participants came from eight Latin American countries with an average of 18 years (SD: 7.2) in the U.S. All women had a diagnosis of breast cancer under age 50 and had a current mean age of 48 (SD: 5.5). Acceptability of the booklet was high among both interview and pilot participants who stated it was easy to understand, culturally appropriate, and informative. Participants mentioned the lack of similar resources in Spanish. They particularly liked the analogy of a flan recipe to understand how genetic alterations lead to disease and also appreciated diverse faces on sample pedigrees, reporting it motivated them to consider sharing information with relatives. Pictographs showing cancer risks associated with BRCA1/2 alterations were harder for participants to interpret, and interviewees preferred greater written explanation to accompany these figures. Among TGC pilot participants, general understanding was good, while recall of specific information was mixed.
Conclusions: Learner verification and revision was effective in the cultural adaptation of a booklet for use in TGC with Latinas in the U.S. This process could be used to adapt interventions for other cultural groups facing barriers to GC. Future studies are needed to determine whether culturally adapted interventions are effective in reducing these barriers.

Determining Hereditary Cancer Risk from Somatic Genomic Profiling: A Challenging Clinical Case

Andrea Forman, MS, CGC, Catherine Neumann, MS, CGC; Michael Hall, MS, MD

Fox Chase Cancer Center, Philadelphia, PA, USA

BACKGROUND
Molecular profiling of tumors is a growing area of precision medicine and treatment of cancer. In recent years, tumor-specific testing has been sought by treating oncologists to guide therapy decisions, while hereditary cancer risks identified through blood or saliva samples have focused on screening, prevention, and family risk implications. Notably, the majority of germline variants will be detected in tumor testing and a growing number of these variants are impacting therapies. However, challenges remain in identifying which tumor variants are also present in the germline and which are new mutations in the tumor (somatic only). Tumor testing may miss germline variants through loss of heterozygosity, variant reversion, and large deletion/duplication undetectable by most commercial laboratories. Here we present a case study demonstrating the failure of tumor testing to diagnose a germline gastric cancer risk.
CASE DESCRIPTION
Patient A is a 54 year old man diagnosed with Stage IV diffuse gastric adenocarcinoma. Somatic tumor testing found a variant of uncertain significance (VUS) in CDH1. Germline CDH1 pathogenic variants are associated with Hereditary Diffuse Gastric Cancer (HDGC) and are found in 1-3% of diffuse gastric cancer (DGC) cases. Somatic variants are found in ~12% of DGC cases. The patient met with a genetic counselor for assessment of his medical and family history and consideration of germline testing and was consented to the Fox Chase Cancer Center Risk Assessment Program (RAP) registry (IRB# 09-831). While the patient did not meet clinical testing criteria for HDGC, he agreed to pay for testing in order to receive future clinical updates on his CDH1 VUS, should it be confirmed in the germline.
Germline results were negative for the somatic CDH1 VUS, but identified a new pathogenic variant in CDH1 not previously reported through somatic testing. This new variant was noted to involve a large deletion that may not have been detectible through the somatic testing laboratory's processes.
CONCLUSION
Limitations in tumor genomics sequencing continue to provide challenges in identifying germline mutations through somatic testing. In additional to technical limitations, recent studies suggest that 1/3 of pathogenic germline variants found on paired somatic/germline testing do not meet clinical criteria for testing. The discovery of this CDH1 variant has allowed for cascade testing of several family members that otherwise could have been missed, highlighting the importance of combining somatic genomic profiling with appropriate genetic counseling and testing in identifying hereditary cancer risks.

Expanding the role of a genetic counsellor to assist with variant analysis in the Victorian Undiagnosed Disease Programme

Lyndon Gallacher 1,2, Tiong Tan 1,2, John Christodoulou 1,2, Ivan Macciocca 1,2, Sue White 1,2

1. Victorian Clinical Genetics Services, Murdoch Children's Research Institute, Parkville, Victoria, Australia 3052
2. Department of Paediatrics, University of Melbourne, Parkville, Victoria, Australia 3052

The Victorian Undiagnosed Disease Program (UDP-Vic) in Australia utilises advanced genomic sequencing and analysis as well as functional studies to establish a genetic diagnosis in children for who previous clinical testing has been inconclusive. Our current diagnostic yield for this group of patients is 40%. Genetic counsellors play a key role in project management for genetics and genomics research initiatives around Australia. In addition to managing consent, enrolment, sample collection, record keeping and psychosocial support for patients in UDP-Vic, we have expanded the role of the genetic counsellor to include first pass genomic data curation for a subset of patients being sequenced through the program. We do this with the goal of expanding the highly skilled, in-demand workforce required for variant interpretation, thereby reducing pressure on clinical geneticists in the research area, and freeing up analysis bottlenecks within our project. We will describe the training and approach to analysis conducted within UDP-Vic while demonstrating how genetic counsellors are well equipped with clinical and analytic skills to contribute to this area in the genomic era. Finally, we will describe the genetic counsellor-led qualitative research being undertaken within the programme, which aims to ensure safe service provision for the families of both diagnosed and undiagnosed children.

The difficulty of genetic counseling in pre-conceptional testing

M. Carmen Garrido-Navas, M. Jose Serrano

GENYO- Centro Pfizer - Universidad de Granada - Junta de Andalucia de Genomica e Investigacion Oncologica, 18016, Granada, Spain.

Currently, pre-conceptional genetic testing (or carrier screening) allows healthy couples to identify germinal mutations in genes related with autosomal recessive diseases. Thus, the risk of having an affected child can be estimated based on the presence/absence of gene mutations for the same gene within the couple. For some of these diseases (e.g. spinal muscular dystrophy, Duchenne dystrophy or cystic fibrosis among others), several reproductive options might be offered to those couples carrying germline mutations in the same gene such as pre-implantation genetic diagnosis (PGD) or pre-natal diagnostic tests (PNDT), reducing risks of having affected children. However, genetic counseling can be challenging when mutations in the same gene might be responsible for two different genetic diseases, one of which is not life threatening. An example of this is the CFTR gene, which mutations mainly cause cystic fibrosis (CF) but can also produce a type of male infertility named congenital bilateral aplasia of vas deferens (CBAVD) among other minor diseases.

Here, we present the case of a young couple, both carrying germline mutations in the CFTR gene with different pathogenicity. The couple decided to carry out pre-conceptional testing because the woman's nephew was affected by CF (c.3140-26A>G and c.1521_1523delCTT) and wanted to rule out the possibility of carrying the same mutation than her brother.

After pre-conceptional genetic testing, the woman was found to carry c.3140-26A>G mutation which was shared with her brother and the man was found to carry c.1727G>C, both mutations located at CFTR. The first mutation was pathogenic for CF however the second was benign for CF and only pathogenic for CBAVD. In fact, only one case of both simultaneous mutations has been reported worldwide and was not affected by CF. Despite evidences suggesting that presence of c.1727G>C, in combination with other CF-associated mutation, might not produce CF, the presence of milder CF-like characteristics could not be ruled out; additionally, the aforementioned case of CF in the family difficulted decision-making for this couple who would opt for PGD to avoid presence of the disease.

Although pre-conceptional genetic testing has some advantages on the early identification of risks for a couple, it can also have some drawbacks. This clinical case highlights one of the shortcomings not only related with the difficulty of risk-calculation when mutations in the same gene are associated to two different autosomal recessive diseases, but also with the psychosocial effect of having an affected relative.

Using technology and counselling to weave a web of support for patients and their families

Selina Goodman (1), Professor Ray Jones (1), Dr Leigh Jackson (2) & Professor Heather Skirton(1)

1. Plymouth University, Plymouth UK, 2. University of Exeter Medical School, Exeter, UK

Introduction: Genetic counsellors (GCs) will seek to identify what sources of support an individual can access in their adjustment to a new genetic diagnosis. Online resources are important for information and peer support but patients can feel cautious about the reliability of what they learn there. However utilising different technologies such as apps, chat bots and websites can provide opportunities to augment GC practice and provide information in patient preferred formats that help adaptation and facilitate sharing the diagnosis with relatives.
Methods: Semi-structured telephone interviews (n=14) were conducted with patients who had a high risk of bowel cancer. Results from these interviews combined with the findings of a cross-sectional survey (n=286) informed the development of a website (www.familyweb.org.uk) designed to help support patients in sharing information with their relatives. All participants knew that their diagnosis had implications for their relatives and that they were eligible for bowel surveillance by colonoscopy. The interviews explored patient's experiences of their diagnosis and how they had attempted to share information with their relatives. Recruitment was via NHS hospital services or charity websites in the UK.
Results: Four major themes were identified from the interview data: impact of the diagnosis, the importance of pyschological adaptation to the diagnosis, the need for practical information, and using appropriate methods of communication. The impact of the diagnosis was often profound and often experienced as a burden. Adaptation was indicated by acceptance, seeking information and taking action. Practical information needed to be clearly understandable and in a format that was appropriate, written with a positive perspective about topics that were considered important (e.g. healthy lifestyle, talking to children, accessing surveillance). Methods of communication varied according to which relative was being contacted. All these factors were interconnected and appeared to modify patients' ability to share information with family members.

Conclusion: Listening to individual patient needs and eliciting their specific requirements could aid adaptation to their diagnosis and the dissemination of information within their families, these findings were consistent with the Family System Genetic Illness model. Therefore, counselling skills remain key to successful delivery of genetic counselling. In addition, genetic counsellors can influence the development of new technologies to support patients and their families.

Changing Paradigms: From Prenatal to Reproductive Genetic Counseling

Hayley Green MA, MS, CGC, Michelle Pacione, Ed.M, MS, CGC, Sophie Adams MSc, MS, CGC, Sam Gbur, MS, CGC, and Lori Dobson MS, CGC

Center for Fetal Medicine and Reproductive Genetics, Brigham and Women's Hospital, Boston, MA, USA

Background: Genetic testing is now routine in many areas of medical care, including a significant increase in the number of genetic tests ordered both prior to and during pregnancy. Simultaneously, aspects of prenatal and preimplantation testing options are becoming more routine, and more complex. To adapt to this changing paradigm, the roles of the genetic counselors (GCs) in our Maternal Fetal Medicine (MFM) clinic have focused on higher-level referral indications while reducing routine indications for referral.

Objective: To present our expanded clinic model where GC knowledge and skills are optimized in a reproductive setting, including case examples.

Discussion: Our model showcases the ability to increase access to genetic testing while maximizing the skill set of the genetic counselors. For routine indications, we educated OBGYN providers to provide patient counseling for aneuploidy screening and pretest carrier screening. These efforts were supported by genetic education opportunities, detailed pretest counseling patient aids and consents and clinical support from the genetic counseling team. GC patient slots increased from 60 to 90 minutes and shifted from 20 to 14 patients per week for full time GCs. As a result, GCs provide more robust care for families with positive test results, fetal anomalies and preimplantation testing. Our services expanded with increased referrals for PGT counseling and establishment a cord blood genetic testing program for families who have declined prenatal diagnostic testing.

Due to our specialization in both PGT and prenatal counseling, we can help families throughout their entire reproductive journeys. This can be in a linear fashion: patients are seen for preconception counseling and then ask to meet with us to re-review prenatal testing options in an established pregnancy. More often, we meet with families multiple times related to abnormal prenatal diagnoses where we identified a genetic etiology. This includes interconception counseling where we can expertly review both PGT and prenatal testing options and coordinate the family's preferred route. This seamless continuity of care across departments creates an ongoing relationship between the GC and family, providing increased psychosocial support and rapport. Job titles and department name have also been updated to "reproductive genetics" to reflect this expanded scope of practice.

Conclusion: Genetic testing options in the reproductive setting continue to increase in complexity. Our clinic model allows for GCs to provide effective care where our skill set is most needed.

“You're going to uncover more stuff and you don't know what to do with it” - Exploring uncertainties encountered by health professionals working in prenatal genomics: An international qualitative study.

Jennifer Hammond 1,2, Jasmijn Kalpwijk 3, Eleanor Harding 2, Stina Lou 4, Ida Vogel 4, Lisa Hui 5,6, Emma Jane Szepe 5,7, Melissa Hill 1,2, Kelly Ormond 8, Lyn Chitty 1,2, Sam Riedijk 3, Celine Lewis 1,2, Consortium for Understanding Uncertainty in Prenatal Genomics

1. North East Thames Regional Genetics Service, Great Ormond Street Hospital for Children NHS Foundation Trust, London, UK. 2. Genetics and Genomic Medicine, The UCL Great Ormond Street Institute of Child Health, London, UK. 3. Dept of Clinical Genetics, Erasmus MC, Wytemaweg 80, 3015 CN, Rotterdam, Netherlands 4. Center for Fetal Diagnostics, Aarhus University Hospital, Aarhus, Denmark 5. Reproductive Epidemiology group, Murdoch Children's Research Institute, Melbourne, Australia 6. Department of Obstetrics and Gynaecology, University of Melbourne, Melbourne, Australia 7. Department of Paediatrics, University of Melbourne, Parkville, Victoria, Australia 8. Stanford University School of Medicine, Dept of Genetics and Stanford Center for Biomedical Ethics, Stanford, CA, USA

Introduction: Use of genomic technologies has dramatically changed the way we screen and test for suspected fetal genetic abnormalities. Tests such as chromosomal microarray analysis and exome sequencing bring significant clinical benefits through higher diagnostic yields, however a key concern is the increasing amount of uncertain information that may be generated. This uncertainty can be challenging for healthcare professionals (HCPs) who play a vital role in preparing families for testing and communicating results which may influence pregnancy outcome decisions.
To identify the different sources of diagnostic uncertainty that occur through prenatal genomic testing and explore attitudes to dealing with this on an international basis.
Methods: This was a qualitative study conducted across five countries with different healthcare systems. To date, 24 semi-structured interviews have been conducted with HCPs (including clinical scientists, geneticists, fetal medicine specialists, obstetricians, genetic counsellors) in the UK, Netherlands, Denmark, Singapore and Australia. Interviews were transcribed verbatim and, where required, translated into English and analysed using thematic analysis.
Results: Sources of uncertainty identified by HCPs were categorised into three broad themes; uncertainties that result from incomplete knowledge, uncertainties that are associated with the condition that was diagnosed, and uncertainties about the reliability of the test. These uncertainties were found to be common across countries.
Knowledge: Uncertainties in this theme are the result of incomplete knowledge and understanding of genotype-phenotype correlations; including limitations in our understanding of human fetal development, not knowing how a genetic anomaly with a well-known postnatal phenotype presents prenatally and unclear variant pathogenicity, in particular variants of uncertain significance. Condition: Uncertainties related to the condition that was diagnosed included; diagnosis of a condition with an uncertain prognosis, including conditions with variable penetrance and/or variable expression, unexpected diagnosis of a condition that may or may not be the cause of the phenotype, diagnosis of a condition not related to the reason for testing (secondary findings) and incomplete results such as finding only one autosomal recessive variant compatible with the fetal phenotype. Test reliability: These uncertainties were associated with the reliability of the test itself and included the diagnostic yield, the technical validity of a result and the potential for false positive and false negative results.
Conclusions: This study confirms that globally we face varied sources of uncertainty for HCPs working in the prenatal genomic setting. We are currently exploring how genomic uncertainty is managed in day-to-day practice in these different healthcare systems.

Development of a framework to include Indigenous Australians in education and training pathways for Genetic Counsellors.

Jan Hodgson, Linda Browne

Department of Paediatrics, University of Melbourne, Parkville, Victoria, Australia

Indigenous Australians are under-represented in many health professional settings including clinical genetic counselling services. Increasing the number of Indigenous practitioners would enhance services' provision of culturally responsive care and support self-determination for Indigenous communities in genetic health.

To date, no Indigenous students have ever enrolled in the Master of Genetic Counselling (MGC) at The University of Melbourne (UOM). Educational institutions, with responsibility for training genetic health practitioners, are faced with specific challenges in recruiting Indigenous students and supporting their successful transition into the professional workforce.

Broadly, these challenges are twofold:

- the cultural relevance of existing genetic health curricula
- the social and educational disadvantage of Indigenous students who experience significant impediments to accessing and completing tertiary education.

The Victorian Government Department of Health and Human Services have provided the University of Melbourne Master of Genetic Counselling (MGC) program with funding to encourage and facilitate Indigenous engagement in the genetic counselling training pathway at UOM. The project will address the range of barriers that hinder Indigenous students' participation in the requisite degree program, subsequent professional employment and certification processes necessary to become a fully certified genetic counsellor.

This presentation will describe the intervention strategy that has been developed to:-

- address academic entry barriers by addition of a preparatory year for the MGC program at UOM
- address financial barriers with a scholarship program to enhance the existing equity-based ABSTUDY program
- address employment transition barriers with a supported graduate training post and supervision to facilitate full certification as a genetic counsellor with the Human Genetics Society of Australasia (HGSA)

Facing uncertainties in the diagnosis of Congenital Long QT Syndrome: A discursive investigation of the clinical management of VUS in genetic counselling

Andy Lok-Chung HUI, N/A

School of English, University of Hong Kong/Research and Impact Initiative on Communication in Healthcare (RIICH)

This study examines the interaction in the context of genetic counselling consultation for Congenital Long QT Syndrome (LQTS) in Hong Kong. A particular focus is put on how genetic professionals inform parents about the choice of familial genetic diagnosis and explain subsequent testing results based on the test result of variant uncertain/unknown significance (VUS) in LQTS-related genes in children.

LQTS is a subtype condition of Sudden Arrhythmic Death Syndrome (SADS) characterized by the clinical features of prolonged corrected QT interval in electrocardiogram and arrhythmic events (and eventually sudden cardiac death) in individuals with a normal cardiac structure (Ackerman et al, 2011; Spoonamore & Ware, 2016). While international guidelines are lucid concerning pathogenic variants in inherited cardiac disorders, (Ackerman et al, 2011; Gollob et al, 2011; Priori et al, 2013) the situation is more contentious with cases of VUS, especially coupled with the variable expressivity and penetrance of LQTS. Although testing first-degree relatives based on VUS can provide insights into the segregation pattern (Cowman et al, 2009; Garrett et al, 2016), such testing runs the risk of becoming predictive testing with little evidence. This amounts to a dilemma that poses interactional difficulties to genetic professionals.

From 41 video-/audio-recordings of genetic consultations for SADS, this study draws on 8 consultations in which 4 symptomatic infants/children carry VUS in LQTS-related genes (KCNH2 and SCN5A). In the dataset, genetic professionals perform genetic testing on first-degree relatives based on the VUS result of child probands. In this study, a discourse analytic approach is adopted as it pays special attention to context and closely examines the moment-to-moment interactions in the clinical setting in order to produce rich insights into medical practices (Roberts & Sarangi, 2005; Sarangi, 2010). Consistently with the recommended practice in international guidelines, professionals avoid framing genetic testing as a predictive test for parents/siblings. Genetic professionals also discursively situate the importance of familial diagnosis/test results in the heart of "helping the child", which functions as a reassuring strategy for parents when facing the ambiguous nature of the VUS result (or in Ackerman's term, genetic purgatory). This study does not attempt to generalize the genetic counselling practice in local contexts; instead, it serves to exemplify how genetic professionals carefully balance different medical and counselling duties (including explanation of genetic testing, interpretation of test results, and emotion support to clients) in the clinic setting and reflects upon the ethical and social implications for such a practice.

An Ethical Framework for Genetic Counseling in the Genomic Era

Leila Jamal, Will Schupmann, Benjamin E. Berkman

Department of Bioethics, NIH Clinical Center (Jamal, Schupmann, and Berkman); National Institute of Allergy and Infectious Diseases (Jamal); National Human Genome Research Institute (Berkman)

The field of genomics has diversified and commercialized since the profession of genetic counseling emerged in the early 1970s. With this evolution, the complexity and number of ethical considerations relevant to genetic counseling has grown. Given this, we see a need to re-visit whether and how ethical principles should be used to guide genetic counseling practice. In this paper, we argue that the concept of non-directiveness, which is grounded in a narrow interpretation of patient autonomy, is conceptually, normatively, and scientifically fraught. We begin with a brief history of non-directiveness and a review of the arguments that scholars have put forth against the concept. After acknowledging the insufficient efforts that have been made to move away from the concept, we turn to a series of arguments about why non-directiveness has become even more untenable given the arrival of the genomic era. Finally, we make the case that genetic counselors should de-emphasize a narrow conception of individual autonomy in favor of a more explicit commitment to the principles of individual and familial beneficence, as well as a more positive understanding of individual autonomy. To translate our arguments into practice, we present an evidence-based framework of six considerations that genetic counselors should consider when deciding if it is ethically acceptable (or even desirable) to provide active guidance to patients.

Intention-formation in rare-disease genetic testing: a study evaluating the Theory of Planned Behaviour (TPB).

Ali Kay
Cardiff University (postgraduate student)

Understanding people's psychological relationship with rare diseases and predictive testing is important in order to target and guide provision (Taylor, 2005). However, understanding individual difference in forming an intention to take or not take a genetic test is challenging because even faced with the same risk factors, some people decide to take undergo testing and some do not (Taylor, 2005). Using a correlational design, this study assessed how well the Theory of Planned Behaviour (TPB) (Ajzen, 1985, 1991, 2015) predicts people's genetic testing intentions in regard to rare diseases. A random sample (n = 81) of the general population and rare disease interest groups was obtained via social media. Participants read stimulus material on three rare disease types, accompanied by questions on their hypothetical testing intentions. Further questions were based on measures developed to address the TPB constructs, along with perceived risk and faith limitation.

Regression analysis revealed the model was able to predict variance in intentions for highly penetrative and incurable rare diseases, consistent with other applications of the TPB (Armitage & Conner 2001; McEachan, Conner, Taylor & Lawton 2011), with intention to take the test being driven by positive attitudes and control beliefs - the only statistically significant predictors. The former is consistent with the findings of Nordin et al. (2004) for an unnamed hereditary disease. Unlike in Frost et al.'s (2001) study relating to Alzheimer's Disease, negative attitudes (e.g. stigmatization) were not found to be a significant predictor, although they were positively correlated with the intention to use a Direct to Consumer (DTC) test privately. Additionally, the model predicted the least variance for this new and growing (Philips 2016) testing method, encouraging further investigation. This study concluded that the TPB can be used to predict people's decision-making towards traditional non-commercial testing for highly penetrative rare diseases.

Note: This independent research project was undertaken in 2018 to meet the requirements of an MSc Psychology (conversion) degree at UCLAN and was awarded the School prize. I am now studying on the MSc Genetic and Genomic Counselling course at Cardiff University and would welcome the opportunity to share, discuss and reflect on this student project.

Cardiac screening uptake in South East Wales

Emily Lamb/Jackie Hill, N/A

All Wales Medical Genomics Service, Cardiff & Vale University Health Board

Aims: To evaluate the uptake of cardiac screening in South East Wales for those at risk of neuromuscular conditions and inherited cardiac conditions
Scope: According to Resta et al's task force report 2006 " Genetic counseling is the process of advising individuals and families affected by or at risk of genetic disorders to help them understand and adapt to the medical, psychological and familial implications of genetic contributions to disease."
In part this also includes accurate information about the screening recommendations for those at risk of inherited genetic conditions. There are many inherited conditions where cardiac surveillance is recommended. This includes inherited cardiac conditions and types of neuromuscular conditions.
Individuals at risk of inherited cardiac conditions such as Hypertrophic Cardiomyopathy, Dilated Cardiomyopathy and Long QT syndrome are recommended to have cardiac surveillance at regular intervals. This is also true for neuromuscular conditions such as Duchenne muscular dystrophy, Becker Muscular Dystrophy and Myotonic Dystrophy which are known to increase the risk of cardiac issues in those affected, and at risk of being affected but also in the case of possible manifesting carrier females.
By exploring the genetic counselling and uptake of surveillance we hope to provide an evaluation of whether these recommendations are met within South East Wales.
Methods: This is a service evaluation involving the use of an evaluation form designed to extract the relevant data. Data will be obtained from reviewing patient genetics and medical records.
Data collection to include patients seen between Jan 2017 to Jan 2018
Results: Pending

Ethical and legal basis of genetic counselling in Russia: areas of improvement

Lapaeva V.V. Advanced Doctor of Law,

Ethical and legal basis for genetic counselling in Russia generally meets international standards despite the fact that the country has not signed the Convention on Human Rights and Biomedicine and is unlikely to sign this document after the presidential decree (dated 11.03.2019), which prescribes genetic certification of the population. However under conditions of insufficient elaboration of the relevant legislation and underdevelopment of specialised forms of medical community's professional self-organisation these general rules do not receive adequate specification in numerous situations. For a number of reasons, the specificity of genetic information, which often poses existential problems for patients, is more acute in Russia than in Western countries. Therefore, there are not enough such already taken measures like the inclusion the basics of clinical psychology in the professional standards of medical genetics. Without highly professional support not only patients, but also geneticists often bear an excessive psychological burden. So it seems reasonable to organise offices of psychological assistance on the basis of state medical-genetic services. Another serious problem faced by Russian counselling geneticists is that according to current legislation they are forced to keep the patient's genetic status in secret, when his right to medical confidentiality conflicts with the rights of his family to receive vital information. The practice shows, that it is necessary to strive for making appropriate amendments to the legislation and finally adopt a code of professional ethics of the medical-genetic community.

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Inherited Predisposition to Mesothelioma, it's not all about Asbestos

Alexandra Lebensohn-1, Grace Fasaye-1, Kathleen Calzone-1, Idrees Mian-1, Raffit Hassan-1

1-Center for Cancer Research, National Cancer Institute (NCI), National Institutes of Health, USA

BAP1 (BRCA1-associated Protein 1) is a tumor suppressor gene located on chromosome 3p21.31-p21.2. Its importance is demonstrated through its involvement in various biological processes including DNA repair, cell growth and cell cycle regulation. Patients with *BAP1* pathogenic germline variants are predisposed to develop *BAP1* Tumor Predisposition Syndrome (TPDS), which carries a significantly increased risk for cancer including uveal melanoma, mesothelioma, cutaneous melanomas and renal cell carcinomas. In addition, patients with germline *BAP1* mutations may be at increased risk of developing meningioma, cholangiocarcinoma and basal cell carcinomas. The exact lifetime risks have yet to be established and will evolve as more data is collected. It is clear however that the cancers involved in this syndrome are often detected at late stages which offer poor prognoses. Mesothelioma has previously been associated mainly with asbestos exposure and therefore hereditary implications have not been considered. It is a difficult disease to diagnose and in fact it is estimated that one of every four to five cases have not been recorded. Up to 12% of mesothelioma patients harbor pathogenic variants in DNA repair genes, with most variants presenting in the *BAP1* gene. Phenotypically, studies have demonstrated that in *BAP1* TPDS there is a higher incidence of mesothelioma in females vs men in contrast to non-*BAP1* related mesothelioma. There is also a tendency toward peritoneal mesothelioma in *BAP1* TPDS as opposed to pleural mesothelioma, which is more common in the general population. Genetic counselors play a critical role in educating primary care physicians and oncologists regarding the hereditary component of mesothelioma and who should be considered for genetic counseling/testing. Our team has developed a multidisciplinary research-focused approach to address the unique needs of *BAP1* carriers. This allows for a distinctive genetic counseling role, incorporating both clinical and research aspects of the profession. In addition to standard education and counseling provided to patients, genetic counselors help implement this new protocol by assisting in the development of eligibility criteria and surveillance guidelines (for which there are no formal guidelines currently), meticulous acquisition of family history, data collection and counseling on cascade testing options on or off study. The study objective is to enrich our understanding of *BAP1* TPDS by observing cancer frequencies, ages of onset and unique phenotypic characteristics in *BAP1* mutation carriers. Participants are followed longitudinally using a robust screening protocol which includes ophthalmological and dermatological exams in addition to abdominal, chest and pelvic MRI. Early detection may lead to a more favorable prognosis.

Genomics in clinical care: Preparing non-genetic health professionals

Ms Elly Lynch, Fran Maher1,2,3, Melissa Martyn1,2,3, Amy Nisselle1,2,3,4, Taryn Charles1,2,3,5, Callum McEwan1,3 and Clara Gaff1,2,3

1. Melbourne Genomics Health Alliance
2. Murdoch Children's Research Institute
3. The University of Melbourne
4. Australian Genomic Health Alliance
5. Victorian Clinical Genetics Services

Genomics has potential to impact almost all areas of medical care, however most non-genetic medical specialists lack confidence to order and interpret genomic tests. Melbourne Genomics has an upskilling strategy to meet education needs of practising non-genetic medical professionals: (1) internships; (2) blended learning short courses in clinical genomics; (3) workshops. Here we describe the case-based, discipline-specific workshops and their outcomes.
Clinical cases address discipline-specific learning objectives. Using a modified Interrupted Case Method (Herreid, 2005), an experienced clinician alternates between presenting case details and directed questioning to guide group discussion and address key learning points. Clinicians experienced in genomics facilitate small group discussions. Pre- and post-workshop surveys evaluate impact.
To date 183 clinicians have attended five of nine planned clinical workshops (cardiology, acute care, congenital deafness and two paediatric neurology). Participants range from medical students to senior consultants. 70% (80/117) of survey respondents already used genetic or genomic testing in their clinical role; however, 76% (89/117) have no formal genetics training. Experience is highest for ordering chromosome and single gene tests (57% and 54%, respectively) and lowest for exome/genome tests (average 32%; range 9-61% across specialties). Self-reporting 'Good confidence' increased for ability to identify the right test for a patient (21% to 45%) and ability to interpret a genomic test report (18% to 41%). Respondents rate case-based learning as the most beneficial aspect. Other strengths include, genomics introduction, targeted discussion groups, and dedicated facilitators with clinical genomics experience. Evaluation is informing development of other components of our upskilling program.

“There’s no such thing as a genetic emergency”: Genetic counselling and the parent experience in acute care

Fiona Lynch1,2,3, Belinda McClaren1,2,3, Amy Nisselle1,2,3, Clara Gaff1,2,3

1Australian Genomics Health Alliance, 2Murdoch Children’s Research Institute, 3The University of Melbourne

Genomics is rapidly being implemented across many areas of healthcare, with the paediatric acute care setting in particular showing great promise. Typically, results of clinical genomic sequencing take between six weeks and six months to report. Through ultra-rapid genomic sequencing (rGS), however, results are received within days. Add to that the immense emotional burden that parents are likely to experience when their child is in intensive care, and it is clear that the use of rGS presents both novel genetic counselling issues and a unique impact on families. Although there is substantial enthusiasm surrounding this new application of genomic technology, it is important that implementation remain patient-centred. It is therefore vital that we understand current genetic counselling practice in the acute care setting, and the experiences of parents, to provide recommendations for practice as technology progresses.

This study aimed to explore both genetic counselling issues and parent experiences of rGS for critically unwell children. Sixteen qualitative, semi-structured interviews were conducted with genetic counsellors (GCs) working in this setting. Interviews were audio-recorded, transcribed, and analysed using thematic analysis. Parents whose children had had rGS in acute care are being approached for interview, commencing in June 2019. Data collection is ongoing (n=1 to date) and will continue until data saturation is reached.

Interviews with GCs revealed a number of themes describing genetic counselling in acute care, including: the need for flexibility (both at an individual and organisational level); concerns for informed consent; ideas about time (lack of preparation time, time invested to see families, and limitations of the traditional '9 to 5' workday of genetic specialists); and the range of existing and new skills required by GCs to practise in this setting.

"Genetics has traditionally been very 9 to 5, there's no such thing as an emergency.....across a weekend there isn't going to be a genetics professional around to see these families, so a lot of it might have to wait, which obviously negates the whole purpose of it being ultra-rapid if you have to wait until a Monday to see them."

These GC perspectives will be complemented by the parent interview data. This project will inform the final phase of a larger study, which aims to develop recommendations for genetic counselling in the acute care setting. Further research will investigate additional stakeholder perspectives (such as other health professionals), resulting in the most patient-centred care for families impacted by this new technology.

Co-development of education and support resources for families with severe early-onset genetic epilepsy

Rebecca Macintosh,2 Suzanne Nevin1, Fleur Le Marne,1 Dr Brittany McGill, 1,3, Dr Rani Sachdev,2 Prof Claire Wakefield,1,3 A/Prof Ann Bye,1 Dr Elizabeth Palmer,1

1School of Women's and Children's Health, University of New South Wales, Sydney, Australia
2Centre for Clinical Genetics, Sydney Children's Hospital, Randwick, New South Wales, Australia 3Kids Cancer Centre, Sydney Children's Hospital, Randwick, New South Wales, Australia

Severe and early onset epileptic encephalopathies (SEE) are characterised by intractable seizures resistant to multi-drug treatment. Developmental outcomes are devastating, with profound cognitive, behavioural and neurological impairments and childhood mortality is ~20%. Over 400 different genetic causes for SEE have been identified, however, information relating to these ultra-rare genetic conditions is limited to research publications, which are inaccessible to families and non-specialist clinicians. Further, little data exists to inform the optimal manner(s) of presenting information following a rare genetic SEE diagnosis to families.

Research Objectives

1. Understand parent experiences undergoing genetic testing and to investigate parent information needs regarding their child's medical management.
2. Examine preferences for content, style and mode of access to information at different stages in their child's diagnostic journey.
3. Collaborate with parents and clinicians involved in the multidisciplinary care of SEE patients to co-design customised information resources.

Methodology Twenty parents (20% male) of children who have undergone genomic testing for a suspected SEE within the past 5 years were recruited from the Sydney Children's Hospital Network. Semi-structured interviews were conducted to investigate pre and post-test experiences and to understand information needs and preferences. Interviews were transcribed, de-identified, coded and analysed for salient themes. Content and iterative thematic analysis was conducted to explain experiences.

Lessons learned

Key themes emerged with participants identifying:

- Difficulties comprehending complex genomic information, and challenges attempting to navigate and gain access to healthcare supports.
- The need for information to be simplified, specific to their child's condition and understandable.
- Desire for accessible and customised information resources, in a variety of formats to complement information received during consultations.
- Importance of having a support network and developing rapport with clinicians.

Implications for practice and future research

The study is ongoing but preliminary findings highlight that parents are likely to benefit from condition-specific information to support them during and after receiving genomic testing. Results are informing the co-development of a customised clinical information resource with active input from both parents and clinicians. The dissemination of an up-to-date clinical resource will facilitate optimal care for children affected with these complex conditions by translating new genetic knowledge to improve health outcomes for patients and families.

One deciding factor: The decision-making process of women offered termination of pregnancy for a serious congenital abnormality

Malebo F Malope1, Karen Fieggen1, Tina-Marié Wessels1

1 Division of Human Genetics, University of Cape Town

Background: A weekly pregnancy counselling clinic is held in conjunction with foetal medicine experts at Groote Schuur Hospital for women with pregnancies complicated by foetal anomalies. Those in whom serious congenital abnormalities with a poor prognosis was diagnosed, may be offered termination of pregnancy (TOP) during genetic counselling. The experiences and decision-making process of these women in this clinic is poorly understood in the South African setting. This project explored how these women make their decisions and the factors that played a role in the decision-making process.

Methods: Ethics approval was obtained from the Human Research Ethics Committee at the University of Cape Town. The women were identified using the Division of Human Genetics pregnancy counselling database. They were initially contacted by either the clinician or the genetic counsellor who had previously counselled them to discuss participation. Those agreeing to participate were contacted by the researcher. Qualitative research drawing on the principles of phenomenology was used as the study design. The data was collected in the form of semi-structured, face-to-face interviews ranging from 30 minutes to an hour in length. Demographic information was obtained through close-ended question. The interviews were recorded and transcribed verbatim. Data was analysed using a thematic data analysis approach.

Results: The women considered a multiple of factors when considering the option of termination of pregnancy. However, each woman had one final deciding factor that lead to the decision to continue with the pregnancy or to terminate. Reaching the final deciding factor was a process from the time of diagnosis until the decision. Following the shock and acute grief the women considered a multiple of factors with external factors also impacting their process. They moved back and forth until the most important factor was identified and then they made their decision.

Conclusion: This decision-making process is unique to each woman. Decision-making process for women considering termination of pregnancy for a serious congenital abnormality is not linear but rather negotiating through multiple factors to reach the deciding factor and the decision. This was surprising as it was expected that multiple factors contribute to the final decision rather than one deciding factor. These results have provided more information about our population and thus better supportive counselling services for women in the same situation can be provided. This information also highlighted areas of focus and the importance of facilitating decision-making during genetic counselling.

‘I want to have both my breasts off even if I’m not BRCA’: 3 case studies of women who requested risk-reducing contralateral mastectomy regardless of their genetic status and contralateral breast cancer risk

Athalie Melville, None

Wessex Clinical Genetics Service. University Hospitals Southampton NHS Trust

Increasing numbers of women are aware that a risk-reducing double mastectomy is an option to reduce breast cancer risk for those at high risk. However, risk-reducing contralateral mastectomy is not routinely offered to women who have a moderate risk of developing a contralateral breast cancer.
Here I present three case studies of women who have each had unilateral breast cancer. Each woman has undergone genetic testing to look for BRCA1 and BRCA2 gene alterations. In two out of three cases, no pathogenic BRCA1 or BRCA2 gene alteration has been identified. The results are awaited in the third case. In two cases, the woman's risk of developing a contralateral breast cancer was assessed to be moderately increased following genetic test results. Routine practice in the UK under NICE guidelines would be to offer these women regular mammographic screening, but not risk-reducing surgery. In all three cases, the risk of a recurrence from the primary breast cancer was greater than the risk of a contralateral breast cancer. However all three women requested risk-reducing contralateral mastectomies with their surgeon. This is despite empirical evidence that the survival rate for breast cancer is not improved after removing the healthy breast. The post-surgical complication rates with bilateral mastectomies are also much higher compared to unilateral mastectomies with or without breast reconstruction.
During the genetic counselling session for each case, we discussed motivations for wanting risk-reducing surgery. In all three cases, the motivations included reducing the fear of developing a new breast cancer, eliminating the need to undergo further chemotherapy and treatment, and avoiding dying from a new breast cancer.
As women's knowledge of available surgical options increases, I anticipate that numbers of such surgical requests will increase. This poses challenges to genetic counsellors and breast surgeons who counsel women on how to manage their future contralateral breast cancer risks. There is a need to evolve our genetic counselling skills to accommodate these challenges.

A study exploring how patients with motor neurone disease view genetic testing and the factors affecting these views

Katrina Merrifield, Laura Boyes

University of Manchester, Birmingham Women's and Children's NHS Foundation Trust

Diagnostic genetic testing for motor neurone disease is becoming increasingly relevant as a single mutation of large effect can be identified in 60-80% of patients with a family history, and approximately 10% of patients without a family history. There are also an increasing number of gene-specific treatment trials available. This has led to proposals to offer diagnostic genetic testing widely to patients with MND at the point of diagnosis, irrespective of family history. However, the complex aetiology, devastating nature of the progression of this disease, and lack of currently available treatment mean that diagnostic genetic testing for MND has the potential to have complicated and substantial implications for patients and their family members. This research aims to fill gaps in our knowledge regarding how patients with MND view genetic testing, and what factors may contribute to these views, and to consider what implications these views may have for testing and counselling protocols and guidelines.

A qualitative methodology, utilising semi-structured interviews and thematic analysis, was used. The results from the two participants interviewed suggested that they held generally positive views about genetic testing, particularly regarding its potential to help in developing treatments. In contrast, their views regarding its utility for family members diverged; however, these views were also affected by misconceptions about the reasons for, and implications of, such testing. Neither participant saw family planning as a potential utility, although their rationale differed. In addition to misconceptions, other factors that seem to have influenced their views were: their lack of family history, the causal attributions they had made for why they had developed MND, and their different coping strategies. Although the study is limited by small sample size, it gives useful and novel insights into the views of those recently diagnosed with MND with regard to genetic testing, and suggests that genetic counselling may be an intrinsic part of diagnostic testing for MND. Based on this study, it is recommended that further consideration be given to the pathways involved and the roles and responsibilities of the multi-disciplinary team in respect of counselling provision.

Interventions in a Specialist Drug Development Unit to improve family history documentation and onward referral of patients with advanced cancer to Cancer Genetics Services

Cathryn Moss, Terri McVeigh, Simon Ward, Elena Cojocaru, Wen Xu, Janet Hanwell, Mary van Zyl, Lorraine O'Leary, Johann DeBono, Udai Banerji, Stan Kaye, Juanita Lopez, Angela George

1. Cancer Genetics Unit, Royal Marsden NHS Foundation Trust, Fulham Road, London
2. Drug Development Unit, Royal Marsden NHS Foundation Trust, Sutton, Surrey
3. Gynaecological Oncology Unit, Royal Marsden NHS Foundation Trust, Fulham Road, London

Molecular aberrations in cancer may represent therapeutic targets, and, if arising from the germline, impact further cancer risk management in patients and their blood relatives. Annually, 600-700 patients are referred for consideration of experimental drug trials in the Drug Development Unit (DDU) in our institution. A proportion of patients may merit germline genetic testing because of suspicious personal/family history or findings of tumour-based testing. We aimed to assess the impact of different multi-disciplinary interventions, involving different genetics professionals, on family history taking in and referral rates from DDU to Cancer Genetics Unit (CGU).
Methods:
Over 42 months, three interventions were undertaken at different intervals;
1. Embedding a Clinical Fellow in DDU review clinic
2. "Traffic light" system flagging cancers with heritable component and Genetic Counsellor review
3. Virtual multi-disciplinary meeting (MDM) by Consultant Geneticist
Comparative analyses between intervals were undertaken, including referral rates to CGU, investigations and patient outcomes. Family history-taking in a sample of 20 patients managed in each interval was assessed by retrospective chart review.
Results: Frequency of family history taking, and referral to CGU, increased with each intervention, particularly, the virtual MDM (40%-v- 85%). Referral rates increased over the study period, from 0.1 referral/week (5/year, 0.36% total referrals) to 1.2/week (projected 63/year (3.81%). Forty-four (52%) patients referred required germline testing, in three of whom variants were identified. Non-attendance rates were low (6, 7%).
Conclusion: Multidisciplinary working between CGU and DDU facilitates germline testing of those patients that may otherwise miss the opportunity. The enhanced engagement between specialist services was sustained, regardless of which genetics professional was leading the intervention. This provides an example of the diverse and evolving role of the Genetic Counsellor, in enabling appropriate referrals from specialist services, without increasing pressure on out-patient clinics.

EXPLORING NECESSITIES IN A RARE DISEASE SERVICE: IDENTIFICATION OF GENETIC COUNSELLING TIME –CONSUMING.

María Mercedes Navarro de Miguel (1,3), Rosario Sánchez Martinez (2.1.), María Elena García Payá (2.2.), María Gutierrez Agulló (2.2.), José Luis Girela López(1.).

(2.1.)-Unidad Multidisciplinar Enfermedades Baja Prevalencia. Servicio de Medicina Interna. Hospital General Universitario de Alicante- .(2.2.) -Laboratorio de Biología Molecular. Hospital General Universitario de Alicante-. (1)- Departamento de Biotecnología. Universidad de Alicante-. (3)-GENIFEN- Gestión y Comunicación de la Información genética. Genetic Counselling Services-.

INTRODUCTION
The increased demand for consultation of low prevalence diseases in recent years has led to an increase in the demand for genetic tests, which arises the assessment of the needs of this service. The main objective is to detect the need to integrate the information of the genetic/genomic technology to the service through the Genetic Counselling, with the purpose of the diagnostic implementation and the quality of life of the patients.
MATERIALS AND METHODS
The analysis has been carried out on how often the genetic information/data is handled, as well as the ranges of time used for each of the types of clinical information in the rare disease department of the Hospital General de Alicante (HGA), Spain.
The subject matter are the consultations of the rare diseases department of the HGA. There has been an evaluation of the time spent treating genetic data per patient, as well as what percentage the genetic information represents in the total clinical data used.
Random consultations have been chosen in the 200-hour period of medical visits during the training period in the Genetic Counselling. The visits have been divided into Diagnostic Consultation and Follow-up Consultation. It has been established in each consultation a classification of the types of clinical data used in each visit and the percentage of time spent for the communication of important clinical data for the patient.
RESULTS
The physician's office of rare diseases of the HGA receives patiens Median age was 45 (37-54). The 50% of pathologies treated are rare hereditary collagen diseases, 20% are rare metabolic diseases, and 20% are other rare diseases.
The average total time spent for each patient is approximately 40 minutes.
The documents and clinical data regarding genetic test results represent 15% of the information concerning a patient. The time spent in communicating genetic information in a first consultation is 15%. The time spent if it is a second consultation where genetic results have to be communicated is 30% and up to 45% if a family tree is made.
CONCLUSION
The augment in the use of genomic data by patient and some family members creates the need to increase the time dedicated to the Genetic Counselling per patient.
The need arises for creating a diagnostic circuit where the Genetic Counselling is introduced. This diagnostic circuit would implement the efficiency of time used by the patient, as well as the diagnostic performance of the service.

Psychological distress following direct-to-consumer testing for a single-gene condition: A case study.

Nadia Preitner,

We present a genetic counselling case where a client was seen after direct-to-consumer (DTC) genetic testing identified that she was a carrier of a BRCA2 pathogenic variant. The client had not previously seen a genetic counsellor and was referred to her local NHS genetics service by her GP after receiving her result. During the genetic counselling appointments, the client exhibited severe anxiety and distress.
The present case illustrates the possible negative psychological impact DTC genetic testing can have on clients who are unaware of the possible results and their implications. We explore how the genetic counselling process was modified to alleviate the client's distress, as well as the difficulties encountered by the genetic counsellor throughout the counselling process.
Finally, we comment on the burden DTC genetic testing places on already strained NHS genetics services and the lack of current guidelines for genetic counsellors on how best to serve such clients.

From Washington State to Sweden & Beyond: Prenatal Genetic Education Videos Spark International Collaboration

Jennifer Rietzler, MS, CGC (1), Charlotta Ingvoldstad Malmgren, PhD (2,3), Kelly Donahue, MS, CGC (1), Katie Stoll, MS, CGC (1), Debra Lochner Doyle, MS, CGC (4)

(1) Genetic Support Foundation, Olympia, WA, USA
(2) Center for Fetal Medicine and Center for Rare Diseases, Karolinska University Hospital, Stockholm, Sweden
(3) Swedish Network on Information about Prenatal Diagnosis (SNIF), Stockholm, Sweden
(4) Washington State Department of Health, Kent, WA, USA

In May of 2015, the Washington State Department of Health (DOH) sent out a request for a project entitled, Educational Videos about Prenatal Screening and Diagnosis Options. Genetic Support Foundation (GSF), a not-for-profit organization based in Olympia, Washington, USA, dedicated to increasing access to independent genetic counseling services and resources, responded to this request and was awarded the contract. With the assistance of Truscribe Animation Studio (Madison, Wisconsin, USA), Washington DOH and GSF partnered to develop seven videos in this series, including topics surrounding prenatal ultrasound, maternal serum screening, cell-free DNA screening, prenatal diagnostic procedures, and the conditions typically identified with these modalities.

These videos were released in June of 2016 and have been widely used by providers throughout the United States and internationally. While the initial videos were produced in English, they contain scripts and storyboards that allow them to transcend the boundaries of individual languages with health literacy parameters and a neutral narrative in mind. Some of the videos from this series have now been translated into Spanish as well as Swedish - with Mandarin on the horizon. We describe how these videos provide an avenue that promotes informed decision-making and patient autonomy in an era where prenatal genetic screening and testing have become increasingly routinized through primary obstetrical care with many patients not appreciating the nuances and voluntary nature of these options. In particular, we describe the impact of the utilization of these videos in Sweden through the Swedish Network on Information about Prenatal Diagnosis (SNIF), an organization with a mission to increase awareness of the availability of prenatal screening and testing as well as the ethical issues encompassing these options. We also present possibilities for future implementation of these videos from Washington State to Sweden and beyond.

Co-creating a knowledge base in the 22q11.2 Deletion Syndrome community

Roberta Rizzo, Profs. Marianne van den Bree
Dr. Rose Thompson

Cardiff University

22q11.2 DS is characterised by its variability, rarity and variety of features ranging from congenital heart conditions to psychiatric and behavioural issues. As a result, health information seeking behaviour is different to other more common conditions. A mixed method exploratory study was carried out to understand how parents access information and support, and how they feel about different information sources. The study also looked into how that information is shared. A survey was carried out with 29 carers of children with 22q11.2 DS and it was found that most information about psychiatric and behavioural conditions came from the internet or support groups. Qualitative interviews were also carried out with fifteen families and support group representatives and thematic analysis was applied. Five main themes emerged; 1) Medical abandonment 2) Parent expert 3) Support groups 4) Internet and Social Media 5) Complexity of the condition. Medical abandonment appears to lead parents towards support groups and the internet where they gain expertise in their child's condition. Parents share this information and provide support to like-minded parents; this contributes to their coping with the complexity of the condition. Support groups facilitate these discussions, gather information and pass these on to researchers and professionals in order to raise awareness and improve clinical practice.

Using a genetic counsellor to streamline patient care in a subspecialty setting: the craniofacial genetic counsellor

Sarah Robart, Louise Wilson

North East Thames Regional Genetics Service, Great Ormond Street Hospital

Craniofacial services in the UK are provided by four specialised Craniofacial Units. The large number of patients with apparently non-syndromic single suture (NSSS) craniosynostosis means this population is difficult to manage in a traditional clinical genetics setting. Although genetic diagnoses are an integral aspect of care for individuals with craniofacial disorders, no genetic counsellor (GC) has previously been specifically appointed to work with any of the Craniofacial Units.

In order to streamline care for this patient population with complex multidisciplinary needs, North East Thames Regional Genetics Service created a new role for a GC to integrate with the Craniofacial Unit at Great Ormond Street Hospital using a combined approach of face-to-face and phone consultations (telemedicine). This position is the first of its kind across the four Craniofacial Units. We report a summary of our experience one year after introducing the role of the "craniofacial GC."

In this new service model, the craniofacial GC is responsible for assessing patients with apparently NSSS craniosynostosis via telemedicine and initiating genetic testing in the form of a gene panel. The majority of these patients have negative results, which prompts a review of their history, pedigree, and photographs by a consultant clinical geneticist with expertise in craniofacial disorders. A small but significant proportion of patients have a detectable genetic diagnosis. These patients are then seen in a traditional face-to-face appointment with a consultant clinical geneticist, as are all patients with multisuture craniosynostosis or suspected syndromic craniofacial disorders. The craniofacial GC is also responsible for providing counselling for teenagers with established diagnoses transitioning from paediatric to adult services, and for pregnant patients seeking information about recurrence risk or prenatal genetic testing.

Using telemedicine has improved care for patients with NSSS craniosynostosis by providing a more timely and convenient investigation pathway, decreasing waiting times, and saving families a significant amount of travel. This model demonstrates the benefits of integrating GCs in subspecialty teams and using telemedicine at a time when this is becoming increasingly common in the profession. Initial success with this model has already prompted a similar GC role to be created in another UK Craniofacial Unit.

Adult Male 46,XX: Challenges at Genetic Counselling

Glykeria SAMOLADA, A.Chatziparasidou, Sr. Clinical Embryologist, N. Christoforidis Consultant Obstetrician & Gynaecologist, A. Kougioumtzi Psychologist

Embryolab

We present a case of a 33 years old man with severe azoospermia. He came to our IVF clinic asking for the possibility to find and cryopreserve few spermatozoa using any medical treatment available. Sperms' total absence confirmed and he referred to our Genetic Department to evaluate the situation, as testicular ultrasound revealed parenchymal heterogeneity, atrophic epididymis and low size of both testis.
Karyotype analysis showed to be 46,XX SRY-positive, whereas further Yq microdeletions analysis revealed a complete deletion at AZFabc. These finding confirmed to diagnosis of testicular disorder of sexual development (DSD).

DSD at males has been described as "Pseudohermaphrodite" "Intersex" or "Sex reverse", terms which are not using anymore, as karyotype and genetical data can not exclusively define human sex. 46,XX SRY-positive males are usually diagnosed in late adolescence or adulthood, there is no clinical sign before puberty, with the exception of small testis and infertility as in the present case.

Couple referred to a Genetic Counsellor (CG) to discuss karyotype and other genetical results, recognising the importance of counselling and communication skills required in such cases which may significantly affect man's life balance. GC's priority was to inform him in a way that will not affect couple's relation and marriage, his sexual life, as also his psychological and social balance. The option to discuss results alone without his wife was given, but he chose being together. He did not mentioned any significant at the three-generation family pedigree, he was in good health mentioning only few finding at a recent fatigue test. GC made it clear that gender is not defined by law, social, phenotypic or biological data and none/nothing can define a 33-years old, educated man better than himself. Karyotype is an important result for his life and health as urologist and endocrinologist should monitor 46XX,SRY-positive men and cardiological evaluation should be done too. Testicular biopsy was not recommended, with sperm donation being a reproduction option.

The interdisciplinary approach is very important at 46XX males management and Genetic Counsellor's role is essential in diagnosis, prognosis and correctly informing couples and families

Attitudes of Genetic Counsellors' towards counselling young adults through predictive testing for Huntington's Disease (HD).

Urvi Savania, 2) Lesley Snadden 3) Leah Marks

1)Pre-Registration Genetic Counsellor, Oxford hospitals, MSc Genetic and Genomic Counselling, University of Glasgow UK.
2)Clinical genetics, NHS Greater Glasgow and Clyde, Glasgow, UK
3)Institute of medical genetics, University of Glasgow.

In the UK, majority of individuals who seek a predictive test for Huntington's disease (HD), have been primarily found to be young adults (YA) (18-30 years old). With no current direct medical benefit to testing, as well as the uncertainty in the severity/age at which symptoms may appear, the emotional, ethical and other psychological consequences associated with the test have been found to be very high. As a result of this, the pre-symptomatic predictive testing protocol (PPT) was established to guide at-risk individuals, as well as healthcare professionals to understand the impact predictive testing can have on one's life, facilitating informed choices. The importance of genetic counselling and additional support for at-risk individuals remains crucial to enable a better understanding on the impact predictive testing can have on one's life. However, YA have previously reported lack of emotional support and a presence of a communication barrier during these consultations. As there is limited research conducted on the explicit experiences of genetic counsellors with predictive testing for HD, it is difficult to establish why this gap in support and communication exists. The aim of this study was to gain a better understanding of experiences of GCs whilst counselling at-risk young adults for predictive testing for HD, as well as explore the HD service provision in Scotland. Nine genetic counsellors from three regional centres in Scotland (Glasgow, Aberdeen and Dundee) were interviewed using a semi-structured interview guide. Thematic analysis identified three key themes: challenges faced with young adults and Huntington's disease, aspects of the pre-symptomatic predictive testing protocol and the relationships with other health care professionals. Being made aware about these experiences can highlight gaps in training and support for genetic counsellors as well as other healthcare professionals, and help update guidelines to better support younger adults at risk of later onset conditions.

Recontacting former breast cancer patients for updated genetic testing

Shiri Shkedi-Rafid*, Michal Sagi*, Tamar Peretz-Yablonski**

* Center for Clinical Genetics, Hadassah Medical Center, Jerusalem Israel; **Sharett Institute of Oncology, Hadassah Medical Center, Jerusalem Israel

Background:
Historically in Israel, BRCA1/2 genetic testing consisted mainly of founder mutations. In 2012 new guidelines were issued, which enabled cancer patients with a ≥10% carrier risk to be eligible for comprehensive BRCA1/2 testing. According to the PennII risk-assessment-model, women diagnosed with breast-cancer aged ≤42 reach this threshold. These new guidelines created a subset of patients who had been found not to be carriers of the BRCA1/2 founder mutations prior to 2012, and had subsequently been discharged from genetics.
Aims:
Our aim was twofold: (1) To assess the feasibility of recontacting former breast cancer patients for further genetic testing; (2) To better understand the perspectives of these women on recontact and re-testing.
Methodology:
We evaluated the medical records of women diagnosed with breast cancer age ≤42 years who had a negative founder BRCA1/2 testing between 1997 and 2012. Where no contraindication was identified, we contacted patients by phone and offered a genetic counselling appointment. Structured interviews were conducted at the end of the genetic counselling session (n=35), analysed using grounded-theory methodology. Women who declined further testing were asked for their reasons during the telephone conversation.
Results:
The uptake of a further genetic counselling appointment was 85%, with 100% of those women consenting to additional testing. Additional testing was considered a responsible behavior to ensure women's own health and that of their family. Some expressed a desire for reassurance. Although some women were stressed by the recontact, all perceived it as an expression of care, for which they were very appreciative.
Of the women recontacted, 15% declined an appointment and testing. They expressed reluctance to reopen "old wounds" and exhaustion from never-ending tests.
Conclusions:
Recontact is highly acceptable to breast cancer patients, and there is a high motivation towards updated genetic testing, which is extant for years after the diagnosis. The complexities of finding patients suitable for recontact, and the communication skills necessary for this task require experienced practitioners.

The BAFopathies: A New Way to Conceptualize Disorders of the BAF Chromatin Remodeling Complex

Ayaka Suzuki, MS, LCGC 1,4, Nicole Z. Mans, MS, LCGC 2,4; Jeanna L. Welborn, MD 2; Katherine A. Rauen, MD, PhD 1

1 Division of Genomic Medicine, Department of Pediatrics, University of California (UC) Davis Medical Center
2 Hereditary Cancer Program, UC Davis Comprehensive Cancer Center
3 Division of Hematology and Oncology, UC Davis School of Medicine
4 These authors contributed equally to this work

Germline variants in SMARCB1 cause several distinct genetic syndromes, including schwannomatosis and rhabdoid tumor predisposition. SMARCB1 is part of the BAF chromatin remodeling complex, the human analog of SWI/SNF, which modulates gene expression through the repositioning of nucleosomes. Problems within the complex lead to the disruption of various cellular processes, including tumor suppression. We report on a novel presentation in a family exhibiting two distinct SMARCB1-related phenotypes and argue for a new disease model for the family of disorders. The proband, a woman of Mexican ancestry, presented at the age of 40 to the University of California Davis (UCD) NF/Ras Pathway Genetics Clinic with a history of 8 schwannomas in the thoracic cavity, hard palate, and sinus. Initial onset was 30 years of age. Testing identified a heterozygous intronic variant in SMARCB1 at c.501-25A>G. Pathogenicity was determined by RNA-based analysis, which showed that the variant led to out-of-frame missplicing of intron 4. It is unknown whether the variant was inherited or de novo in the proband. Of note, she has no known family history of schwannomas or other SMARCB1-related findings in her generation and those previous. Meanwhile, the proband's son presented at 6 years of age in the UCD Hereditary Cancer Program clinic for evaluation of Li-Fraumeni syndrome due to a history of choroid plexus carcinoma (CPC) at 8 months of age. Once the SMARCB1 variant was identified in the proband, he was tested and confirmed to have inherited it. CPC is not one of the malignancies generally recognized as being associated with SMARCB1 variants. Although schwannomatosis and rhabdoid tumor predisposition syndrome are typically considered distinct, a few families have been reported with co-occurrence of multiple SMARCB1 phenotypes. This family's phenotype broadens our understanding of SMARCB1-associated disease and highlights the need for a new disease model for SMARCB1 and related disorders. We propose the term "BAFopathies" to refer to disorders caused by germline variants in genes within the BAF chromatin remodeling complex. Historically, these medical genetic conditions have been defined separately due to their disparate phenotypes. This is particularly true because pediatric genetics and cancer genetics patients are often seen by separate specialists. The relatively recent identification of causative genes allows us to link the seemingly unrelated phenotypes together. Experiences such as this family's in which multiple phenotypes are caused by the same familial variant further support the need to rethink how we approach and counsel families with these disorders.

Genetic Counselors Belgium: Need for professional recognition

Virginie Szymczak, Aude Lombard, Ileen Slegers

Center for Pathology and Genetics (IPG), Charleroi
Center for Medical Genetics Ghent, University Hospital Ghent
Center for Medical Genetics, University Hospital Brussels

Background: The genetic counseling profession is continuing to develop globally. In some countries the profession has been well established, while others are still outlining their scope of practice, like in Belgium. As the use of genetic and genomic testing is being greatly extended across a range of specialties, there is a high demand for genetic counselors trained to provide appropriate information to families about their condition, genetic and genomic testing, facilitate reproductive decision-making and support to adjust to a diagnosis or genetic risk situation. The working group of genetic counselors in Belgium was launched in 2015, in association with the Belgian Society of Human Genetics (BeSHG).

Methods: The professional background of the genetic counselors in the BeSHG working group was explored by questionnaire.

Results: Currently there are 19 genetic counselors in Belgium. It is a very heterogeneous group in terms of educational background, professional background, level of work experiences, autonomy, tasks, etc. The majority has a Bachelor degree. In Europe, there are only 6 recognized training programs in the profession of genetic counselor (UK, France, Spain, Portugal, Norway and Romania). For this reason, some genetic counselors have followed an additional training abroad.

Conclusion: Generally, we can conclude that the current population of genetic counselors in Belgium is very heterogeneous. Since the profession genetic counselor is not recognized in Belgium, there is currently no national registration process, no recognized training program, no quality control, etc. As a consequence the current main goal of this working group is the recognition of our profession.

Clinical implications: The professional recognition would protect the title of genetic counselor and consequently would imply quality control and good practice guidelines. Recognition of this profession will make it possible to answer - the ever - increasing demand for genetic counseling. According to the EBMG: 'genetic counselor' should be a protected professional title referring to a health professional who had been educated and trained at Master's level to enable them to develop the core competence defined for the role and to practice according to the Code of Ethics.

Pitfalls of prenatal exome sequencing: evolving phenotypes

Dagmar Tapon, Tina Prendeville, Christoph Lees, the Prenatal Assessment of Genomes and Exomes Consortium, Lyn Chitty

Centre for Fetal Care, Queen Charlotte's & Chelsea Hospital
Imperial College Healthcare NHS Trust
Du Cane Road, London W12 0HS

Prenatal genome and exome sequencing is increasingly being employed to increase diagnostic yield of prenatal diagnosis. In the UK, prenatal exome sequencing will be rolled out nationally later this year. One recognised difficulty of interpreting results of prenatal exome sequencing is the lack of phenotypic data that may only present postnatally, such as intellectual disability. However, another emerging limitation of prenatal exome testing is the existence of new phenotypes not previously recognised in the prenatal period. Phenotypes may only evolve postnatally into recognised conditions. We present an example of a case with intrauterine growth restriction recruited for prenatal exome sequencing as part of the Prenatal Assessment of Genomes and Exomes (PAGE) study. Exome sequencing revealed a likely pathogenic missense mutation in NSD1, a gene associated with the overgrowth condition Sotos Syndrome. While there were no recognised prenatal features of Sotos Syndrome, postnatal assessment of the baby showed macrocephaly and clinical features of Sotos syndrome including developmental delay, confirming the diagnosis.
A similar case of intrauterine growth restriction with an NSD1 microdeletion has recently been published, however the described pregnancy was terminated, eliminating the possibility of confirming the diagnosis postnatally. Our case highlights the difficulty that previously unrecognised evolving phenotypes will present to the interpretation of prenatal exome sequencing and the associated complexity of counselling parents about findings of uncertain significance from new prenatal genomic testing options.

Experiences with transition of gene panel DNA-diagnostics from clinical geneticists to treating physicians in breast cancer patients

Angela van Remortele[1], Maaike Haadsma[1], Beppy Caanen[2], Kim van Kaam[2], Arjen Mensenkamp[1], Rien Blok[2], Edward Leter[2], Michel van Geel[2], Wendy van Zelst-Stams[1], Marjolijn Ligtenberg[1], Nicoline Hoogerbrugge[1];
[1]Radboud University Medical Center, Nijmegen, the Netherlands, [2]Maastricht University Medical Center, Maastricht, the Netherlands.

Introduction
In most European countries requesting DNA-diagnostics for hereditary breast cancer has traditionally been the field of clinical geneticists. Since test results can now be available within 2-3 weeks, these are increasingly considered for determining treatment options in breast cancer patients. Treating physicians therefore increasingly refer eligible, recently diagnosed breast cancer patients for rapid DNA-testing to the clinical geneticist. To facilitate timely test results, we aimed to shift counseling and requests for rapid DNA-diagnostics for these patients from clinical geneticists to treating physicians and evaluate this transition towards so-called 'mainstreaming'.
Methods
The project was initiated by the departments of Clinical Genetics from Maastricht University Medical Center and Radboud University Medical Center, Nijmegen, The Netherlands. Regional hospitals were included one-by-one from July 2018 onwards. Treating physicians were asked for their needs and barriers to participate. DNA-diagnostics consisted of gene panel analysis for *BRCA1, BRCA2, PALB2, CHEK2* and *ATM.*
Results
The website www.DNAfirst.nl was developed to provide treating physicians with hands-on information. Barriers included time investment during consultation and perceived incompetence of providing breast screening advice for family members. Therefore, clinical geneticists joined multidisciplinary meetings. Up to April 2019, nine hospitals were included, 135 requests for DNA-diagnostics made and fifteen pathogenic variants found (11%). All patients with a pathogenic variant were subsequently referred to a clinical geneticist. In total, 9/15 pathogenic variants were found in patients that met the Dutch criteria for DNA testing for hereditary breast cancer (n=104). Interestingly, 4/15 of pathogenic variants were found in the much smaller group of patients who did not meet these criteria (n=21). Additionally, 2/15 pathogenic variants were found in patients for whom it was unknown from the request form whether the testing criteria were met (n=10). Structured evaluation of quality of care and experiences of doctors and patients will follow shortly.
Conclusion
Transition of rapid gene panel diagnostics from clinical geneticists to physicians treating breast cancer patients appears to be feasible.

Genetic counselling, in different sittings. Does the genetic counselling program at UCT measure up?

Tina-Marié Wessels 1, Monica Araujo1,2; Susan Louw1; Malebo Malope1; Kalinka Popel1; Tarryn Shaw1,3; Katryn van Niekerk1,2.

1.Division Human Genetics, University of Cape Town, South Africa. 2.Division Human Genetics, University of the Witwatersrand and the National Health Laboratory Service, South Africa. 3.Cancer Genetics Service of the National Cancer Centre, Singapore

The training of Genetic Counsellors aims to prepare students to become competent healthcare professionals that are able to deal with any case they encounter throughout their careers. Each training unit has their unique training model and it is inevitably that their practical experience will be limited to the setting in which they are training. Evaluation or feedback on the effectiveness of training in equipping the students with the necessary skills to be able to work in different settings is limited. Our trainees have visited different sites and they were asked to write a narrative on their experiences. These sites varied from first world cities to rural South Africa. Common themes in their narratives included: challenges of advocacy for Western based interventions vs. traditional medical practices; communication issues; vocabulary; addressing psychosocial issues; and counselling approaches. While these concepts are not new, it echoes what literature has found. However, these themes demonstrate some of the nuances of working in different settings. From their narratives, it seems they have the necessary skills to adapt to the different settings and that their training is adequate in providing them with a basis from which they could practice. The most important insight from their narratives is the need for practical exposure. This is invaluable as it allows trainees to experience counselling under different circumstances that cannot be taught in a classroom. To this end, UCT will continue to ensure that trainees have an opportunity to experience counselling in a variety of different settings.

Hosting a BRCA gene carrier information day: the experience of the South West Thames Clinical Genetics Service

Elizabeth Winchester, Jessica Bailey, Kelly Kohut, Stephanie Burcher, Virginia Attard, Erin Baker, Heidy Brandon, Beth Coad, Sharne Limb, Sarah Cable, Tanya Davis, Dr Helen Hanson, Dr Katie Snape

South West Thames Clinical Genetics Service, St George's Hospital, Tooting

All 1316 patients on the South West Thames Regional Genetics BRCA Carrier Register were invited to an information day hosted by the genetics team. Carriers were invited by letter and were asked to register on Eventbrite for the 100 available places, on a first come first served basis. The day consisted of a mix of talks given by health professionals, researchers and charity representatives. The topics covered included ovarian cancer management options, research opportunities, the management of psychological issues associated with menopause and communication with children.

On the day we asked our BRCA carriers three questions:
1. What has helped them the most since finding out they were a carrier
2. What they wish they had known when they first found out they were a carrier
3. Suggestions for ways to improve our service.

We also asked patients to complete a feedback form at the end the day.

Results: 93 BRCA carriers attended our patient information day. 98% of the carriers strongly agreed, or agreed that the day had been a positive experience. A number of themes came out from their responses to the questions above, including patients valuing the contact they have had with their genetic counsellor, patients wishing they knew more about the support available when they got their result and to have better understood the impact the result would have for their family. Carriers were incredibly grateful that we had arranged this information day and wish to have more regular information days arranged in the future, and more regular updates and follow up appointments with genetics.

Conclusions: Attendee feedback from a BRCA patient information day was overwhelmingly positive. Speakers and patients requested regular patient information days and a summary on our website to help them keep up to date with current advice and research. We also had a large number of requests from patients for an appointment with the genetic service following the information day. We share our experience of organising the day, the challenges we encountered and we reflect on ways we can continue to improve the ongoing support we provide to our carriers.

Unique: working with genetics professionals worldwide to support families affected by rare chromosome and genomic disorders

Sarah Wynn, Arti Patel, Beverly Searle

Unique, Cardiff University

Unique was founded in 1984 as the Trisomy 9 Support Group but soon expanded to include all rare chromosome disorders. Over the subsequent years, with rapid advances in genomic technologies and the clinical introduction of microarrays and more recently sequencing, Unique has now expanded to support those individuals with copy number variants and autosomal dominant novel genomic variants linked to developmental disorders. Unique works worldwide, offering contact, support and easy-to-read information to families affected by these disorders and with the professionals who work with them. At present (July 2019), Unique has 20220 member families with a rare chromosome or genomic disorder, representing 22715 affected individuals. Unique works with genetics professionals in a myriad of ways. Unique holds detailed developmental, health, behavioural, educational and social information on these members in a confidential database - issues that are important to families in caring for their affected children over a lifetime. This database can be utilised to provide professionals with anonymised phenotypic information charting the natural histories of Unique members with a rare chromosome or genomic disorder.

Families receiving a diagnosis of a rare chromosome or genomic disorder frequently experience severe distress. This distress is compounded by a lack of information about the lifetime effects of these disorders, e.g.two thirds of families receiving a diagnosis of a rare chromosome disorder for their child reported of a lack of medical information. To attempt to answer the types of question that families need answered and that go way beyond solely medical information, Unique has drawn on its confidential database together with the published medical literature to produce more than 250 information guides on a wide range of rare chromosome and genomic disorders. These guides have been written, reviewed and edited in collaboration with a vast army of genetics professionals including genetic counsellors, generously working pro bono. Written information such as Unique's information guides can relieve anxiety and stress and improve understanding. Unique has members in 110 different countries and, to ensure that these information guides are freely accessible (www.rarechromo.org) to as many people as possible, works with bilingual genetic or medical professionals to translate as many as possible into other languages.

With wider access worldwide to genomic testing, Unique membership is expanding rapidly. Working together with both patients and genetics professionals, Unique offers support and information to every family so that the isolation and distress of receiving a rare genetic diagnosis may be alleviated.

Notes

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Speaker & Delegate List

Irene Abreu Rodriguez
Reference Laboratory, S.A
iabreu@referencelaboratory.es

Fizza Akbar
Aga Khan University Hospital
fizza.akbar@aku.edu

Elizabeth Alexander
Manchester Centre for Genomic Medicine
elizabeth.alexander@mft.nhs.uk

Anwar Alharban
Private
Anwar.alharban@yahoo.com

Martha Antwi
Bonsua Government Health Centre
afyafrema99@gmail.com

Stephanie Asher
The University of Pennsylvania
stephanielbyers@gmail.com

Jehannine Austin
University of British Columbia
jehannine.austin@ubc.ca

Cecilia Avila
Stony Brook University Hospital
cecilia.avila@stonybrookmedicine.edu

Samantha Ayres
Victorian Clinical Genetics Services
sam.ayres@vcgs.org.au

Tina Babineau Sturk
LifeLabs
tina.babineausturk@lifelabs.com

Oluwatayo Bamidele
University College Hospital, Ibadan
tayo.moyin@yahoo.com

Elena Baranova
RMACPE
baranova.gen@gmail.com

Helen Batchelor-Regan
Birmingham Women's and Children's Hospital
helen.batchelor-regan@nhs.net

Barbara Biesecker
RTI International
bbiesecker@rti.org

Patricia Birch
University of British Columbia
patricia.birch@ubc.ca

Michelle Bishop
Health Education England
Michelle.Bishop@hee.nhs.uk

Felicity Boardman
Warwick Medical School
felicity.boardman@warwick.ac.uk

Gerdien Bosman
University Medical Centre Groningen
g.bosman@umcg.nl

Laura Boyes
West Midlands Regional Genetics
laura.boyes@nhs.net

Jacqueline Boyle
Hunter Genetics
jackie.boyle@health.nsw.gov.au

Matilda Bradford
Royal Devon & Exeter NHS Trust
matildabradford@nhs.net

Deanna Brockman
Massachusetts General Hospital
deanna.brockman@mgh.harvard.edu

Claire Brooks
Liverpool Centre for Genomic Medicine
Claire.Brooks@lwh.nhs.uk

Tootie Bueser
King's College London
tootie.bueser@nhs.net

Abrar Buhlaiqah
NHS Greater Glasgow and Clyde
2302414B@student.gla.ac.uk

Stephanie Burcher
St George's Hospital
stephanie.burcher@nhs.net

Sarah Cable
St Georges NHS Trust
cable.sarah@btinternet.com

Caty Carrera
Stroke Pharmacogenomics and Genetics Lab
caty.carrerav@gmail.com

Kate Carter
Nottingham Clinical Genetics
katherine.carter@nuh.nhs.uk

Gemma Chandratillake
Eastern Genomic Lab Hub
g.brown.97@cantab.net

Megan Cho
National Institutes of Health
megan.cho@nih.gov

Louise Christie
Hunter Genetics
louise.christie@health.nsw.gov.au

Emily Clarke
Genetic Disorders UK
ekmc2014@gmail.com

Nuala Cody
Dept. of Clinical Genetics
nuala.cody@olchc.ie

Naama Nahama Cohen Kfir
Bar Ilan University
nehamakfir@gmail.com

Gaya Connolly
St George's Hospital
gayaconnolly@gmail.com

Harriet Copeland
Royal Devon and Exeter NHS Foundation Trust
harriet.copeland1@nhs.net

Christophe Cordier
SYNLAB
christophe.cordier@synlab.com

Alessia Costa
King's College London
alessia.costa@kcl.ac.uk

Adriana Costal Tirado
Hospital Parc Taulí
adrianact6@hotmail.com

Alice Coulson
Guy's Hospital
alice.coulson@gstt.nhs.uk

Kathleen Dady
Great Ormond Street Hospital
kfdady@gmail.com

Megan Dancer
Birmingham Women's Hospital
megan.dancer@nhs.net

Pooja Dasani
Guy's and St Thomas's NHS foundation trust
pooja.dasani@nhs.net

Jill Davies
GeneMatters
jilldavies@gene-matters.com

Eleanor Davies
Addenbrooke's Hospital
eleanor.davies1@addenbrookes.nhs.uk

Tanya Davis
St. George's Hospital
Tanya.davis001@gmail.com

Varisha Desai
University of Glasgow
2345590d@student.gla.ac.uk

Carolyn Dickinson
University of Rochester Medical Center
carolyn.dickinson@villanova.edu

Laura Diskin
Maccabi Health services
laura28@gmail.com

Emily Dixon
Guy's and St Thomas' NHS Foundation Trust
emilyvdixon.93@gmail.com

Kelly C Donahue
Genetic Support Foundation
kdonahue@geneticsupport.org

Elisabeth Dramstad
Oslo university hospital
uxdrel@ous-hf.no

Jacqueline Dunlop
NHS Tayside
jacquelinedunlop@nhs.net

Samantha Edwards
Harry Perkins Institute of Medical Research-UWA
samantha.edwards@uwa.edu.au

Fatima Elmugadam
Institute of Endemic diseases
fatimahakam49@gmail.com

Katarina Englund
Centre for Cardiovaskular Genetics
katarina.englund@regionvasterbotten.se

Lori Erby
National Human Genome Research Institute
lori.erby@nih.gov

Nezzy Evans
Liverpool Women's Hospital
nezzy.evans@lwh.nhs.uk

Flavia Facio
National Institutes of Health
flavia.m.facio@gmail.com

Hannah Fagg
UH Bristol
hannahfagg28@gmail.com

Katie Fiallos
Johns Hopkins University
kfiallo1@jhmi.edu

Julia Finch
Genomic Medicine, Liverpool
julia.finch@lwh.nhs.uk

Andrea Forman
Genetic Counsellor
aformanCGC@gmail.com

Elisabeth Forsman
Centre for Cardio vascular Genetics
elisabeth.forsman@regionvasterbotten.se

Lyndon Gallacher
Victorian Clinical Genetics Services
Lyndon.Gallacher@vcgs.org.au

Carmen Garrido
GENyO Center
carmen.garrido@genyo.es

Barbara Girerd
APHP
barbara.girerd19@gmail.com

Selina Goodman
University of Plymouth
selina.goodman@plymouth.ac.uk

Inge Goovaerts
University Hospital Antwerp
inge.goovaerts@uza.be

Jennifer Gorrie
NHS Greater Glasgow & Clyde
jennifergorrie@nhs.net

Abby L Grant
Children's Health Ireland
slane25@gmail.com

Hayley Green
Brigham and Women's Hospital
hgreen7@bwh.harvard.edu

Jennifer Hammond
Great Ormond Street Hospital
jennifer.hammond@ucl.ac.uk

Alia Hashmi
St Georges, University of London
ahashmi12@hotmail.com

Kirsten Henderson
NHS Tayside
kirsten.henderson@nhs.net

Laura Hercher
Sarah Lawrence College
Lhercher@aol.com

Jackie Hill
Cardiff and Vale UHB
jackiewedderburn@hotmail.com

Jan Hodgson
University of Melbourne
hodgson@unimelb.edu.au

Lauren Hogan
Beth Israel Deaconess Medical Center
lauren.hogan24@gmail.com

Julie Horsting
University of Texas Health Science Center at San Antonio
jmhorsting@gmail.com

Esther Horton
NUH
estherlhorton@yahoo.com

Catherine Houghton
Liverpool Genomic Medicine
catherine.houghton@lwh.nhs.uk

Lok Chung Hui
The University of Hong Kong
huilok@connect.hku.hk

Angela Iley
NHS Greater Glasgow & Clyde
angela.iley@ggc.scot.nhs.uk

Charlotta Ingvoldstad Malmgren
Karolinska University Hospital
charlotta.ingvoldstad-malmgren@sll.se

Leila Jamal
National Institutes of Health
leila.jamal@nih.gov

Harsha Jani
Omionics Ltd
Thesis25@gmail.com

Kiley Johnson
GeneMatters
kileyjohnson@gene-matters.com

Galen Joseph
U of California, San Francisco
galen.joseph@ucsf.edu

Ali Kay
Cardiff University
KayAC@cardiff.ac.uk

Stephen Kearney
University College Dublin
Skearney82@gmail.com

Stephanie Kearton
Royal North Shore Hospital
stephanie.kearton@health.nsw.gov.au

Kamron Khan
Leeds Teaching Hospitals
medknk@leeds.ac.uk

Robin King
PWNHealth
rking@pwnhealth.com

Lotte Klansoe
Royal Hospital of Copenhagen
lotte.klansoe@regionh.dk

Emily Lamb
All Wales Medical Genomics Service
emily.lamb@wales.nhs.uk;
emily.r.lamb88@gmail.com

Michelle Lane
IWK Health Centre
lane.michellem@gmail.com

Valentina Lapaeva
Russian Academy of Science
lapaeva07@mail.ru

Alexandra Lebensohn
National Institutes of Health
alexandra.lebensohn@nih.gov

Sara Levene
Centre for Reproductive & Genetic Health
sara.levene@crgh.co.uk

Lauren Limb
NW Thames Regional Genetics
lauren.limb@nhs.net

Joana Lindbom Gomes
NHS Grampian
joana.gomes@nhs.net

Caroline Lintott
Genetic Health Service NZ
Caroline.Lintott@cdhb.health.nz

Wenche Listol
Haukeland University Hospital
wenche.listol@helse-bergen.no

Gina Liu
Kaiser Permanente
gina.liu@kp.org

Marianne Lodahl
Rigshospitalet Denmark
marianne.lodahl@regionh.dk

Aude LOMBARD
Institute of Pathology and Genetics
aude.lombard@ipg.be

Mark Longmuir
NHS Greater Glasgow & clyde
mark.longmuir@ggc.scot.nhs.uk

Birte Lundhaug
Haukeland University Hospital
birte.lundhaug@helse-bergen.no

Fiona Lynch
Murdoch Children's Research Institute
fiona.lynch@mcri.edu.au

Elly Lynch
Melbourne Genomics Health Alliance
elly.lynch@melbournegenomics.org.au

Rebecca Macintosh
Sydney Children's Hospital
bekmac@emailme.com.au

Rhona MacLeod
Manchester Centre for Genomic Medicine
rhona.macleod@mft.nhs.uk

Malebo Malope
University of Cape Town
mlpmal005@myuct.ac.za

Manami Matsukawa
Kyoto University
matsukawa.manami.43a@st.kyoto-u.ac.jp

Jessica Matthews-Kelly
Cambridge University Hospital
jessmk20078@gmail.com

Marion McAllister
Cardiff University
mcallistermf@cardiff.ac.uk

Belinda McClaren
Murdoch Children's Research Institute
belinda.mcclaren@mcri.edu.au

Alison McEwen
University of Technology Sydney
alison.mcewen@uts.edu.au

Claire McLaughlin
Kyowa Kirin International
cfm_mcl@hotmail.com

Hema McSara
YRGS (Leeds)
hema.mcsara@nhs.net

Dawn Melville
Liverpool Centre for Genomic Medicine
d.melville55@hotmail.co.uk

Athalie Melville
Wessex Clinical Genetics Service
athalie.melville@nhs.net

Álvaro Mendes
i3S, University of Porto
alvaro.mendes@ibmc.up.pt

Katrina Merrifield
Birmingham Women's hospital
katrina.merrifield@nhs.net

Sylvia Metcalfe
MCRI & University of Melbourne
sylvia.metcalfe@mcri.edu.au

Anna Middleton
Wellcome Genome Campus
am33@sanger.ac.uk

Kelly Minks
University of Rochester
kelly_minks@urmc.rochester.edu

Baharak Mohammadi
UCL
Baharak.mohammadi.16@ucl.ac.uk

Lorena Moreno Calle
Hospital Clinic Barcelona
lomoreno@clinic.cat

Natalie Moreton
Manchester Centre for Genomic Medicine
natalie.moreton@mft.nhs.uk

Cathryn Moss
The Royal Marsden Hospital
cathryn.moss@rmh.nhs.uk

Arijit Mukhopadhyay
University of Salford
a.mukhopadhyay@salford.ac.uk

Anne MURPHY
HUG Genève
anne.murphy@hcuge.ch

Jessica Myring
N/A
Jessicamy48@gmail.com

Maria Mercedes Navarro de Miguel
GENIFEN
genifentotal@gmail.com

Shivani Nazareth
Clear Genetics
shivani@cleargenetics.com

Danielle Newby
The University of Cardiff
danielle.laetitia@gmail.com

Joshua Nolan
Oxford University Hospitals NHS Foundation Trust
joshua11nolan@gmail.com

Emma O'Donoghue
Cardiff University
odonoghuee@cardiff.ac.uk

Judith Ansaa OsaeLarbi
West Africa Genetic Medicine Centre
gjlsprings@yahoo.com

Denise Oxnard
Greater Glasgow & Clyde NHS
denise.oxnard@ggc.scot.nhs.uk

Christina Palmer
UCLA
cpalmer@mednet.ucla.edu

Gayathri Parasivam
Children's Hospital at Westmead
gayathri.parasivam@health.nsw.gov.au

Belen Pastor
Made of Genes
belen3090@gmail.com

Christine Patch
Genomics England
christine.patch@genomicsengland.co.uk

Annette Patterson
Cancer Genetics
annetterp@sbcglobal.net

Rebecka Pestoff
Centre for Rare Diseases
rebecka.pestoff@regionostergotland.se

Amanda Pichini
University Hospitals Bristol NHS Foundation Trust
amanda.pichini@nhs.net

Heather Pierce
Addenbrooke's NHS Trust
heather.pierce@addenbrookes.nhs.uk

Josep Pla
IVI
josep.pla@ivirma.com

Nadia Preitner
NW Thames Regional Genetics
Nadia.preitner@nhs.net

Sarah Pugh
Bristol Clinical Genetics
sarahpugh@nhs.net

Claire Radford
Birmingham Women's Hospital
claire.radford@nhs.net

Lone W Rasmussen
University Hospital of Aarhus
loneramu@rm.dk

Joyce Refuge
UZ Brussel
cmg@uzbrussel.be

Jennifer Rietzler
Genetic Support Foundation
jrietzler@geneticsupport.org

Roberta Rizzo
University Hospital Southampton
roberta.rizzo@nhs.net

Sarah Robart
Great Ormond Street Hospital
sarah.robart@gosh.nhs.uk

Jonathan Roberts
Wellcome Genome Campus/Addenbrooke's
jr23@sanger.ac.uk

Eulàlia Rovira Moreno
Hospital Vall d'Hebron
eroviramoreno@gmail.com

Erin Royer
Sanford Health
erin.royer@sanfordhealth.org

Nathalie Saen
CMG Antwerpen
nathalie.saen@uza.be

Karen Sage
CARE Fertility UK
karen.sage@carefertility.com

Diana Salinas
Hospital Sant Joan de Déu
salinaschaparro.diana@gmail.com

Glykeria SAMOLADA
Embryolab AG
g.samolada@embryolab.eu

Jette Sandgaard
University Hospital of Aarhus
jetsandg@rm.dk

Urvi Savania
Oxford Centre for Genomic Medicine
urvi.savania@ouh.nhs.uk

Kari Schou
Rigshospitalet
kari.schou@regionh.dk

Shiri Shkedi Rafid
Hadassah Medical Center
shiri.shkedi@gmail.com

Elen Siglen
Haukeland University Hospital
elen.siglen@helse-bergen.no

Ileen Slegers
UZ Brussel
cmg@uzbrussel.be

Nandini Somanathan
West of Scotland Genetics Service
Nandini.Somanathan@ggc.scot.nhs.uk

Bethany Stafford Smith
Cardiff University
bstaff04@gmail.com

Siv Hege Stemshaug
KG Jebsen center for genetic epidemiology
shstemshaug@gmail.com

Ayaka Suzuki
UC Davis Medical Center
ayzsuzuki@ucdavis.edu

Virginie Szymczak
University Hospital of Ghent
virginie.szymczak@ugent.be

Chieko Tamura
FMC Tokyo Clinic
c_tamura@t3.rim.or.jp

Dagmar Tapon
Imperial College Healthcare NHS Trust
dagmar.tapon@nhs.net

Nicki Taverner
Cardiff University and All Wales Medical Genetics Service
tavernern@cardiff.ac.uk

Johanna ter Beest
University Med. Centre Groningen
j.g.ter.beest@umcg.nl

Claudia Terry
Kaiser Permanente
claudiavanessaterry@gmail.com

Hanne Teule
University Hospital Louvain
hanne.teule@uzleuven.be

Isobel Turbin
CUH NHS Foundation Trust
Isobelturbin@gmail.com

Bente Udsen
Klinisk Genetisk Afdeling
benuds@rm.dk

Gisela Urgel Reig
NHS Tayside
gisela.urgelreig@nhs.net

Kathryn Van Diemen
TrakGene Pty Ltd
kvandiemen@trakgene.com

Angela van Remortele
Genome Diagnostics Nijmegen Maastricht
angela.vanremortele@radboudumc.nl

Anja Vestergaard
Klinisk Genetisk Afdeling
anjavest@rm.dk

Sally Watts
Guy's and St Thomas' Hospital
sally.watts@gstt.nhs.uk

Jon Weil
California St Univ. Stanislaus
weilj@berkeley.edu

Jonathan Wells
Cardiff University
wellsjp1@cardiff.ac.uk

Tina-Marié Wessels
University of Cape Town
tina.wessels@uct.ac.za

Sarah Wilcox
Addenbrooke's hospital, Cambridge
festivalswilcox2@hotmail.co.uk

Tahni Ann Wilson
Queen Elizabeth University Hospital
tahniann@gmail.com

Lizzie Winchester
South West Thames Regional Genetic Service
elizabeth.winchester@stgeorges.nhs.uk

Sarah Wynn
Unique
wynns@cardiff.ac.uk

Katarzyna Zawadzka
MNM Diagnostics
kasia@mnm.bio

Olga Zayts
The University of Hong Kong
zayts@hku.hk

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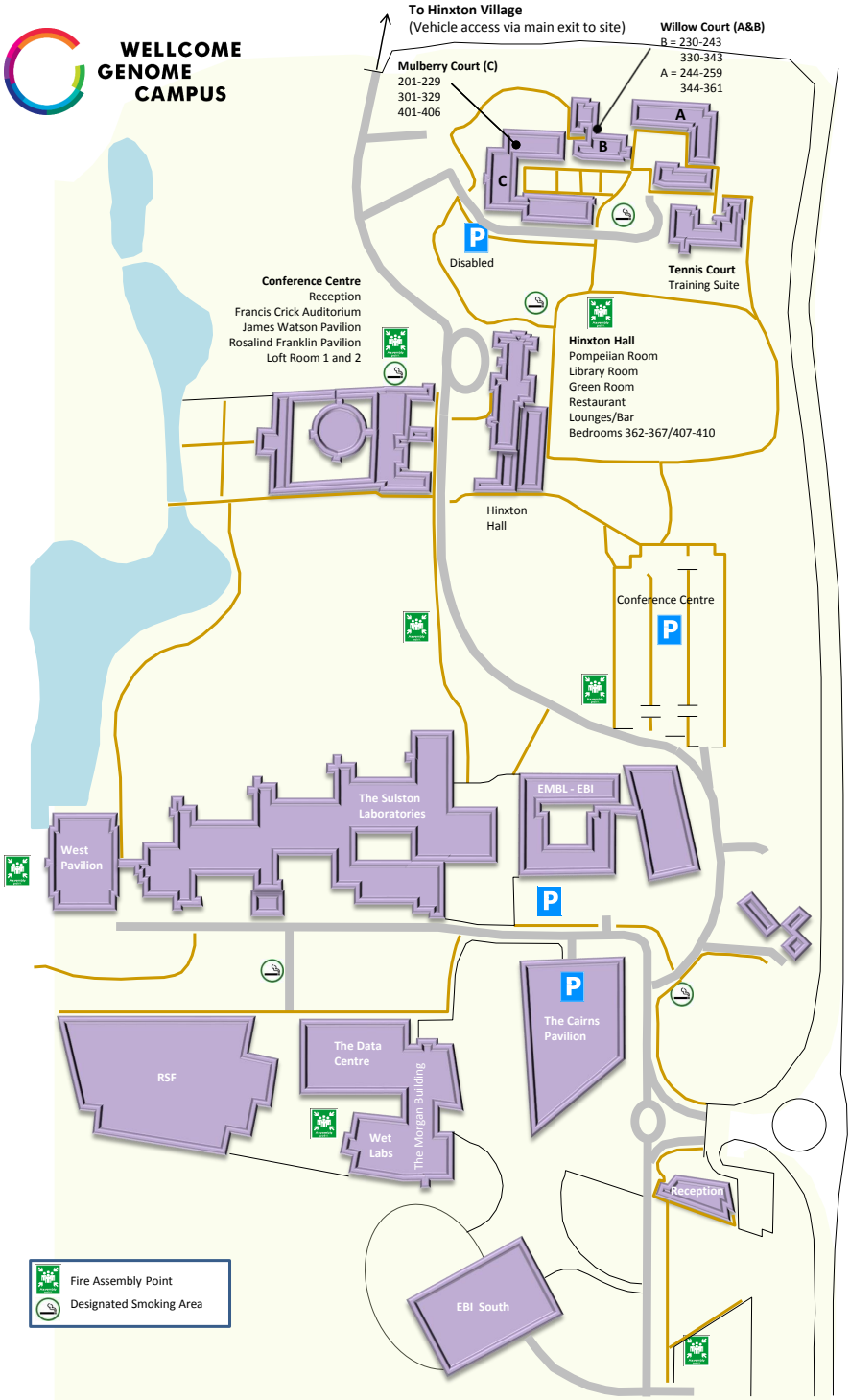
WELLCOME GENOME CAMPUS
To Hinxton Village
(Vehicle access via main exit to site)
Willow Court (A&B)
B = 230-243
330-343
A = 244-259
344-361
Mulberry Court (C)
201-229
301-329
401-406
A
B
C
Disabled
Tennis Court
Training Suite
Conference Centre
Reception
Francis Crick Auditorium
James Watson Pavilion
Rosalind Franklin Pavilion
Loft Room 1 and 2
Hinxton Hall
Pompeiian Room
Library Room
Green Room
Restaurant
Lounges/Bar
Bedrooms 362-367/407-410
Hinxton Hall
Conference Centre
The Sulston Laboratories
EMBL - EBI
West Pavilion
The Cairns Pavilion
RSF
The Data Centre
Wet Labs
The Morgan Building
Reception
EBI South
Fire Assembly Point
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