Name: _____

World Congress on Genetic Counselling

Wellcome Genome Campus Conference Centre, Hinxton, Cambridge, UK
4-6 October 2017

Scientific Programme Committee:

Barbara Biesecker
National Human Genome Research Institute, USA

Anna Middleton
Wellcome Genome Campus, UK

Christine Patch
King's College London and Genomics England, UK

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Conference Organiser

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.

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 **#WCGC17**. 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 and 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, 4 October at 18:00 – 19:30.

Even number poster assignments will be presenting in poster session 2, which takes place on Thursday, 5 October, at 17:30 – 19:00.

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

Filming
We are hoping to catch some of the World Congress on film to make a summary piece about the event. If you do not want us to capture your image in any way, please let us know.

Conference Meals
Lunch and dinner will be served in the Hall. Please refer to the conference programme in this book as times will vary based on the daily scientific presentations.

Please inform the conference organiser if you are unable to attend the conference dinner.

All conference meals and social events are for registered delegates. Please note there will be

no lunch or dinner facilities available outside of the conference timetable.

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.

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.
If you plan to arrive late at night you can check into your room as 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 Red Lion, Hinxton Guests
Check in
is available between 13:00-15:00 and 18:00-23:00 (19:00-22:30 on Sundays) unless special arrangements are made.

Breakfast
Your breakfast will be served in the restaurant from 07:30-08:30 on weekday mornings and 08:30-09:30 on weekend mornings

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

Departures
You must vacate your room by 10:30 on the day of your departure. Please remember return the key fob – there is a £20 replacement fee for non-returns.
A luggage store is available in the Conference Centre please ask at the reception

For Holiday Inn Express, 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
Your breakfast will be served in the hotel, Great Room from 06:30 – 09:30.

Telephone & Internet
A telephone and free wireless internet access is available in your room, wireless is complimentary.
The hotel also offers a relaxed licensed bar and lounge area.

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

Wellcome Genome Campus Scientific Conferences guests receive a 15% discount on food at the Red Lion, Whittlesford Bridge Hotel.

Transfers
For those of you staying off campus, we have a complimentary shuttle bus running to and from the Holiday Inn Express in Duxford organised with Richmond's Coaches. The shuttle collection point is next to Whittlesford Parkway train station, where regular trains operate to Cambridge station. The shuttle service is as follows:

Wednesday 4 October	
Holiday Inn Express – Wellcome Genome Campus	12:30
Wellcome Genome Campus – Holiday Inn Express	21:20
Thursday, 5 October	
Holiday Inn Express – Wellcome Genome Campus	08:30
Wellcome Genome Campus – Holiday Inn Express	21:20
Friday, 6 October	
Holiday Inn Express – Wellcome Genome Campus	08:30

Taxis
Please find a list of local taxi numbers below:

All Journeys
Cam-Air-Connect (Airport Specialist)
bookings@cac-bookingroom.com
+44 (0) 1223 750 850

Sawston Cab Co Ltd (Airport Specialist)
info@sawstoncabcoltd.co.uk
+44 (0)1223 517008

For Cambridge & the airports
Panther Taxis
www.panthertaxis.co.uk +44 (0)1223 715715

For Audley End & Great Chesterford Station
Walden Cabs +44 (0)1799 500500
Crocus +44 (0)1799 525511

For Whittlesford Station & The Holiday Inn Express
Mid Anglia +44 (0)1223 836000
Caz Cars +44 (0) 1223 513693

Return Ground Transport
Complimentary return transport has been arranged for 14:45 on Friday, 6 October to: Cambridge station and city centre, Stansted and Heathrow airport

Please note: a sign-up sheet will be available at the registration desk. Places are limited so you are advised to book early.
Please allow a 30-minute journey time to both Cambridge and Stansted Airport and 2.5 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, 4 October

12:30	Coach departs from Holiday Inn Express & Whittlesford Parkway station
12:30-13:50	Registration with lunch
13:50-14:00	Welcome and Introduction
14:00-15:00	Keynote lecture by *Bob Resta, Swedish Medical Centre, USA*
15:00-16:00	Session 1: The public: understanding of genomics – before becoming a patient
16:00-16:30	Afternoon tea
16:30-17:45	Session 2: Becoming a patient: the first conversations about genomics
18:00-19:30	Poster Session 1 (odd numbers) with Drinks Reception
19:30	Dinner & Cash bar
21:20	Coach departs from Holiday Inn Express & Whittlesford Parkway station

Thursday, 5 October

08:30	Coach departs from Holiday Inn Express & Whittlesford Parkway station
09:00-10:30	Session 3: Counselling under scrutiny
10:30-11:00	Morning coffee
11:00-12:30	Session 4: Inside genetic counselling
12:30-14:00	Lunch
14:00-15:30	Session 5: Outcome measurements
15:30-16:00	Afternoon tea
16:00-17:30	Session 6: Debate session – How & when are psychotherapeutic models relevant in the genomic era
17:30-19:00	Poster session 2 (even numbers) with drinks reception
19:00	Conference dinner & Cash bar
21:20	Coach departs from Holiday Inn Express & Whittlesford Parkway station

Friday 6 October

08:30	Coach departs from Holiday Inn Express & Whittlesford Parkway station
09:00-10:30	Session 7: Ethical, legal and social issues
10:30-11:00	Morning coffee
11:00-12:00	Keynote lecture by *Clara Gaff*, *University of Melbourne, Australia*
12:00-13:35	Session 8: Spreading the word post consultation
13:35 -14:45	Lunch
14:45	Coaches depart to Cambridge city centre and train station & Heathrow airport via Stansted airport

Conference Sponsors

We would like to acknowledge the generous support from the following organisations:

Exhibitors:

www.Illumina.com

http://www.genomediagnosticsnijmegen.nl

www.undiagnosed.org.uk

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www.gcrb.org.uk

World Congress on Genetic Counselling

Wellcome Genome Camps Conference Centre,
Hinxton, Cambridge

4 – 6 October 2017

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, 4 October

12:30-13:50 Registration with lunch

13:50-14:00 Welcome and Introduction
Anna Middleton
Wellcome Genome Campus, UK

14:00-15:00 **Keynote lecture**
Chair: Anna Middleton, Wellcome Genome Campus, UK

What have genetic counselors been doing and have they been any good at it?
Bob Resta
Swedish Medical Centre, USA

15:00-16:00 **Session 1: The public: understanding of genomics – before becoming a patient**
Chair: Christine Patch, King's College London and Genomics England, UK

15:00 Socialising the genome –how to make genomics resonate for the public
Anna Middleton
Wellcome Genome Campus, UK

15:30 Genomics in society – attitudes of the Australian public to personal genomics
Sylvia Metcalfe,
MCRI & University of Melbourne, Australia

16:00-16:30 Afternoon tea

16:30-17:45 **Session 2: Becoming a patient: the first conversations about genomics**
Chair: Clara Gaff, University of Melbourne, Australia

16:30 The UK 100,000 genomes project: views, expectations, and experiences of the first patients recruited
Sandi Dheensa
CELS, University of Southampton, UK

17:00 Parents' and adolescents' reasoning related to receiving secondary variants from whole genome sequencing
Robin Hayeems
The Hospital for Sick Children Research Institute, Canada

17:30 Parental interest in genomic sequencing of newborns: Enrollment experience in the BabySeq project
Shawn Fayer
Brigham and Women's Hospital, Boston, USA

17:45 How do consent forms for diagnostic next-generation sequencing address unsolicited and secondary findings? A qualitative content analysis
Danya Vears
KU Leuven, Belgium

18:00-19:30 **Poster Session 1 (odd numbers) with Drinks Reception**

19:30 Dinner & Cash bar

Thursday, 5 October

09:00-10:30 **Session 3: Counselling under scrutiny**
Chair: Tara Clancy, University of Manchester, UK

09:00 Adapting evidence based strategies for effective communication in cancer genetic counseling
Robin Lee
University of California, San Francisco, USA

09:30 The 'counselling' in genetic counselling: empirical evidence
Tina-Marie Wessles
University of Cape Town, South Africa

10:00 Counselors' experiences with uncertainties in counseling about multigene panel testing for cancer. A focus group study
Niki Medendorp
Academic Medical Center, Netherlands

10.15 Next-generation counseling: a model for non-invasive prenatal screening results disclosure and patient management
Carrie Haverty
Counsyl, USA

10:30-11:00 Morning coffee

11:00-12:30 **Session 4: Inside genetic counselling**
Chair: Barbara Biesecker, NIH, USA

11:00 New interventions to facilitate family communication about genetic risk and preparing the workforce
Alison Metcalfe
Kings College London, UK

11:30 Facilitating personalized communication in genetic counselling for Cancer
Eveline Bleiker
Netherlands Cancer Institute, The Netherlands

12:00 Challenges for genetic counseling of families with dilated cardiomyopathy and truncating titin mutations
Renee Johnson
Victor Chang Cardiac Research Institute, Australia

12:15 Psychological impact and genetic counselling preferences of multi-gene cancer testing in a Spanish multi-centric cohort: a 12-month analysis
Irene Esteban
Ninewells Hospital, UK

12:30-14:00 Lunch

14:00-15:30 **Session 5: Outcome measurements**
Chair: Marion McAllister, Cardiff University, UK

14:00 What counts as 'success' in genetic counselling? Systematic review of RTCs in assessing outcomes in genetic counselling?
Barbara Biesecker
NIH, USA

14:30 Measuring the economic value of genetic counselling
Katherine Payne
University of Manchester, UK

15:00 Using data collected using patient-reported outcome measures for quality improvement in clinical genetics
Marion McAllister
Cardiff University, UK

15:15 The ongoing search for a patient reported outcome measure (PROM) in genetic counseling: the psychometric properties of the Genetic Counseling Outcome Scale for a Dutch study sample
Jan Voorwinden
University Medical Center Groningen, Netherlands

15:30-16:00 Afternoon tea

16:00-17:30 **Session 6: Debate session – How & when are psychotherapeutic models relevant in the genomic era**
Chair: Anna Middleton, Wellcome Genome Campus, UK

Jehannine Austin, University of British Columbia, Canada
Tara Clancy, University of Manchester, UK

17:30-19:00 **Poster session 2 (even numbers) with drinks reception**

19:00 Conference dinner & cash bar

Friday 6 October

09:00-10:30 **Session 7: Ethical, legal and social issues**
Chair: Christine Patch, King's College London and Genomics England, UK

09:00 Bioethics and genetic counselling
Heidi Howard
Uppsala University, Sweden

09:30 The duty to inform: HD case legal going through the English courts
Vicky Chico
University of Sheffield & Wellcome Genome Campus, UK

10:00 BME women's decision-making for risk-reduction after BRCA testing
Mavis Machirori
King's College London, UK

10.15 Reporting clinical whole genome sequencing results via telephone genetic counseling in a diverse healthy population
Molly McGinniss
Illumina, USA

10:30-11:00 Morning coffee

11:00-12:00 **Keynote lecture**
Chair: Christine Patch, King's College London and Genomics England, UK

The priorities for research in genomic counselling
Clara Gaff
University of Melbourne, Australia

12:00-13:35 **Session 8: Spreading the word post consultation**
Chair: Bob Resta

12:00 Genetics in popular culture
Jon Roberts
Wellcome Genome Campus, UK

12:30 Personal Genomic Screening: how best to facilitate preparedness of future clients
Jane Flemming,
Sydney University Medical School – Northern

12:45 Panel discussion: The future of the genetic counselling profession
Clara Gaff, Tina-Maria Wessles, Christine Patch, Jehannine Austin

13:30 Closing remarks and conference summary
Programme Committee

13:35 -14:45 Lunch

14:45 Coaches depart to Cambridge city centre and train station & Heathrow airport via Stansted airport

Spoken Presentations

What Have Genetic Counselors Been Doing And Have They Been Any Good At It?

Robert Resta

Swedish Medical Center, Seattle, WA USA

The effectiveness of genetic counseling has been assessed by different measures since Sheldon Reed first coined the term in 1947. These measures of effectiveness reflect temporal changes in the perceived goals of genetic counseling, as well as changes in social, medical, professional, and ethical factors.

This presentation reviews and critiques the historical changes that have occurred in research addressing the effectiveness of genetic counseling over the last 70 years, with attention to underlying factors that influenced the choice of these measures. The historical development will be broken down into 3 broad periods, and the somewhat arbitrary reasons for this particular categorization will be explained:

1) 1947-1982, when the nearly universal measures of effectiveness were patient recall of recurrence risks and its effect on reproductive choices
2) 1982-1995, which saw a broadening of the goals to include measures how effectively genetic counseling reduced anxiety and stress
3) 1995 – Present, when other psychosocial and adaptational measures were introduced as genetic counseling moved beyond being a service concerned primarily with reproductive decisions.

The discussion will end with questions and issues that should be taken into consideration as research into the effectiveness of genetic counseling enters the third decade of the 21st century.

Notes

Socialising the genome –how to make genomics resonate for the public

Anna Middleton

Wellcome Genome Campus, Hinxton, Cambridge, CB10 1SA, UK

Genomics ('Geno-what'?) is a term that 82% of the British public have not heard of. This creates a challenge when conducting public attitude research. The thematic analysis of 6 focus groups with members of the British public reveals some novel 'conversation starters' about genomics that could be used in public engagement. Building on these we created films that describe various aspects of genomic data sharing. These films sit within a survey, 'Your DNA, Your Say', we designed for the Global Alliance for Genomics and Health. The films give participants the necessary background to be able to answer the questions (without biasing them). The survey gathers attitudes towards genomic data donation and the perceived harms associated with this. It has been translated into 14 languages and is on track to be the largest global survey of public opinion on genomic data donation.

Results from the English-speaking survey (n = 9742) offer views from a representative UK, USA, Canada and Australia public. We show that familiarity with DNA, genetics and genomics is at the heart of a willingness to donate one's genomic data for use by others. Those who are most unfamiliar with the concepts are least likely to want to donate their data. English-speaking publics are able to articulate their fears about data donation, but this does not affect their willingness to donate.
The results of the survey will feed into the work of GA4GH, including the development of new policies to address the ethical and moral questions – both personal and political – about how we use people's genetic information. The survey can be found here: www.YourDNAYourSay.org.

Notes

Genomics in society – attitudes of the Australian public to personal genomics

Sylvia Metcalfe[1] for the Genioz study team

1 Murdoch Children's Research Institute and The University of Melbourne

Online personal genomic testing (PGT) is offered for a diverse range of purposes, including health, nutrition, pharmacogenetics, sporting ability, physical traits, ancestry, behaviours, personalities and even social attributes. PGT is available globally and marketing in Australia has grown in the last few years, as is PGT offered through complementary/alternative health practitioners. Research about Australians' attitudes to and experience with personal genomics has been limited. Therefore, to explore the Australian public's perceptions of these tests, a multi-stage study called Genioz (Genomics: National Insights of Australians) has conducted focus groups, an online survey, interviews and deliberative workshops. Approximately 3000 Australians have participated in one of these activities.

Although the genetics community refers to these online tests as "direct-to-consumer", many participants in the focus groups did not recognise this term. Rather, they were able to conceptualise the term "personal genomics", which echoes the language and rhetoric used by the testing companies that focus on "personal", "your DNA", "unique". About two-thirds of survey respondents had heard of PGT previously. Of 2539 Australian respondents, 22.5% (572) reported having had some type of genetic test, with 8% (202) having had online testing only, and 36.4% (925) were interested in having PGT for health-related and/or "recreational" reasons. The majority of online tests were for ancestry DNA testing. Some interviewees who had ancestry DNA testing discussed downloading their raw data to conduct further analysis using online genealogical software or other software to obtain health information, occasionally taking these results to a health practitioner for interpretation. Some interviewees had *MTHFR* and other 'genomic wellness' tests, turning to alternative health practitioners for support. The majority of survey respondents said they would prefer healthcare professionals (HPs) to help them understand their health-related results (GPs: 78%; other specialist HPs: 65%; independent genetic specialists: 57%), while 17% would seek help from a company-based HP. About half would even seek help from HPs for non-health related results.

In 2011, a survey of Australian genetic health professionals showed that 11.3% saw clients about PGT results, increasing to 51.1% in 2017. Many are not comfortable discussing PGT results and some services now refuse to do this. Findings from the Genioz study suggest workforce implications, not only for genetic health services but also for GPs and complementary/alternative health practitioners, who often have limited understanding of these types of tests and their clinical significance.

Notes

The UK 100,000 genomes project: views, expectations, and experiences of the first patients recruited

Sandi Dheensa [1] Anneke Lucassen [1 2], Angela Fenwick [1]

1 Clinical Ethics and Law, University of Southampton, Southampton, United Kingdom.
2 Wessex Clinical Genetics Service

The 100,000 genomes project (100kGP) is unprecedented and introduces several practices that are novel for the UK health service, such as the offer of additional findings, and the test being conditional upon also participating in broad research. We have been exploring views, expectations, and experiences of this new venture, using questionnaires and longitudinal interviews with participating patients and families. At abstract submission, we had analysed questionnaires from 450 patients or parents thereof, and conducted interviews with 23 of these respondents, all from the rare disease arm of 100kGP. We analysed qualitative data using framework analysis, focusing on four key areas: project information, diagnosis, additional findings, and research. We analysed our survey data using univariate statistics. Our qualitative research showed that regarding project information, generally, participants had not read or understood the materials provided in great detail. They found it was also challenging to take in the information imparted in the consent session. Participants had realistic expectations about receiving a primary diagnosis. They thought a diagnosis would be of limited value and saw the receipt of good ongoing care in frontline NHS as more important. They worried about receiving a worse diagnosis than they were expecting and about when, how, and by whom results would be delivered. Regarding additional findings, participants felt privileged to have such testing and thought limiting the tests to certain findings was a good idea. However, some would have preferred to receive individual carrier results. Research participation was seen as a privilege, as an act of solidarity with other families affected by rare disease, and as worth doing despite several perceived costs. Participants emphasised the importance of ongoing oversight and governance over their data and research practices. Our survey data reflected the qualitative findings but interestingly showed that between a third and a fifth or participants had identified types of research they would not want their data used for—such as research using animals or embryos or research that could lead to patents. While patients/families spoke positively of whole-genome sequencing and 100kGP, we have identified areas of the consent process that warrant improvement. To this end, we are designing digital tools based on our ongoing research to help families understand their results and, where relevant, communicate them to relatives.

Notes

Parents' and adolescents' reasoning related to receiving secondary variants from whole genome sequencing

Hayeems RZ[1], Anderson JA[1], Byrne R[2], Meyn MS[1], Shuman C[1], Zlotnik Shaul R[1], Mantella LE[3], Szego MJ[1], Bowdin S[1], Kaufman M[1], Sappleton K[1], Chitayat D[1], Monfared N[1]

[1]Hospital for Sick Children, Toronto, ON, Canada; [2]GeneDx, Gaithersburg, MD, USA, [3]Queens University, Kingston, ON, Canada

To inform clinical implementation of whole genome sequencing (WGS) in paediatrics, we aimed to understand parent and adolescent reasoning related to participating in WGS and receiving adult-onset secondary variants (i.e. medically actionable variants that are unrelated to a presenting medical condition but predict adult-onset disease). Embedded within the Hospital for Sick Children's Genome Clinic, a multi-disciplinary platform for translational research in genomics, we conducted qualitative interviews with parents whose children were undergoing WGS and adolescent participants. Interviews probed parents' and adolescents' understanding of WGS, experience with and motivation for pursuing it, preferences related to receiving secondary variants, and associated decision-making strategies. Interviews were transcribed and analyzed thematically. Twenty-three parents of young children and eight additional parent-adolescent pairs participated. Parents of young children supported WGS as a diagnostic test, perceiving clear intrinsic and instrumental value. However, many parents were ambivalent about receiving secondary variants, conveying a sense of self-imposed obligation to take on the 'weight' of knowing their child's adult onset health risks, however unpleasant. Some parents chose to learn about adult-onset variants for their child but not for themselves. Analogous to the notion of 'inflicted insight', we call this phenomenon 'inflicted ought'. Importantly, the finding that not all parents wanted to learn about adult onset variants for themselves challenges one of the underlying justifications for current professional guidelines on reporting secondary findings from genome wide sequencing. Adolescents (age 11-18 years) demonstrated a good understanding of WGS and secondary variants and perceived this information to be valuable to managing their emerging health and life choices; all but one opted to receive secondary variants if identified. Concerns identified related to potential constraints placed on insurance eligibility and fears of being at risk for a disorder that may not be curable. When considering their decision about secondary variants, adolescents used both abstract and concrete thinking strategies, sought input from parents or other valued relationships, considered personal health and familial factors, and endorsed a shared decision making approach. Parents felt their adolescent made informed and capable decisions regarding secondary variants and valued a shared decision making approach. Pre-test counseling strategies should attend to parents' and adolescents' understanding of, motivation towards, and reasoning related to learning secondary variants. Whether, when and for whom it is justifiable to identify adult onset secondary variants in paediatric medicine remains controversial. Findings herein contribute to this ongoing dialogue.

Notes

Parental Interest in Genomic Sequencing of Newborns: Enrollment Experience in the BabySeq Project

Shawn Fayer1, Casie Genetti2, Grace E. VanNoy2, Jill Robinson3, Talia Schwartz2, Stacey Pereira3, Amy L. McGuire3, Ingrid Holm2,5, Pankaj B. Agrawal2, 5, 7, Alan H. Beggs2,5, Robert C. Green1,4,5,6, Richard B. Parad1, 5

1)Brigham and Women's Hospital, Boston, MA. 2)The Manton Center for Orphan Disease Research, Division of Genetics and Genomics, Boston Children's Hospital, Boston, MA. 3)Center for Medical Ethics and Health Policy, Baylor College of Medicine, Houston, TX. 4)Partners Personalized Medicine, Boston, MA. 5)Harvard Medical School, Boston, MA. 6)The Broad Institute of MIT and Harvard, Cambridge, MA. 7)Division of Newborn Medicine, Boston Children's Hospital, Boston, MA

The BabySeq Project is the first randomized clinical trial assessing the impact of providing genomic sequencing (GS) on newborns to their parents. Parents of newborns from well baby nursery (WBN) and intensive care unit (ICU) settings were approached and offered enrollment via a consent session with a genetic counselor (GC). Among parents who were approached, we assessed how often and why participation was declined. Of 4,079 families approached (3,624 parents of WBN and 455 parents of ICU infants), 10% were willing to attend the 1-hour education/consent session (ECS) with a GC. 63% of these families, representing 6% of total approached families, ultimately enrolled. Of the declining families who provided a reason, 67% did not want to participate in any research studies at that time. The remaining 43% reported reasons including: study logistics, overwhelmed after childbirth, discomfort with genetic testing, reluctance to receive unfavorable/uncertain results, concern for privacy and fear of insurance discrimination. Those families that declined study participation after an ECS with a GC were significantly more likely to cite concerns related to unfavorable results, privacy, and insurability compared to those who declined at initial approach, who more often cited logistical concerns, feeling overwhelmed, and disinterest in genetic testing. Despite preliminary data suggesting strong theoretical interest in newborn GS (Waisbren, 2015), execution of a randomized trial offering newborn GS in a postpartum setting yielded far lower study uptake. Participation in ECSs shifted the categories of concerns towards issues of privacy, insurability and emotional effect of results, highlighting the impact of the consent process on enrollment decisions. Understanding motivations of parental apprehension to participate in newborn GS will help shape counseling content, emphasize the important role of GCs in public GS education, and inform policy development around potential population-wide newborn GS.

Notes

How do consent forms for diagnostic next-generation sequencing address unsolicited and secondary findings? A qualitative content analysis

Danya F Vears 1,2, Emilia Niemiec 3,4,5, Heidi Carmen Howard 6 & Pascal Borry 1,2

1 Centre for Biomedical Ethics and Law, Department of Public Health and Primary Care, KU Leuven, Kapucijnenvoer 35 Box 7001, 3000 Leuven, Belgium.
2 Leuven Institute for Human Genomics and Society, 3000 Leuven, Belgium.
3Erasmus Mundus Joint International Doctoral (Ph.D.) Degree Programme in Law, Science and Technology, University of Bologna, Via Galliera 3, 40121 Bologna, Italy.
4 Department of Law, University of Turin, Lungo Dora Siena 100 A, 10153 Turin, Italy.
5 Centre for Ethics and Law in the Life Sciences, Leibniz University Hannover, Am Klagesmarkt 14-17, 30159 Hannover, Germany.
6 Centre for Research Ethics and Bioethics, Uppsala University, Box564, SE-751 22, Uppsala, Sweden.

Despite considerable debate, the question of whether, and to what extent, unsolicited findings (also known as incidental findings) should be returned to patients following next-generation sequencing (NGS) remains unanswered. This is likely partially exacerbated by confusion in the terminology used to describe both disease-causing variants unrelated to the original rationale for testing identified inadvertently (unsolicited findings; UF) and those that are actively searched for (secondary findings; SF). Research indicates that a large proportion of patients and members of the general public are keen to receive UF. However, this is a) not always the case, b) not necessarily feasibly in practice, and c) not necessarily the most responsible approach given the uncertainties related to UF and SF in asymptomatic individuals. Despite this, many authors have suggested that patient choice regarding return of UF and SF should be determined prior to testing.

In light of these complexities, we aimed to analyse consent forms being used for WGS, WES, and/or large NGS panels in the diagnostic setting in order to determine if and how they address reporting of UF and SF, and whether patients (or their parents) are provided with options regarding the return of these results. Forms were primarily identified through systematic online searches. The consent forms were analysed using inductive content analysis where categories were derived from the data, rather than pre-determined.

A total of 54 forms in English met our inclusion criteria, which represented 38 separate institutions from 7 different countries. One quarter of the forms (13/54; 24%) did not mention that findings extraneous to the clinical question might be identified during the course of the analysis. The other forms predominantly referred to "incidental" and "secondary" findings, although "unsolicited", "unexpected", and "co-incidental" were also used. Regarding UF reporting practices, 20 forms indicate that there is a possibility for unsolicited findings to be returned to patients, although which specific types of UF they return varied considerably. Seventeen forms seem to indicate that they actively search for SF. Of these, 2 provide solely 'opt in' options for patients and 6 provide the patient with an option to opt out of receiving SF, often indicating that the laboratory will report SF by default.

The differences we identified between the consent forms used for diagnostic next-generation sequencing suggest important inconsistencies in how UF and SF are addressed or explained to patients. This raises concerns about the quality of informed consent being obtained.

Notes

Adapting Evidenced Based Strategies for Effective Communication in Cancer Genetic Counseling

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Thirty-six percent of Americans have limited health literacy. Gaps in effective communication are widely recognized as a major contributor to health disparities. As criteria for cancer genetic services expand, insurance coverage increases, and costs go down, counselors need strategies to communicate effectively with their increasingly diverse patient population.

To examine current communication practices in cancer risk counseling, we used multiple inductive qualitative methods including systematic direct observation and audio-recording of genetic counseling sessions conducted in English, Spanish and Cantonese (n=170), semi-structured interviews with observed genetic counselors (n=10) and stimulated recall interviews with observed patients (n=51) at two public hospitals.

We identified a fundamental mismatch of patient information needs and information provided by counselors. Components of communication that contributed to this mismatch and resulted in ineffective communication include: (1) provision of information that lacks relevance for the patient; (2) provision of too much information; (3) conceptually difficult presentation of information; (4) imprecise discussion of screening and prevention options. To address these findings, we adapted evidenced based strategies developed in other medical settings, such as teach-back, plain talk, and proven risk communication methods, to the cancer genetic counseling context. In a pilot test, counselors learned about these strategies in a four-hour workshop, and then spent two months in practicing in clinic. Results of the pilot indicate that counselors are able to apply these strategies to improve patient comprehension and engagement.

Our findings indicate a need to transform the standard model of genetic counseling communication. Particularly for pre-test counseling, counselors need to adapt to the communication needs of the increasingly diverse patients who now have access to hereditary cancer testing, including the many with limited health literacy. These findings have been incorporated into a large randomized control trial which is comparing the effectiveness of three modes of genetic counseling (in-person, by phone and over video conferencing), in a diverse patient population at high risk for hereditary breast and ovarian cancer in three public hospitals. This current study will enable us further explore qualitatively and quantitatively what is lost and gained across the three counseling modes in a lower health literacy population.

Notes

The 'counselling' in genetic counselling: empirical evidence

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A genetic counselling session is a complex interaction (communication process). A genetic counsellor (GC) has multiple tasks to perform, education regarding the medical and complex scientific aspects of genetics, consideration of the impact on the patient's health and the implications to the individual, couple, family and culture while taking into account emotional functioning and reactions such as grief, fear and anxiety. The GC has to consider and use this knowledge to facilitate autonomous and informed decision-making. Typically, all this is to be accomplished in one 60-minute session, making it very challenging to maintain a balance between the medical and counselling components of the consultation. To understand the process, studies have been conducted investigating the patients' and/or the GC's (and other healthcare providers) perspective of the genetic counselling session. This has in turn been used to examine the applicability and value of established theories and models. These insights have helped to shape and develop genetic counselling models, such as the reciprocal engagement model of genetic counselling. One source of evidence that has been underutilized in genetic counselling is data from interactional research (IR). IR scrutinizes an interaction by analysing the turn for turn discussion between the participants. Existing international IR on the genetic counselling process has provided insight into risk management, information communication and contextual influences. IR, utilizing principles of conversation analysis, was used to study the process of advanced maternal age (AMA) prenatal genetic counselling in South African. Analysis of these sessions revealed that patient decision-making is not as non-directive as we believe. It was found that despite GCs' non-directive strategies, the women perceived the option of amniocentesis to be an offer that should be accepted. There is also evidence that the women dichotomise the risks irrespective of the numerical values given. These findings seem to be linked to the South African health care setting. The resulting empirical evidence provided insight into the genetic counselling process and although conducted on AMA sessions in one setting, it may be applicable to other settings as the function and role of the GC is similar. Further studies using IR methods, as it has demonstrated its value, may assist with providing a better understanding of genetic counselling, the goals and success of the process, and alternate practice options for the future.

Notes

Counselors' experiences with uncertainties in counseling about multigene panel testing for cancer. A focus group study.

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Next-generation sequencing based panel testing is increasingly used in the cancer diagnostic setting because it enables the analysis of multiple genes to improve the identification of a hereditary predisposition for cancer. However, multigene panels may yield high levels of uncertainty, for example by the increased identification of variants of unknown significance. So far, it remains unknown how counselors experience uncertainties concerning multigene panels and discussing these uncertainties with counselees. Consequently, it is unclear whether and what difficulties counselors experience with these uncertainties. Therefore, we explored what uncertainties counselors perceive, experience and communicate concerning decisions about multigene panel testing. Six focus groups were conducted in six academic medical centers in The Netherlands. In total, 38 counselors participated; group size ranged between 4 and 10. Counselors' socio-demographic characteristics were assessed in a questionnaire. Topics discussed were the uncertainties experienced by counselors as well as dilemma's and needs in discussing these uncertainties with counselees. Focus groups were audio recorded and transcribed verbatim. The transcripts were analyzed inductively by two independent coders. Counselors reported several uncertainties related to multigene panels, such as incidental findings and inconclusive test results. These uncertainties were not necessarily experienced as problematic. Most counselors did however report having difficulty in deciding to what extent and even whether uncertainties should be communicated to patients before testing. This was particularly the case for less experienced counselors. Most counselors reported to inform patients more extensively after the test, in order to restrict the information about uncertainties to those relevant for that specific patient. Counselors indicated a need for more consensus between counselors and between centers about the extent and manner of communicating uncertainties before testing. These findings warrant further investigation on how best to communicate uncertainties concerning multigene panel testing before testing.

Notes

Next-generation counseling: a model for non-invasive prenatal screening results disclosure and patient management

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OBJECTIVES
Non-invasive prenatal screening (NIPS) utilization has grown dramatically and is increasingly offered to the general population by non-genetics specialists. All major guidelines recommend patients with both negative and positive results be counseled regarding limitations of testing. As a genetic testing laboratory that provides a results delivery system, including telecounseling, we report how this service is utilized for patients undergoing NIPS.

METHODS
Upon results availability, providers are notified. If negative, a patient is contacted by automated email to access results through a secure portal where she may watch tailored informational videos, request "on-demand" genetic counseling, schedule a later consult, or decline all of the above. If a consultation is elected, a summary is sent to the ordering provider. If results are positive, either the ordering provider or our own genetic counselor contacts the patient directly.

RESULTS
Over a 29-month period, 27,827 NIPS results were issued through the system. Of these, 1,975 patients elected genetic counseling, 96.6% of whom received negative results. 65.2% (n=1,244) of patients with negative results and 72.1% (n=49) with positive results requested an on-demand consult. Average consultation time was 15 minutes (range: 3-54 minutes) for positive results and seven minutes (range: 1-40 minutes) for negative results. The average patient satisfaction rating for consultations was 4.9/5.0.

CONCLUSION
Combining web education, counseling, and automated notifications, we implemented a service that efficiently manages results disclosure. The majority of patients choosing to schedule a consultation had negative results, demonstrating a desire for post-test genetic counseling irrespective of test results. We describe an efficient and scalable means of manifesting medical guidelines on post-NIPS patient management, which is imperative to quality care as uptake grows among the general population.

Notes

New interventions to facilitate family communication about genetic risk and preparing the workforce.

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Genome sequencing is becoming routine and an integrated part of medical care. Despite the advancements in the science, preparation for the acculturation of the genomic information into family narratives has not taken place. Between 50-60% of families avoid discussing genetic risk information and even when they do, the quality of information sharing is very variable. The lack of communication about genetic conditions affecting the family can have severe consequences for the mental health of parents and their children, with some young people self-harming and even attempting suicide. Families report that more assistance is required from health professionals to assist parents in having these sensitive conversations with their children about a genetic condition that affects them or their family.

Using a co-design process and family systems theory we are involving families and health professionals in developing new interventions to facilitate better communication about genetic conditions affecting the family. The interventions draw on a strengths based model to develop parental confidence and family resilience in managing these potentially difficult and sensitive conversations and emotional responses.

One of the interventions developed, Multi-Family Discussion Groups (MFDG) was designed through a series of focus groups with parents, children, young people and genetic counsellors. Three genetic counsellors were trained in systemic family practice techniques before delivery of a MFDG intervention to families, co-facilitated by two systemic family practitioners and three genetic counsellors.

The findings from MFDG's design the showed there was strong support from families and health professionals for interventions to facilitate better family communication, and the genetic counsellors were able to deliver the interventions with the support of the practitioners. However a number of challenges also emerged that need to be taken into account going forward and these will be discussed as well as the future role of systemic family interventions in genetic counselling.

Notes

Facilitating personalized communication in genetic counselling for cancer

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Approximately 25% of individuals undergoing genetic counseling for cancer experience clinically relevant levels of distress, anxiety and/or depression. However, these general psychological outcomes do not provide detailed information on the specific psychosocial problems experienced by counselees, and are difficult to use in clinical practice of the genetic counselor. Therefore, a Patient Reported Outcome Measure (PROM) was developed to assess the specific psychosocial issues encountered by individuals undergoing genetic counseling for cancer.
In order to develop a PROM, which we named the Psychosocial Aspects of Hereditary Cancer (PAHC) questionnaire, we adopted the EORTC-Quality of Life Group guidelines. From the literature, we identified the following six overarching themes: coping with cancer risk, practical issues, family issues, children-related issues, living with cancer, and emotions. In total 26 items were developed and tested, and the screening properties of our new questionnaire were evaluated. In the final step, a Randomized Controlled Trial was used to evaluate the efficacy of this cancer genetics–specific questionnaire in facilitating communication about, awareness of, and management of psychosocial problems, as well as in lowering distress levels. Therefore, individuals referred to genetic counseling for cancer were randomly assigned to an intervention or a control group. All participants completed the PAHC before counseling. In the intervention group, the counselors received the results of this questionnaire before the counseling session.
The results showed that the frequency with which psychosocial problems were discussed with 246 participating counselees was significantly higher in the intervention group (n= 127) than in the control group (n =119), as was the counselors' awareness of psychosocial problems regarding hereditary predisposition, living with cancer, and general emotions. We found that the routine assessment of psychosocial problems by this questionnaire facilitated genetic counselors' recognition and discussion of their clients' psychosocial problems, without affecting the length of the counseling session. Furthermore it reduced clients' distress levels. We therefore recommend the use of a screening questionnaire like the PAHC in the genetic counseling sessions.

Notes

Challenges for genetic counseling of families with dilated cardiomyopathy and truncating titin mutations

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Dilated Cardiomyopathy (DCM) is a commonly-occurring heart muscle disease characterized by dilation and impaired contractility of the left or both ventricles, which is a major cause of heart failure, heart transplantation, stroke and death. Genetic factors have an important role in the pathogenesis of DCM but the molecular etiology of this disorder is incompletely understood. Truncating variants in the TTN gene (TTNtv) that encodes the giant sarcomeric protein titin are present in approximately 15-20% of cases and are proposed to be the most common genetic cause of DCM. However, TTNtv are also found in up to 3% of healthy individuals in the general population and it is currently unclear whether TTNtv are sufficient to cause disease or modify disease susceptibility. This unresolved conundrum raises enormous challenges for counselling individuals in whom a TTNtv is identified.
We performed whole genome sequencing and/or targeted re-sequencing of a cardiomyopathy gene panel in 174 probands with familial DCM. TTNtv were identified in 38 probands (22%) and detailed analysis of genotype-phenotype correlations was performed in 137 individuals in eleven families. Three families showed good co-segregation with the TTNtv present in all affected and none of the unaffected family members tested. In seven families, there was incomplete co-segregation with several unaffected TTNtv carriers. Overall, the median age of DCM onset in TTNtv carriers was 44 years (range 16-75 years) with DCM penetrance increasing from 44% at 40 years to 88% at 70 years of age. Males had a higher DCM penetrance (53% vs 34% at 40 years) and a lower median age of onset (39 vs 53 years) compared to female TTNtv carriers. Interestingly in one family there was poor segregation and incomplete penetrance with two genotype-negative affected individuals suggesting that TTNtv are unlikely to be the only cause of DCM.
Truncating TTN mutations are present in a significant proportion of familial DCM cases. Segregation analysis can be highly informative in determining the potential clinical significance of TTNtv and age and sex effects on disease penetrance needs to be taken into consideration. Care should be taken when implicating TTNtv as potentially disease-causing as clinically important second genetic and/or environmental factors can be involved.

Notes

Psychological impact and genetic counselling preferences of multi-gene cancer testing in a Spanish multi-centric cohort: a 12-month analysis

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Genetic testing for hereditary cancer has evolved from single-gene to multiplex panel testing including high and moderate penetrance cancer genes. The emotional reactions to the different testing approaches might differ. One hundred and eighty-four patients with clinical suspicion of hereditary cancer undergoing a 25-gene panel test completed psychological questionnaires after pre-test genetic counselling and at one week, three months and twelve months after results disclosure. Cancer panel testing was not associated with an increase in cancer worry or a detrimental psychological impact regardless of the genetic test result ($p=0.14$ and $p=0.874$ for CWS and R-IES) 12 months after disclosure. One year after results, carriers of a moderate penetrance variant were less likely to understand their medical options ($p=0.026$), and had higher distress (MICRA and R-IES), and uncertainty (MICRA) compared to carriers of high penetrance variants ($p=0.026$, $p=0.031$, and $p=0.025$, respectively). Baseline cancer worry was a relevant predictor of genetic testing psychological impact in the short and long term. The majority of patients reported the wish to know all genetic results and health-care professionals expressed the need for new genetic counselling models. Our results suggest that patients can emotionally cope with cancer panel testing. However, the distress and uncertainty identified in carriers of moderate penetrance variants warrants further research.

Notes

What counts as 'success' in genetic counselling? A systematic review of RTCs in assessing outcomes in genetic counselling?

Barbara Biesecker

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With the advancements in precision medicine and health care reform, it is critical that genetic counseling practice respond to emerging evidence to maximize client benefit. We conducted a systematic literature review to synthesize evidence on outcomes from randomized controlled trials (RCTs) of genetic counseling to inform clinical practice. Studies were selected for inclusion if they were: (a) RCTs published from 1990 to 2015, and (b) assessed a direct outcome of genetic counseling. A review of 1654 abstracts identified 58 publications of 54 unique RCTs that met inclusion criteria, the vast majority of which were conducted in the cancer genetic counseling setting. The most common client outcomes hypothesized to be affected by genetic counseling were psychological wellbeing, knowledge, perceived risk, and patient satisfaction. RCTs of genetic counseling demonstrate enhanced client outcomes in a many of the studies and pave the way to evidence-based practice. Results from this review will be used to deliberate what constitutes sufficient evidence to incorporate an intervention or delivery mode into clinical practice.

Notes

Measuring the economic value of genetic counselling

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The decision to allocate resources to a particular service excludes those resources from alternative possible uses within a healthcare system. The need to measure the relative value of alternative uses of the same budget is the basic premise that underpins the use of the concept of opportunity cost and the use of economic evaluation to inform resource allocation decisions. Methods of economic evaluation, in general, and cost-effectiveness analysis (CEA), in particular are now increasingly used as an evaluative framework to measure the economic value of new technologies or healthcare services. In this context, genetic counselling, and any modifications to existing models of service delivery, should ideally be able to show added value using robust methods of economic evaluation. As an example of a complex intervention conducting economic evaluations of genetic counselling may be problematic on two levels (i) how to identify and collect resource use data for the intervention, subsequent treatments and pathways of care for the proposed new intervention compared with current practice and (ii) how to identify and quantify the impact of the intervention and current practice on the relevant patient population, which may sometimes include family members. This presentation focuses on the second challenge drawing on the implications of a programme of work that provided empirical evidence to show genetic services may have broader objectives than maximising health status, which is the standard outcome valued in CEA of healthcare interventions. Specifically the presentation will explain the value judgements that underpin the use of CEA and the need to take account of opportunity cost if outcomes other than health status are introduced into economic evaluations.

Notes

Using data collected using patient-reported outcome measures for quality improvement in clinical genetics

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International advocacy of patient-centred care is driving attempts to evaluate and (re)design healthcare processes and outcomes from the patient's perspective. Patient-reported outcome measures (PROMs) have significant potential to contribute to these attempts.
The aim in this study was to explore the views of genetics clinicians in Wales about usefulness and feasibility of using PROMs data for quality improvement in clinical genetics.
PROMs data were collected using mailed self-completion questionnaires between February and July 2015. PROMs used were the 24-item Genetic Counselling Outcome Scale (GCOS-24) and the generic 5-item Euroqol (EQ-5D) before and after clinic attendance, both of which enable pre-post intervention comparison, and a post-clinic service audit tool. GCOS-24 is a well-validated clinical genetics-specific PROM, with demonstrated validity, reliability and sensitivity to change over time. EQ-5D is the generic PROM preferred by the UK National Institute for Health & Clinical Excellence. Monthly meetings with clinical staff were used to monitor progress and discuss the data collected. Qualitative interviews with eight participating clinicians explored their perceptions of benefits and challenges associated with use of PROMs.
Paired before-after PROMs data were collected from 89 patients (response rate=23% time1, 44% time2). Quantitative data analysis demonstrated statistically significant improvement in patients' GCOS-24 scores following clinic attendance ($p>.001$), and high post-clinic scores on the audit tool but no significant change in EQ-5D scores. Monthly meetings with clinicians increasingly focused on change scores on individual GCOS-24 items and strategies the clinical team could adopt to improve these change scores over time. Qualitative analysis of interview transcripts demonstrated that participating clinicians valued use of PROMs data to inform quality improvement initiatives using Plan-Do-Study-Act cycles, a quality improvement approach commonly used in the NHS. Participating clinicians considered the clinical genetics-specific GCOS-24 the most appropriate PROM. The main challenge of PROMs use identified was low patient response rates. Participating clinicians reported that GCOS-24 data provided insight into patients' needs, complementing clinical judgement; identified and quantified where patient needs were being met, evidencing the benefit of services provided; prompted consideration of areas of their practice requiring attention and encouraged professional development.

Notes

The ongoing search for a patient reported outcome measure (PROM) in genetic counseling: the psychometric properties of the Genetic Counseling Outcome Scale for a Dutch study sample

Jan Voorwinden[1], Mirjam Plantinga[2], Wim Krijnen[3], Margreet Ausems[4], Nine Knoers[4], Mary Velthuizen[4], Erwin Birnie[2], Anneke Lucassen[5] Irene van Langen[2] and Adelita Ranchor[1]

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An internationally validated and agreed upon patient reported outcome measure (PROM) where the service quality in genetic counseling is evaluated by counselees is urgently needed, but not yet available. A concept that covers many aspects of the service quality in genetic counseling is empowerment. Empowerment has been identified as a key patient outcome goal of genetic counseling and seems an overarching construct that represents many other patient reported outcome measures of services in clinical genetics. Empowerment in genetic services has been operationalized before in a 24-item questionnaire: the Genetic Counseling Outcome Scale (GCOS). The objective of this study was to validate this questionnaire for a large Dutch study sample of 2158 patients who were referred for genetic counseling (oncogenetics, cardiogenetics, reproduction, other) in two university medical centres. There were three assessment points: before the intake (T0), direct after the intake (T1) and one week later (T2). Results showed that we could detect a 7-factor structure that was found by the developers. Besides a total score for “empowerment” we suggest seven subscales: “hope and coping”, “family response”, “knowledge about the condition”, “knowledge about genetic services”, “uncertainty about the treatment”, “negative emotions” and “uncertainty about heredity”. Four items of the GCOS had to be dropped due to insufficient factor loadings and insufficient relatedness with all other items. Internal consistency and test-retest reliability of the total score and subscales were satisfactory. Convergent validity was confirmed by moderate positive and negative associations between the GCOS and other validated outcome measures, including PPC and STAI. Sensitivity to change was comparable to those of the other outcome measures. This study contributes further to the international validation process of the GCOS.

Notes

Bioethics and Genetic Counselling

Heidi Carmen Howard

Uppsala University, Sweden

The domain of Bioethics is a relatively new field. It has developed over the last decades as a multidisciplinary and comparatively applied field using different approaches including, among others, those from philosophy, theology, sociology, anthropology, policy studies, as well as the biosciences and medicine. There are clear parallels between the fields of Bioethics and Genetic Counselling; the work done helps to protect vulnerable persons and to support autonomy, all the while considering risks and benefits as well as justice. Starting from the perspective of the field of Bioethics, I will first provide an overview of the field and how I consider it to relate to Genetic Counselling. I will then discuss different cases, such as consent, direct-to-consumer genetic testing, and gene editing in an attempt to map out the perspectives from each field.

***Please bring your smartphone or tablet, as this presentation will involve audience input via Mentimeter*

Notes

The duty to inform: HD case legal going through the English courts

Victoria Chico

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In May 2017 in the case of ABC v St Georges Healthcare NHS Trust the Court of Appeal held that it is arguable that clinical genetics professionals owe a duty of care to patients' relatives to inform them of genetic risks. This presentation considers this judgment, the potential next legal steps, and the implications of these for practising genetics professionals.

Notes

BME women's decision-making for risk-reduction after BRCA testing

Mavis Machirori, Dr Christine Patch, Professor Alison Metcalfe

King's College London

Familial cancer syndromes present challenges to health perspectives of individuals and their families. In breast cancer, women concerned about their family history are sometimes offered genetic testing and subsequent treatment options based on their histories and genetic results. Discussions around genetics in Black and Minority Ethnic (BME) groups are rarely documented in literature, and information regarding interactions with genetic testing and subsequent decision-making is even rarer. Counselling sessions based only on medical information miss out the many reasons participants consider in making health decisions, information which can be used to encourage BME women to engage in cancer genetics services. 15 BME women with a mixed personal and family history of breast and ovarian cancer backgrounds were interviewed in a study exploring beliefs about familial breast cancer syndromes. 11 of these had undergone genetic testing.

Participants demonstrated mixed biomedical, social and individual cultural reasons for undertaking genetic testing and the role of the genetic results on their decision-making towards risk-reduction surgeries and treatment options. They evaluated their views regarding similar options for their family members which at times differed from the routes they themselves had taken. Narratives about suspicion of scientific utility of genetic knowledge, the perceived predictive value of mutations for future cancers or the origin of mutations and family disease patterns feature heavily in how participants evaluated genetic information and treatment decisions. In addition, social circumstances, individual health identities and cultural values were juxtaposed with medical information and personal disease histories to make judgements about how much or how little participants wanted to be involved with cancer genetics services. The diversity of results shows that women from BME groups are interested in engaging in genetic information but use multiple sources for evaluating the extent of involvement in genetic services and the place of genetic information and treatment options for themselves and their families. Concerns of familial breast cancer syndromes are contextualised to personal cultural values and the impact of genetic information on health decision-making is aligned with these personal values. Counselling for treatment options should explore and incorporate as many of these personal cultural values to ensure more individualised and inclusive discussions that address these multiple sources of concerns.

Notes

Reporting clinical whole genome sequencing results via telephone genetic counseling in a diverse healthy population

Molly A McGinniss1, Erica Ramos1, Erin Thorpe1, Lindsay Fosler1, Kimberly Schahl2, Karmen Trzupek2, Abigail Hata2, Tricia See2, Helen Bixenman3, Doug Morton3, David Wellis3

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Clinical whole genome sequencing (cWGS) for healthy individuals is rapidly gaining acceptance as cost decreases and the identification of disease-causing variants increases. The majority of early adopters of research or clinical cWGS have a narrow demographic profile - wealthy, highly educated, and Caucasian. This lack of diversity is a barrier to assessing issues that may arise when implementing cWGS in a broader population. The growing availability and uptake of cWGS also highlights the importance of assessing various genetic counseling methods to address the increasing demand for genetic counseling services. The San Diego Blood Bank (SDBB) and Illumina conducted an IRB-approved pilot project in which cWGS was offered to a diverse group of 70 SDBB blood donors whose demographics (race, income, age, etc.) were representative of San Diego County. Participants had cWGS and received interpreted results for ~ 1200 Mendelian disorders and a pharmacogenomics panel. There were an average of 2.17 pathogenic/likely pathogenic variants and 1150 variants of unknown significance (VUS) per participant. Nine participants (13%) had pathogenic or likely pathogenic (P/LP) variants expected to be clinically significant (heterozygous for a dominant condition or homozygous/compound heterozygous for a recessive condition). Two participants (2.9%) had a likely pathogenic variant in one of the 59 genes defined as actionable by the ACMG. One participant had a personal and family history consistent with his genetic results and the other had limited family history information. Certified genetic counselors conducted post-test telephone genetic counseling sessions, including family and medical history collection and psychosocial assessment. Preparation, counseling, and documentation times were tracked. Average counseling time was 66 minutes with 39% of counseling sessions taking 45-60 minutes. Average time spent preparing for patients and completing a consultation summary were 18 and 35 minutes respectively. These metrics are comparable with the 2016 NSGC PSS.
This pilot study demonstrates the feasibility of providing consistent, efficient genetic counseling services to a diverse healthy cohort with cWGS results. The ability to integrate telephone genetic counseling services into cWGS supports increased diversity and quality of care as it improves access to certified genetic counselors.

Notes

The priorities for research in genomic and priorities

Clara Gaff

University of Melbourne, Australia

Genetic counselling takes place in an ever-changing landscape shaped by scientific advancements and health system constraints. The most recent technology to impact on genetic counselling has been genomics, accompanied by claims that it will transform health care. Currently, genomic technologies are used clinically to better diagnose single gene disorders; the genetic counselling issues are largely familiar but – like the technology itself – on a larger scale.

This does not mean that the body of scholarship and research available to us is sufficient to navigate the next era of “genomic counselling”. In this presentation, I project forward into the near future when understanding of our genome allows us to more accurately predict disease risk for common conditions. I explore the evidence that will be required to assist genetic counsellors and society to meet the challenges of this new era and propose priority areas for research to provide this evidence.

Notes

Genetics in popular culture

Jon Roberts

Wellcome Genome Campus, UK

Media representations of genetics are regularly criticised by scientists as exaggeration or fear-mongering. For many, the way that science is presented in popular culture is inaccurate, negative and a barrier to effective science communication. However for the majority of people it is through media such as film, TV shows and books that they become familiar with many scientific terms and ideas.

There has been comparatively little research to date that explores the way people draw on and utilise pop culture when discussing genetics. However new work from scholars working in this area has found evidence that audiences deal with texts actively, selectively and critically. People don't believe everything they watch or read. Instead the relationship between the media that people consume and their attitude towards genetics is complex.

In this talk I explore some of the ways that genetics is presented in pop culture, outlining some of the main narratives, frames, metaphors and tropes utilised in fiction and the media. I will then describe some research, including work from my PhD, that looks at the ways in which people are able to draw on these texts as resources to articulate their views and opinions about genetics.

I will conclude by outlining the relevance this has for genetic counselling practice as genetic moves mainstream and we are called on to make real the 'genomic dream.'

Notes

Personal Genomic Screening: how best to facilitate preparedness of future clients

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Personal genome screening (PGS) is increasingly being offered as a screen for future health and wellness, and to identify carrier status pertinent to future generations. The aim of this study was therefore to explore the experience of individuals undertaking PGS to identify how best to prepare future clients. Individuals who undertook PGS through the 2014 Sydney "Understand Your Genome (UYG)" event and 2015 offer by biotechnology company Life Letters (LL) were invited to participate by their clinical geneticist (UYG), or email (LL). Semi-structured telephone interviews were conducted, audio-recorded, transcribed and de-identified. Transcripts are currently being analysed by two coders using thematic analysis with an inductive approach. Seventeen individuals were interviewed: nine genetic health professionals and eight non-genetic health professionals. Individual PGS results included: an autosomal dominant condition neurofibromatosis type 1 not previously clinically identified; carrier status for recessive condition(s); a number of variants identified as likely pathogenic but many of uncertain significance; and pharmacogenetically relevant mutations. Analysis to date has identified some common themes between the groups. The majority of participants noted the importance of pre-testing information and consent with positive experiences with clinical geneticists/genetic counsellors. Some barriers to uptake were identified, including skepticism of colleagues, family members and privacy concerns. The rationale for testing by many genetic health professionals was cited as professional interest and/or curiosity, without anticipating personal or family impact. On reflection, despite this initial objective motivation, the impact of the test result had unanticipated personal impact and changed over time and several later recognized their relevance, as health problems developed or family history was interrogated more closely. Non-genetics health professionals were mostly motivated by curiosity with two participants influenced by a prior undiagnosed medical condition. For the majority of participants, disclosure of results to extended family members has been limited. Most participants felt that expectations; residual risk; changes in interpretation with developing phenotypes; and personal and family impact and communication needed greater emphasis at the pre-test session. Non-genetics professionals' highlighted complexity of information provided. These results will inform development of more accessible resources, and counselling approaches to address expectations, dissemination of results, and preparedness for unexpected findings.

Notes

Poster Presentations

Genetic counselling and lifestyle changes in acute intermittent porphyria

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The porphyrias are a group of rare and mainly inherited diseases caused by reduced activity of different enzymes in the heme synthetic pathway. Acute intermittent porphyra (AIP) is characterized by attacks of abdominal pain, headache, muscle aches, muscle weakness, paresis, hypertension, tachycardia and respiratory paralysis. A range of medications can trigger symptoms, in addition to hormones, alcohol, physical and psychological stress, and dieting. Inheritance is autosomal dominant, with reduced penetrance (10 %). Disease severity varies from minor symptoms of abdominal pain to serious and frequent attacks with potentially lethal complications. In order to prevent activation of the disease, the Norwegian Porphyria Centre (NAPOS) routinely offers genetic counselling and predictive testing for AIP to healthy at risk relatives.

The aim of this cross-sectional study was to investigate what motives healthy at risk persons have for undergoing genetic testing and whether they were satisfied with the genetic counselling they received. In addition, we wanted to investigate in both predictively tested and manifest patients whether receiving a diagnosis led to lifestyle changes.

The study was conducted among persons with genetically predisposed (n = 28) and manifest (n = 106) AIP. Self-administered questionnaires of motives for genetic testing, satisfaction with genetic counselling (SCS) and lifestyle changes were used.

Most participants reported that their motives for genetic testing were the possibility to prevent symptomatic disease (82 %) and the consideration of risk for children (76 %). Persons who had received genetic counselling were highly satisfied with the relevance of the content. After getting the diagnosis the participants became more conscious of checking their medications (92 %). Participants also changed their lifestyle in regards to eating habits (56 %), alcohol (54 %) and tobacco consumption (54 %).

It seems the genetic counselling session contains elements that individuals at risk of AIP find informative. Our findings showed that the possibility to prevent symptomatic disease is an important motive to get tested and that receiving the diagnosis motivates both manifest and predictively tested patients to make healthier lifestyle choices.

Addressing religious beliefs and spirituality in genetic counselling sessions

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Many factors impact on the lived experience of patients with a personal and/or family history of a genetic disorder. One important factor is religious beliefs and spirituality (henceforth R/S) because an estimated 68% of the world's inhabitants attribute importance to R/S in their everyday lives. We will discuss the main studies that have explored the impact of R/S on patients' decisions about whether or not to undertake genetic testing, their uptake of screening and reproductive options and on their coping processes. This will help to raise Genetic Counsellors' awareness of the importance of R/S values for many patients. We will also review approaches to assessing R/S to provide Genetic Counsellors with tools to use to become comfortable with initiating discussions about R/S.

The first assessment of psychological and educational aspects of the genetic counseling's efficacy in Russian Federation

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In spite of that there are no genetic counselors in Russia medical geneticists provide the genetic counseling for the patients. Until recently the efficiency of genetic counseling especially its psychological and educational aspects were poorly investigated. The present research is dedicated to these problems. The qualitative study is aimed to estimate the genetic counseling efficiency by a number of parameters, including satisfaction, reduction of stress, the guilt feeling relieving after the birth of affected child. The study is conducted in the outpatient department of the Research Centre for Medical Genetics from 2007. To the present we have 226 completed cases. Our respondents were from 17 to 67 years old and had an affected child/children or affected relatives. All families were interviewed before and after genetic consulting. An attempt was made to find whether the respondents understand the importance of genetic counseling and the goals of the latter; the initial level of respondents' knowledge and some psycho-emotional characteristics of the respondents. The interviews were analyzed using Statistica 10.0 software. Most respondents (n=213, 94%) indicated that genetic counseling was important or very important to them. We found that understanding of the risk of birth of a affected child after genetic counseling increased significantly (Wilcoxon test, $p < 0.05$) . Also significantly increased knowledge of patients about the possibility of prenatal diagnosis (χ^2 - distribution, $p < 0.05$). We identified a high level of anxiety and guilt feelings among respondents (60% respondents with affected children experienced guilt feelings and 40% were very concerned about the possibility of the disease recurrence in their family). The level of anxiety is significantly reduced after counseling ($p<0.05$). We are pleased to record a high level of satisfaction with genetic counseling among respondents: 197 (87%) of respondents considered it useful or very useful, 212 (93%) indicated the investment of time and money to be entirely or substantially justified, 181 (80%) reported satisfaction with advice. This investigation is proposed that psychological and educational skills of the medical geneticist involved in genetic counseling is important for efficiency of the genetic counseling. This work was supported by the Russian Foundation for Humanities, project 15-03-00822.

Australian life insurers use of genetic test results in underwriting decisions

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Australian Life Insurers use of genetic test results in underwriting decisions
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In Australia, the USA and many Asian countries, there are disclosure requirements of genetic test results in applications for new policies or when renewing previously guaranteed polices for life insurance including cover for critical care and income protection and death. Concerns have been expressed as to how insurance companies are managing the underwriting for polices with such disclosures. Since 1999, the Financial Services Council (FSC) has requested its Australian life insurance member companies provide data on applications where a genetic test is disclosed. The FSC provided data collected 2010-2013 to enable repetition of an independent analysis undertaken of applications 1999-2003 (Otlowski et al 2007). Data included de-identified insurer; age; gender; genetic condition; reason for testing and result; underwriting decision-maker; and insurance cover. Data was classified as to test result alone or in addition to other factors relevant to risk, underwriting decision. Where necessary, the FSC facilitated clarification by insurers. 340/547 applications were for adult-onset conditions: hereditary haemochromatosis (HH-200); cancer (51); thrombophilia (31); cardiovascular (17), neurodegenerative (13), neuromuscular (9); and other (19). The genetic test result solely influenced the underwriting decision in 170/340 applications: 24 positive, 139 negative, 2 uninformative, 3 pending and 2 unknown. Policies were provided at standard rate for all negative test results with evidence of reassessment of previous non-standard decisions and 20/24 positive results with recognition of risk reduction strategies. Non-standard polices were provided for positive BRCA2 (2) and Lynch syndrome results; for the two BRCA1/2 uninformative results, breast cancer exclusion and 50% loading were applied respectively; and for results pending (cancer-2, Huntington disease-1) applications were denied. Limitations in the data influence interpretation and generalisability of the findings including the context of the testing setting (research/clinical).

The evolving landscape of genetic counselling practice in the genomci era

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The evolving landscape of genetic counselling in the genomic era
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Background: Facilitating informed decision-making regarding genetic testing is a core component of genetic counselling practice. Internationally, and more recently in Australia, genetic testing is moving from single genes to genomic testing (including gene panels, wholeexome or whole-genome sequencing). The rationale is improved diagnostic yield and cost effectiveness.
Aim: To explore genetic counselling practice in Australia and the UK in the context of genomic testing.
Method: Recruitment was purposive sampling via email invitations of 14 genetics practitioners residing in Australia or the UK, who were known to the authors to have experience with the offer and delivery of panel and/or genomic results; a request to snowball to colleagues was included. Semi-structured telephone interviews explored their views, experiences and practice. Interviews were transcribed verbatim, de-identified, coded with concordance from two coders and analysed using an inductive thematic approach .
Results: 14/17 practitioners have been interviewed to date: Australia (9), UK (5). Three themes have emerged. (I) Role delineation including increased autonomy and the influence of increasing complexity; multi-disciplinary team involvement and responsibility; and responsibilities for current and future variant interpretation and result delivery. (2) The evolving spectrum of practice including that practice was building on core genetic counselling skills with emphasis that these remained the same regardless of the type of test used; blurred boundaries between research and clinical services; consent processes and streamlining based on experience; and return of results strategies. (3) Policy and Governance needs including access to testing; achieving consistent variant interpretation, reporting and responsibility for review; managing incidental findings; and issues regarding professional registration for Australian genetic counsellors.
Significance: These exploratory data highlight that genetic counselling practice in the genomic era is evolving but remaining patient-centred, with core skills underpinning practitioners' capacity to adapt.

Predictive Testing for BRCA1/2 and Lynch Syndrome: A service evaluation

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In the UK there has been a general move towards reducing the number of patient contacts prior to presymptomatic predictive testing for inherited cancer. At MCGM we conducted a prospective service evaluation for predictive testing for BRCA1/2 and Lynch syndrome. In accordance with departmental guidelines, the blood sample is usually taken at the 2nd appointment and the result given in person 4 weeks later. Increasingly patients are offered the option of receiving the results by telephone.
We devised 2 separate questionnaires to evaluate the acceptability of the current Manchester protocol to both patients and Genetic Counsellors (GCs). Questionnaires were completed prospectively over a 3 month period by 59 patient and GC pairs at the time of testing.
98% of patients reported that they found it helpful to discuss genetic testing with a GC and 97% reported that speaking to a GC had improved their understanding of the condition in the family and the options available. Only 10% of patients were tested at their first appointment (mainly males from BRCA1or 2 families). Of those patients not tested at the first appointment, the GCs assessed 49% of patients as ready to be tested at their initial contact. Interestingly, only 13% of patients not offered testing at the first contact suggested this as a possible improvement to the service they received. 71% of GCs felt the patient had benefitted from a second appointment. 45% of patients were offered telephone results but only 65% accepted the offer with 35% preferring to return to clinic for a results appointment. 100% of patients reported satisfaction with arrangements made to receive their results, regardless of whether they were offered results by telephone or not.
Data from these questionnaires suggest a high level of patient satisfaction with the current predictive testing protocol. The current system of 2 face to face sessions with flexibility for GCs to test earlier in specific cases seems to be acceptable to the majority of patients. From a health economics perspective, it would be interesting to compare patient and GC responses with those of a cohort of patients routinely offered testing at the first appointment and given results by telephone.

Impact of Privately Funded Genetic Testing in the Austin Familial Cancer Clinic of Genetics in the North East

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Over time, the cost of mutation detection testing has decreased making privately funded testing more accessible to patients. Some of the patients choosing to have privately funded testing are referred for genetic assessment as it may impact their cancer treatment decisions, although the likelihood of identifying a pathogenic mutation is low. Other patients wish to arrange privately funded testing to refine risk assessment advice for themselves or family members.
The number of patients proceeding with privately funded testing at the Austin Familial Cancer Clinic (FCC) has increased. The tests available to patients has also changed over time with single gene or paired gene (eg. BRCA1 and BRCA2) testing being replaced with routine testing of panels of genes. An audit of privately funded mutation detection testing arranged through the Austin FCC was performed and the results will be presented.
We examined which tests are privately funded by Austin FCC patients and the frequency of these tests over time to assess their impact on the Clinic workload. We also compared the mutation detection frequency of privately funded tests and tests performed when clinical criteria for State funded testing were met.
Uptake of privately funded testing is likely to continue to increase as costs further reduce. By examining these trends we aim to gain insight into the impact this type of testing has on service delivery, risk estimation, and overall management. These results will allow an improved understanding of service provision and facilitate mechanisms through which these patients may potentially be managed by treating specialists outside the Austin FCC. We will also assess the outcome of privately funded testing to better appreciate its utility.

Genetics first… and then genetic counselling. The experience of returning whole genome sequencing results to healthy volunteers in Singapore.

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A major barrier to identifying variants associated with disease traits is the lack of ethnically diverse genomic data. The SingHealth Duke-NUS Institute of Precision Medicine (PRISM) was established to promote precision health by focusing on medical conditions relevant to South East Asian populations, currently an under-represented cohort. Specifically, the aims of this initiative are to: 1) determine the range of South East Asian normality by detailed phenotyping and genotyping of 5,000 healthy Singaporeans; 2) establish a clinical framework to identify and return medically significant variants; 3) define the frequency of disease-causing variants within the Singaporean population. So far, data analysis of 124 genes from whole genome sequencing of 455 healthy volunteers has identified nine variants associated with autosomal dominant conditions, and that 8 in 100 individuals are carriers of recessive conditions. Variants are annotated according to ACMG guidelines, reviewed as a multidisciplinary team and returned to consenting participants. To our knowledge, this is the first evolving reference of Asian genotypic and phenotypic data coupled with a clinical workflow to return results. The development of this framework has enabled the discovery of novel pathogenic variants and carrier frequencies of genetic conditions in our local population, which are underestimated in existing data sources. However, determining which genetic variants to return, classifying variant pathogenicity in absence of patient phenotype or apparent family history, and retuning pathogenic results to volunteers considered "healthy" are experiences which diverge from the traditional norms of genetic counselling practice when genetic testing takes place. These professional challenges and experiences will be discussed in context of developing a framework to integrate genomics into South East Asian healthcare.

An Evaluation of Preimplantation Genetic Diagnosis Outcomes in Johannesburg, South Africa.

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Preimplantation genetic diagnosis (PGD) is a process that involves testing embryos that were created using in vitro fertilisation (IVF) or intracytoplasmic sperm injection (ICSI) for genetic conditions before being transferred to a woman's uterus for implantation. In South Africa, PGD is still a relatively new service, with 2006 marking the first PGD case managed through the Division of Human Genetics at the National Health Laboratory Service (NHLS) and the University of the Witwatersrand (Wits) Johannesburg, South Africa. When couples are seen for PGD-related counselling, the main concerns that are usually raised centre around the cost, length, and complexity of the PGD process, as well as the relatively low IVF success rates. To make an informed decision about whether to undergo PGD, couples need to understand all the limitations involved and the possible outcomes that they may experience. Current PGD-related genetic counselling can only provide generalisations of PGD outcomes and expenses. Though several PGD cases have been managed in the Division since 2006, no audit of this service has ever taken place. Therefore, the aim of this study was to conduct a retrospective case review to determine the number and nature of PGD cases that have been managed through the Division, and to assess the outcomes at various stages of the PGD process, as well as to conduct a cost analysis of the PGD process. Since 2006, 33 patients have been managed through the Division for PGD-related genetic counselling of which 22 met the inclusion criteria. 42 IVF/ICSI cycles were completed among these couples. After PGD and PGS, 31.1% of biopsied embryos were suitable for transfer. 34% of transferred embryos successfully implanted, and 78.6% of these resulted in a liveborn baby. We report a clinical pregnancy rate of 29.3% per embryo transfer. Overall, 45.5% of couples included in this sample had a successful cycle resulting in a liveborn baby. On average one cycle of PGD costs R117,513.20. This study shows that our PGD success rates are comparable to those achieved globally as reported in the literature. The findings from this study will enable genetic counsellors in South Africa to offer couples evidence-based information regarding PGD outcomes, success rates and costs involved.

“This is the first I’ve heard about it”: An assessment of awareness and perceived genetics training needs of the healthcare workforce in the East of England

Gemma Chandratillake, Mohammad Yusuf Ali Olath, Joann Leeding

East of England Genomic Medicine Centre

Mainstreaming of genomics in the U.K. will require a genetically literate healthcare workforce. To assess current awareness and inform education and training strategy, the East of England Genomic Medicine Centre conducted a Training Needs Assessment survey. Responses were received from >1000 NHS staff, from 40 NHS England Trusts (hospitals) and 25 Clinical Commissioning Groups (primary care). While the response rate is unknown, 5-7% of clinical staff in our lead organisation participated.

Through quantitative and qualitative analysis, general themes and specific educational needs emerged. Doctors and healthcare scientists generally received genetics training during their university education, but few received professional training. The majority of nurses & midwives, pharmacists, and allied health professionals received no genetics training at any time. Awareness of the 100,000 Genome Project stood at 50%, with the appetite for training being high.

The responses to this survey have guided immediate and longer term awareness and educational strategy in the region. Efforts include a re-work of our website to provide a one-stop-shop regarding DNA and patient care, and establishment of a region-wide email bulletin highlighting training opportunities. Accordingly, we have seen a substantial increase in uptake of the modules of our Genomic Medicine Programme by a wide range of NHS staff. The survey may have been the first, but it's certainly not the last our workforce will hear about genomics!

Everyone who goes to genetic counseling has a 'black spot'. Attitudes regarding genetic counseling, among Arab communities in Northern Israel.

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Aims and Backgrounds
Prenatal genetic counseling is offered before or during pregnancy in order to identify couples at risk for fetal abnormalities and genetic disorders. Although the service is offered to all Israeli citizens, findings show underutilization of genetic counseling services by pregnant Israeli Arab couples.
We explored attitudes and beliefs regarding genetic counseling and genetic testing among Arab women in the Galilee, in order to improve the service and develop tailored genetic counseling processes for different Arab communities.
Methods
The study is based on a purposeful sampling of 18 women who attended mother and infant clinics either for routine checkup during pregnancy or for their new-born children. These women were drawn equally from the 3 principal subgroups of the Arab population in Israel: Muslim, Druze, Christian. In depth semi-structured interviews were conducted that explored knowledge, perceptions of the counseling service and risks related to prenatal diagnosis
In addition, we held two focus groups: one with nine "Mother and Infant Clinic" nurses, and the second with seven genetic counselors. Content analysis based on the transcribed interviews and focus group was performed.

Results and conclusions
The following themes emerged from the interviews:
1. Most participants exhibited a lack of knowledge regarding the genetic counseling process, and prenatal genetic testing.
2. Referral to genetic counseling was perceived as unnecessary or "threatening" and as promoting termination of pregnancy.
3. Perception of risk regarding screening tests and prenatal diagnosis was often inaccurate and perceived as much higher than reality.
4. "Passive coping" with risk information - Acceptance of risk for negative fate for the fetus with significant doubt based on previous "lay stories" of an actually positive pregnancy outcomes when negative predictions were given to couples.
5. Key themes derived from the focus groups were consistent with themes emerging from the interviews, although there were some discrepancies such as considering effect of language and family role in the decision making process.
We conclude that our target populations are concerned about the complex process of genetic counseling in part due to misinterpretation of the messages that we aim to put across. Genetic counseling, tailored to the culture and beliefs of each Arab ethnic group is needed to improve the awareness and the genetic counseling outcome among couples and to help them with informed decision taking.

An audit of a patient information leaflet - options for prenatal diagnostic genetic testing.

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At Monash Health there has been implementation of a new prenatal diagnostic testing protocol for patients with an increased risk result for Trisomy's 13,18 and 21 on screening tests. This protocol differs from that of other service providers in Australia. It includes a publicly funded rapid test (FISH) to resolve the risk result from screening, but no further publicly funded testing. This is similar to other publicly funded healthcare services such as those provided in the UK. The patient is also given the option to self-fund a banded karyotype or chromosome microarray if they would like further information.

It is important that patients undergoing an invasive procedure in pregnancy understand the scope and limitations of testing. These results impact on life changing decisions and quality of life issues. Research indicates that patients typically only remember a small percentage of information given to them verbally. Written supplements can increase the retention of information up to 50%.

A leaflet was developed to support genetic counselling discussions. It outlines options available for testing, test scope and limitations, and funding available. It was given to patients after discussions either in the genetics department, or at the time of the procedure in the Fetal Diagnostic Unit, depending on the patient's care pathway.

The aim of the audit is to formally evaluate the quality of the patient information leaflet one year after it was implemented. This process includes formal assessment tools for readability/presentation as well as a tool to identify actions to be taken after the assessment. The evaluation includes a survey of patient opinions on the quality/usefulness of the leaflet.

Patients will complete a questionnaire asking for their opinion on the leaflet prior to chorionic villus sampling/amniocentesis. The leaflet will also be assessed using a readability tool and the Ensuring Quality Information for Patients (EQIP) tool. We will present the results of this audit.

It is important to include patient opinions in the development of information materials designed for them. Patient participation has been described as a key aspect of quality healthcare information materials, and is important to understand what patients actually want/need. This process of involvement should help reduce social inequities and empower patients.

An adaptable practitioner and patient visual aid for prenatal aneuploidy screening and testing options

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The introduction of prenatal chromosomal microarray and Non-invasive prenatal testing (NIPT) as options for patients at increased risk of fetal aneuploidy (on first or second trimester maternal screening) has significantly increased the volume and complexity of prenatal consultations. Increased risk prenatal counselling is particularly challenging, because it is emotionally driven and time restricted. In response to the added volume and complexity and to ensure consistency of information offered across multiple genetic counsellors, we designed a visual aid with dual purpose. Its primary purpose is to assist the genetic counsellors to standardise the application of new technologies and options offered during increased risk prenatal counselling. A secondary purpose is to provide patients with a visual aid to assist their understanding during consultation and enhance informed decision-making. We present this unique, adaptable visual aid, designed to assist both practitioner and patient. The aid is easily adaptable as new technologies become available, for different prenatal scenarios, and for use by different practitioners (e.g. midwives, obstetricians, family practitioners). We have subsequently adapted the "increased risk aid" and developed a "routine prenatal aneuploidy screening aid" and an "advanced maternal age aid". Genetic counselors at a major Australian maternity hospital have been using the three aids for approximately two years, and we have recently implemented the two latter visual aids for use by midwives at the hospital. We have surveyed the midwives using the aid and present findings from the surveys. Feedback showed that midwives valued the additional resource when discussing aneuploidy screening and testing with women, however a woman's first visit for antenatal care in the public hospital was often too late to discuss all available options. We are currently providing the counselling aids to primary care providers who have the opportunity to discuss options with women earlier in pregnancy.

Understanding the information needs of parents of children with rare copy number variant disorders

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On receiving news that their child has been diagnosed with a rare chromosomal disorder the first question parents often ask is, "What happens next?". Meaningful answers can be elusive. Personalising information about genomic copy number variants (CNVs) may be compromised by substantial clinical heterogeneity and a weak evidence base, but understanding parents' worries about their child's diagnosis ranks alongside the need for a comprehensive evidence base to guide health professionals. To better understand parents' concerns about their child's diagnosis we developed a 47-point questionnaire exploring their experiences and views concerning, (i) satisfaction with communication and explanation of genetic test results; (ii) beliefs about physical, neurodevelopmental and psychiatric disorders linked with the diagnosis; (iii) sources of health information; and (iv) the amount and utility of information parents receive. The questionnaire was launched through Bristol Online Surveys and was publicised through rare disorders support groups, primarily by Unique. We report observations from 199 UK participants recruited since December 2014.
Satisfaction with communication of genetic test results varied significantly depending on how they were delivered. Parents were more satisfied if results were communicated by genetic specialists compared with paediatricians or other health professionals. Receiving the diagnosis at clinic appointments was associated with greater satisfaction compared with clinical letters or telephone calls, but 102/199 (51%) said they did not receive results in person. Similarly, satisfaction with explanations of results was significantly greater when given by geneticists. Substantially greater rates of satisfaction were reported if information and support were offered when communicating the diagnosis. However, 72/199 (36%) of participants reportedly received no additional information at diagnosis and 148/199 (74%) reported no support was given. Participants believed that multiple mental health and developmental disorders are associated with their child's CNV. Information about neuropsychiatric disorders was significantly more likely to originate from sources other than clinicians when compared with developmental and physical disorders, where clinical specialists were the primary source. Information from support groups was significantly more helpful compared to internet websites and clinical specialist sources. Internet-derived information was rated significantly more helpful relative to geneticists and paediatricians. Our findings uncover important messages about current provision of services, support and information for parents of children with developmental and neuropsychiatric disorders attributed to genomic variation. Developing best practice in informing and supporting parents and delivering effective interventions to improve mental health outcomes should reflect parity of esteem with medical healthcare for this community.

Genomics Education strategy at the West Midlands Genomic Medicine centre

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The West Midlands is the largest Genomic Medicine Centre (WM GMC) comprising of a consortium of 18 Local Delivery Partner (LDP) Trusts. We have taken a different approach to most GMCs by focussing on transformation, including recruitment across the entire region and outside of clinical genetics.

A suite of new and innovative educational programmes has been developed within the region to underpin the delivery of the project, support nationally available related educational initiatives and to ensure the current and future workforce is equipped to understand how genomics and personalized medicine might impact on their role.
These include:

- A Genomics Access Course
- 100,000 Genome Consent and Ethics Training leading to development of a National Consent and Recruitment Train the Trainer Day, which used a blended learning approach to combine online (Genomics Education Programme) training and face to face training
- A 'Personalising medicine and mainstreaming genomics' masterclass

A varied genomics education programme has been established to support delivery of the 100,000 Genomes Project across the West Midlands, not only to equip staff to work within the project but also to embed basic genomics knowledge and skills within the NHS throughout the region.

Systematic Development of a Genetic Counseling Coursework using Curriculum Mapping

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With the increasing demand for genetic counselors, there is a need for more training programs of excellence. The rapidly advancing field of genetics challenges educators to create curricula to address this evolving field and continually improve the experience for the learners. The purpose of this study was to explore how curriculum mapping could be used when building a Master's degree in Genetic Counseling.
A curriculum committee convened with 12 genetic counselors divided into two work groups (first and second year). Two committee members took the Accreditation Council for Genetic Counseling's (ACGC) Standards and Practice-Based Competencies (PBC) and mapped them on a curriculum worksheet by key topic area, goals, and objectives. The two working groups used this curriculum map to assign potential courses to each objective. It was also noted if the objectives would be met through clinical rotations and/or research projects. The worksheet could then be sorted by these categories. Each working group then reviewed the curriculum map and outlined each course by the learning objectives. The full committee came together and each working group presented their proposal for their assigned year. Committee chair and leaders of the working groups met to create a congruent curriculum with course sequencing. Course descriptions were drafted including planned educational strategies.
The results of this process created a two-year curriculum with 170 learning objectives and 18 courses with 60 credit hours over 5 semesters. The curriculum addresses all the Standards and PBC put forth by the ACGC and integrates the objectives across coursework, clinical rotations, and research thesis. A variety of educational strategies are incorporated including lecture, discussion, reflection, small groups, problem-based learning, team-based learning, role play, and simulated patients.
This exercise has demonstrated that curriculum mapping based in the ACGC Standards and Practice-Based competencies can produce a robust two-year curriculum for master of genetic counseling. Advantages of this process include ease of documentation of learning for accreditation purposes and a process to track and add in new learning objectives. This model could be used to facilitate program development universally.

Raising Awareness for Rare Diseases – A Newly Recognized Aspect of the Genetic Counselor Role

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There are 7000 different rare diseases known today, most are genetic and affect children. People with rare diseases make up to 6-8% of the population, therefore it is estimated that in Israel alone there are half a million children and adults affected with rare diseases.

Along with the role of providing accurate information regarding the implications of a newly diagnosed rare genetic disease, there is also a strong demand for information accessibility in various aspects related to the care of individuals with rare diseases. Indeed, families and health care professionals often have very few clues in regards to the care needed for rare diseases, information which unfortunately is not always available and only partially known.

Here I will present a unique way of educating the public about complex medical information. Since the beginning of 2012 I am writing a personal blog called "Excerpts from the Diary of a Genetic Counselor", the only blog of its kind, written in Hebrew (for the moment), which contains short stories inspired by real families with the aim to increase awareness regarding various issues related to genetic counseling and rare diseases in Israel, along with helping rare families to have their voices heard.

As of today, over 60 different stories have been posted. Social media, which has revolutionized the life for rare disease families permitting them to create effective networks in terms of knowledge and research, made it possible for me to pass on the stories more widely and effectively, thus reaching a greater readership.

Each story contains medical information, including a short section describing the disease and its inheritance pattern, yet in a unique way, which is very easy to read. For example, one story is about Alice, a little girl with MPS IV (Morquio disease), who chooses to cope with her disease by pretending she is Alice in Wonderland.

With the main goal to raise awareness about genetic counseling and rare diseases, reaching as many people as possible including the rare disease community, health care professionals, the general public, and even policy makers, this blog adds one more stone to the "building of life" that will hopefully then serve as a better supportive world for those affected with rare diseases.

Beyond family history: communicating genetic risk in frontotemporal dementia

Elle Elan, -

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Frontotemporal dementia (FTD) refers to a heterogeneous group of neurodegenerative conditions involving progressive degeneration of the frontal and anterior temporal lobes of the brain. FTD is the second most common cause of early-onset dementia and is characterised by a spectrum of clinical symptoms, including changes in cognition, personality, behaviour, language deficits, and impaired social functioning. Genetic counselling is an essential component of both clinical and research genetics due to the complex aetiology of FTD and its co-occurrence with other diseases, including motor neuron disease (MND), which occurs in 15% of FTD patients. Ambiguity surrounding the genetic basis of FTD presents a unique genetic counselling challenge. Approximately 40% of individuals with FTD have a family history of dementia, but only 10% have a clear autosomal dominant pattern of inheritance. Furthermore, pathogenic mutations are found in 15-30% of all FTD cases, including those with seemingly sporadic FTD. This ambiguity is compounded by the condition's clinical and genetic heterogeneity, variable expressivity, phenocopies, unclear penetrance, and variable age of onset. Therefore, genetic risk assessment cannot be solely based on family history. In addition to the challenges involved in genetic risk assessment in FTD, communicating this risk to patients and their families is inherently difficult. Phenotypic heterogeneity means there is no 'one size fits all' model of genetic counselling in this cohort. Features such as the patient's lack of insight, as well as their inability to communicate due to language deficits, raise unique counselling considerations, both practically and ethically. Presented are two cases of FTD that highlight the challenges commonly faced in genetic counselling for FTD, as well as future research directions proposed to address these.

The current Australasian genetic counsellor workplace

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National and international reports have highlighted the importance of the genetic counselling workplace, and the limited up-to-date information available. This study therefore surveyed employed Australasian genetic counsellors to identify the current workplace demands and future considerations. Invitations to participate in an anonymous online survey (RedCAP) were disseminated via genetic counsellor listservs and promotion at relevant meetings. Inclusion criteria included currently working in a genetic counselling role for greater than 1FTE. The survey was a modification of the questionnaire developed by James et al, with additional questions relevant to current and future practice. All 112 respondents (RR estimate 30% based on ASGC data) reported they are employed as a genetic counsellor: either as a sole practitioner, team member, manager, in research or other setting. Respondents work in all states, territories and New Zealand in traditional public genetics services, subspecialties, private practice and other settings, with 44% employed for >5 years. 55% work full-time. 31% have HGSA board certification with 57% in training; and 33% work almost exclusively in cancer genetics. Respondents reported increases in client volume (85%) and changes in workload: with most respondents spending greater than 50% of their time on direct/indirect client care. Overall, 60% of participants were involved in variant interpretation and the majority use genome databases. Free text comments included 'more autonomy/responsibility'; 'manage increased complexity in information'; and 'implement new technologies'. Overall, a third had considered changing to a non-clinical role. Board certification was valued, and the majority of respondents felt certification was recognised in terms of career pathway, but less so in terms of salary/awards. Most participants undertaking Board certification received supervision but the amount received per month was variable. Most genetic counsellors were very or moderately happy with the level of supervision received. Analysis is continuing. These findings may inform strategies to improve inequalities and insufficiencies across genetics services, development of the profession, future workforce planning, and state and federal health policies related to genetic counselling service provision.

Lessons from genetic counseling of a pro-band with MODY2 due to a novel mutation (S441W) in Glucose Kinase (GCK) gene discovered by Next Generation Sequencing (NGS)

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Monogenic diabetes or maturity-onset diabetes of the young (MODY) is characterized by early onset (<25 years old), non-insulin dependence and autosomal inheritance. The major candidate genes include HNF4α (MODY1), GCK (MODY2) and HNF1α (MODY3). A slim lady (BMI 22.4 kg/m2) was diagnosed with Type 1 diabetes based on abnormal fasting glucose and oral glucose tolerance test at age 21. She was started on daily insulin injections (total daily dose 18-22 units/day) with good glycemic control (HbA1c 6.2%). Glutamic acid decarboxylase (GAD) autoantibody was negative. On occasions when she ran out of insulin supply, there were no incidences of diabetic ketoacidosis. These features atypical of type 1 diabetes prompted the team to perform genetic testing for MODY.

A novel missense mutation in the GCK gene (MODY2) was discovered by next-generation sequencing (NGS, Ion Torrent™, Thermo Fisher Scientific) and confirmed by Sanger's sequencing. In accordance with clinical practice guideline, treatment is not effective or needed in GCK-MODY and there is no evidence that pharmaceutical therapy in the doses used in GCK-MODY benefits glucose control. Genetic counseling based on best practice recommended for MODY2 was provided to the patient. Engagement of her family members was highly encouraged as off springs of the affected parent might have inherited the GCK gene mutation. After careful consideration, the patient accepted the recommendation of weaning off all insulin (and anti-diabetic) therapy.

The successful treatment-cessation based on accurate genetic diagnosis (informed by precise phenotyping) can improve clinical outcome and quality of life. Genetic counselling and testing are vital especially to well-phenotyped patients and family members as early detection with an accurate diagnosis may inform specific choice of therapy as well as avoiding unproductive (sometimes hazardous) diagnostic odyssey and treatments. Diagnosing MODY2 accurately has also opened-up the opportunity for the patient to be reconsidered for health-insurance coverage eligibility, some of which may be biased against individuals with type 1 or 2 diabetes.

The lived experiences of adolescents and young adults with Li-Fraumeni Syndrome: are their psychosocial needs being met?

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Adolescents and young adults (AYAs: aged 15-29 years) with Li-Fraumeni Syndrome (LFS) have significantly increased risks of developing primary malignancies at multiple sites from an early age. AYAs with LFS experience their transitional life stage of emerging adulthood concurrently with increased cancer risk and the potential of a reduced lifespan. The prospect of intensive life-long surveillance and, for some, cancer treatment may create a complex and unique set of psychosocial needs that remain inadequately understood. The aim of the present study is to explore the lived experiences and identify the psychosocial support needs of AYAs who live with, or at risk of, LFS.

Thirty participants aged 15-29 years with, or at 50% risk of having, a TP53 germline mutation are being recruited across Victoria, Australia. Participants complete semi-structured interviews examining the psychosocial impact of LFS for AYAs. Data analysis is ongoing, and informed by interpretive phenomenology.

We present preliminary data from six interviews collected during the first three months of this ongoing qualitative interview study. All young participants understood their high cancer susceptibility but possessed a varied understanding of which cancers are associated with LFS. Females were able to detail their age-specific breast cancer risk. All participants felt their identity was not defined by LFS, and that genetic testing and comprehensive screening provided them control over their future. Two participants reported LFS-related distress and had sought out professional support. Four were enrolled in research-based or personalised comprehensive screening programs and felt that screening offered them a sense of control over LFS. One was concerned about potential screening fatigue associated with longitudinal comprehensive screening and one did not engage in MRI-based screening due to anxiety of confined spaces. One young female had sought consultation to undergo bilateral prophylactic mastectomy.

These preliminary data suggest young people with, or at risk of, LFS have a range of genetic and health literacy. Some experience LFS-related distress, and this may be addressed by active risk management strategies and seeking professional support. The experiences of AYAs in this context are still under exploration to develop a better understanding of their ongoing psychosocial support needs.

“Just another straw on the stack” – Men with Lynch Syndrome’s experiences of recontact about potential increased risks of prostate cancer

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In cancer genetics, efficacious risk management strategies are available to carriers of high-risk germline mutations (e.g., Lynch Syndrome). Additional cancer risk information has important implications for the clinical management of these individuals, including surveillance and treatment options. Preliminary findings suggest that individuals affected by other inherited conditions (e.g., cystic fibrosis) may wish to be recontacted about additional risk information if it is perceived as useful for improving individual and familial health. However, to date in cancer genetics, no known studies have examined how carriers of high-risk germline mutations experience the receipt of additional cancer risk information. Recontact in this context is especially problematic when corresponding effective risk management strategies are yet to be established.

The aim of this qualitative study was to explore how men with Lynch Syndrome understand and experience the return of uncertain prostate cancer risk information and its influence on their health behaviours.

Using a modified grounded theory approach, sixteen men with Lynch Syndrome were purposively recruited from the Australian IMPACT study (Identification of Men with a genetic predisposition to ProstAte Cancer: Targeted screening in men at a higher genetic risk and controls) to undergo a semi-structured interview.

The majority of men (mean age 51 years) acknowledged that they may be at above population risk of prostate cancer, though evidence linking Lynch Syndrome and prostate cancer was still emerging. Many men felt that their risk of prostate cancer was overshadowed by the high-risk status of Lynch Syndrome; a potential prostate cancer risk was "just another straw on the stack". The offer of targeted prostate cancer screening via enrolment in IMPACT appeared to moderate the emotional effects of receiving additional cancer risk information. Consequently, men's experience of recontact was characterised by low cancer worry and acceptance. Overall, the men demonstrated high engagement with personal and familial health.

Findings suggest that participants integrated new prostate cancer risk information into pre-existing belief-sets regarding Lynch Syndrome, including the importance of regular screening. Recontact was found to have a limited emotional impact, suggesting these men were well adapted to their high-risk status. Participants' high levels of engagement in screening played a critical role in their perceived sense of control over their cancer risk. This sample was uncharacteristically engaged in personal and familial health compared to general men's health literature. Optimally, new cancer risk information from research should be returned in tandem with the offer of research-based or clinically available cancer-specific screening.

Young Australian women's decision-making about managing breast cancer risk due to a BRCA1/2 mutation

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Young women aged 18-40 years with a BRCA1/2 mutation experience their young adulthood, a formative developmental life stage, interwoven with an awareness of significantly increased cancer risks. Tension may exist between engaging cancer risk management strategies (breast screening with or without chemoprevention, bilateral prophylactic mastectomy (BPM), bilateral salpingo-oophorectomy) and normative developmental tasks, including, forming intimate relationships, and childbearing and rearing. This study explores how young Australian women with a BRCA1/2 mutation experience their young adulthood while engaging breast cancer risk management strategies.

Data were collected using a grounded theory approach with qualitative, semi-structured interviews. The inclusion criteria included women who have a BRCA1/2 mutation, aged 18-40 years, who received their genetic test result more than 12 months prior. Data were analysed iteratively and inductively to identify themes, ideas, concepts and categories.

Forty semi-structured interviews were conducted with women aged 20-40 years. Fertility, childbearing, and children (future or existing) were key considerations for women when choosing how to manage their breast cancer risk. Most of the younger women (≤30 years) in this cohort chose breast screening because they felt 'too young' to have a bilateral prophylactic mastectomy (BPM). These women also wanted to wait until after they had completed childbearing to potentially have a BPM due to their desire to breastfeed. Reproductive planning was influential in women's choice regarding chemoprevention. Most who were offered this medication declined to use it so they could conceive if they chose to within the following five-year period. The women who had a BPM at the time of interview frequently described future or existing children as their reason for pursing this surgery to negate any potential impact of breast cancer on their fertility or existing children.

These findings indicate the women who participated in this study prioritised their fertility and normative developmental tasks involving childbearing and rearing when making decisions about how to manage their breast cancer risk. This indicates these women continue to make decisions about risk management strategies long after the genetic counselling and testing process. Hence, there is a need to ensure these women have access to longitudinal care from genetic health professionals to provide evidence-based information about risk management options. This care could include personalised, empirical risk assessment in short term increments to facilitate informed decision making about cancer risk throughout their young adult stage of life.

Application of Gerard Egan's Skilled Helper Model to Genetic Counselling Practice

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Almost 25 years ago, Kenen & Smith (1995) wrote about practice models for the genetic counselling profession going into the future. They predicted that "innovative genetic counsellors with expanded visions of counselling goals and practices will play an increasingly important role in the future of genetic counselling" (Kenen & Smith, 1995, p115). In this presentation, I explore the application of Gerard Egan's Skilled-Helper Model (Egan, 2014) for use in genetic counselling. The model has been taught in continuing professional development courses in regional genetics centres in the United Kingdom, with Open Awards accreditation for Solution-focussed Counselling (see http://openawards.org.uk). However, there is no published literature about incorporating the Skilled Helper Model and its associated theory into genetic counselling practice on a local or global level. I will begin by outlining historical discussions about using psychotherapeutic techniques and models in genetic counselling, and the argument for further development in this area. I will then present the Egan (2014) model and compare it with the Reciprocal Engagement Model, which itself has been proposed as a means of conceptualising genetic counselling practice in the United States of America (McCarthy Veach et al., 2007). Finally, I will explore how the Skilled-Helper Model may be applied in genetic counselling practice; as well as the benefits and limitations of such a model.

The social dilemmas of offering Preimplantation Genetic Diagnosis (PGD) for Huntington's disease by exclusion testing at The Centre for PGD, Guy's Hospital

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Huntington's disease (HD) is an autosomal dominant, late-onset disorder, caused by a mutation within the HTT gene. Some individuals prefer not to know their carrier status, but still wish to prevent the birth of a carrier child. For these patients, prenatal diagnosis has been available for many years with the option of termination of pregnancies at 50% risk of HD, known as exclusion testing. The main disadvantage is that couples may be terminating a pregnancy that is not at risk of HD. Preimplantation genetic diagnosis (PGD) is an alternative option and is offered to couples using embryos created in vitro. Only embryos free of HD risk are used for embryo transfer after being selected using exclusion testing. Similarly the main disadvantage is that unaffected embryos may still be discarded if the parent at risk (50%) has not inherited the HD gene. Factors around exclusion testing are complex and require extensive consideration.

The Centre for PGD at Guys Hospital has been offering PGD since 1999 and the second most common referral indication for PGD is HD. We offer both HD direct and exclusion testing on blastocyst embryos. Between January 2014 and July 2017 we undertook 106 cycles of PGD using HD exclusion testing. Here we present three cases that highlight the ethical and social dilemmas surrounding PGD with HD exclusion testing.

Case 1: female A developed a life threatening haemorrhage following egg collection. Her partner was at 50% risk of HD. Case 2: female B's partner was also at 50% risk of HD. After several unsuccessful PGD cycles the at- risk partner reluctantly opted for HD testing which was positive. They subsequently conceived spontaneously and had 3 terminations affected pregnancies. Case 3: female C (at 50% risk of HD) focuses on PGD test development using microsatellite linked markers that were uninformative. To ensure accuracy of the PGD test, the partner's HD status had to be established. We will discuss these cases in relation to the dilemmas of having non 'at-risk' partner go through PGD treatment, health risks surrounding multiple PGD cycles, recurrent failed cycles of PGD, discarding embryos which are at 50% risk, HD testing of the unaffected partner for PGD work-up and appropriate use of NHS resources.

Section of Genetic Counseling at CHOP: Organizing for Advocacy and Professional Development.

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The Section of Genetic Counseling at Children's Hospital of Philadelphia (CHOP) was created in 2014 to provide a unified professional voice for all genetic counselors across the institution, aid in professional development and promote collaboration. The purpose of this presentation is to provide insight into the process of formalizing the Section of Genetic Counseling at CHOP, outline internal leadership and committee structure, and highlight initial three year accomplishments. Key deliverables of the Section include an institute-wide salary review that resulted in adjustments for new hires and employed genetic counselors, development of an enhanced career ladder for professional advancement, creation of a centralized system for student placement, institution of incident-to billing for genetic counselors, organization of educational and promotional activities, and provision of onsite CEU opportunities. The methods and structural organization employed to achieve these deliverables will be reviewed.

The Section of Genetic Counseling at CHOP currently has 44 members practicing in a wide array of disciplines including pediatric genetics, metabolic medicine, prenatal diagnosis, hereditary cancer, cardiology, neurology, internal medicine, research, and pathology. The strategies utilized by the Section have not only resulted in increased camaraderie and collaboration amongst genetic counselors but also in increased visibility as well as recognition of the profession at the institutional level across different disciplines. Here we will offer strategies for genetic counselors and other professionals to identify opportunities for advocacy and professional growth, recognize potential challenges to formal organization and present examples of forthcoming projects within the Section which aim to advance professional development of genetic counselors across the country.

Review of Outcomes Measures in Genetic Counselling

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Evaluation of services is essential for providing consistent high quality healthcare. Numerous studies have both defined and measured outcomes of genetic counselling and clinical genetics services in various ways, but there remains a lack of consensus about the most suitable approach. The field of genetic healthcare is expanding rapidly with the integration of genomic technologies, mainstreaming, and direct-to-consumer genetic tests becoming increasingly available. It is an important time to clearly define the potential benefits of genetic healthcare, and to assess how best to evaluate genetic counselling practice to ensure consistent goals are being met in a changing field.
I will present a review of qualitative and quantitative studies regarding the intended outcomes of genetic counselling and the existing tools to measure these outcomes. Furthermore, I will highlight some of the complexities and challenges, including the existence of hundreds of desired outcomes of practice but few validated measurement tools available, discrepancies in the views of patients compared to genetic counsellors regarding what they perceive to be the benefits of genetic counselling, and outcomes that have not yet been studied in depth in the literature (for example the therapeutic alliance). I will propose suggestions for overcoming some of these challenges by identifying areas for further collaborative multidisciplinary research.

Making genetic counselling services accessible - Results of a pilot service delivery feasibility study of telemedicine appointments

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The Merseyside & Cheshire Clinical Genetics Service provides genetic counselling to a population of 2.8 million, including a population of approximately 80,000 living on the Isle of Man. In 2017 the clinical genetics department trialled a telemedicine service as an alternative to traditional face to face consultations.

Telemedicine is not widely used in genetic counselling. A study comprising of 104 clinically active European respondents from 30 different countries identified only 9% currently use telemedicine or videoconferencing facilities (Otten et al., 2016).

This small pilot study was developed to test the feasibility and acceptability of a telemedicine service for patients referred for, cancer or general genetic counselling, and in particular to reduce travel for patients currently travelling by plane or boat from the Isle of Man, in the hope of making genetic counselling more accessible.

Prior to the trial, 21 clinical genetics patients were surveyed regarding the general acceptability of telemedicine, of which two thirds replied positively. However in the trial, of the 60 patients offered a telemedicine appointment only six accepted. Following their appointment, patients and genetic counsellors completed a questionnaire about their experience. Advantages included convenience for the patient and reduction in travel, which were consistent with the findings of Buchanan et al., (2015) and Otten et al., (2016). However, issues such as; reduction in quality of counselee-counsellor interaction, technology problems, reliance on IT provider input and genetic counsellor training of the system were all highlighted as problematic. These results are consistent with those of a pilot study of 51 consultations in The Netherlands.

We conclude, whilst telemedicine was acceptable to some patients, others preferred a face to face consultation. The IT system needs to be robust and support from IT consultants was required to set up each consultation. Overcoming these problems would enhance the acceptability to some patients and genetic counsellors.

How practical experiences, educational routes and multidiciplinary teams influence genetic counsellors' clinical practice in Europe

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The need for genetic counsellors increases rapidly in the world. Still, genetic counsellors' roles in health-care services are not harmonized, and in most countries in Europe the profession is still emerging. Also the educational and experiential backgrounds diverge noticeably.
This study aimed to explore how genetic counsellors' characteristics impact on their tasks in practice. We focussed on relevant tasks of genetic counsellors, according to themselves and according to the medical geneticist colleagues.
The results are from the quantitative part of a mixed-method study. Participants were ascertained via national and international associations. The sampling involved the snowball technique and participants were invited via an email with an online survey. Various statistical analyzes was conducted.

104 genetic counsellors and 29 medical geneticists providing genetic counselling completed the questionnaire, representing 15 European countries. Results showed that most genetic counsellors in Europe perform similar tasks, irrespective of their backgrounds. Factors influencing genetic counsellors' roles showed that the type and volume of tasks performed by genetic counsellors is associated with the years of experience in the field, not with with their background or education. Genetic counsellors and medical geneticists both agreed that tasks with more psychosocial implications were seen as genetic counsellors' responsibility while tasks with more medical implications were seen as medical geneticists' attribution.
In summary, genetic counsellors work in tune with international recommendations and seem to be supportive of multidiciplinary teams. Analysis points to the importance of practical experiences, which can have implications for practice and training in genetic counselling.

Complementariness between medical geneticists and genetic counsellors: its added value in genetic services in Europe

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New knowledge, discovery of new rare conditions, availability and complexity of new genetic tests, and new legislation within genetics progressed significantly the last few decades The need for non-medical health-care professionals working as genetic counsellors has therefore increased rapidly in Europe and worldwide. However, there is no unified approach to genetic counsellors' role in health-care services in Europe, as in most countries the profession is still emerging and the educational backgrounds diverge noticeably, within and between countries.
This qualitative study aims to describe the potential added value of genetic counsellors in clinical genetics teams and to explore their tasks and responsibilities in different European countries.
This study was based on the qualitative part of a mixed-method study. Potential participants were ascertained via national and international associations, and received an invitation letter, a consent form and a link to the online survey via email. The sampling was based on a snowball technique. A total of 143 participants providing genetic counselling in Europe at the time of the survey responded. Thematic analyzes were conducted to analyze the open ended questions from the survey.
The results show differences in activities of genetic counsellors, although there is a wide range of roles, which are similar. The ability to establish a quality relationship with consultands was frequently mentioned as one of the strengths of genetic counsellors, as well as a patient-centred approach. It is believed that genetic counsellors add a more holistic approach of psychosocial and familial dimensions of genetic concerns to the multidisciplinary teams.
This study provides examples of successful integration of genetic counsellors in teams, as complementariness with medical geneticist became clear in several cases. Although the added value of genetic counsellors was manifested, professional recognition of genetic counsellors across Europe is still needed in order to support the quality of patients care and safety of practice.

How do carriers of Barth syndrome navigate reproductive options? A potential fault line for patient-based support organizations

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Barth syndrome (BS) is an X-linked cardiomyopathy characterized by pediatric onset, neutropenia, and skeletal myopathy caused by mutations in tafazzin (TAZ). Carriers are unaffected, but navigate psychological challenges and increasingly complex reproductive options. A multinational patient organization, the Barth Syndrome Foundation (BSF), plays a prominent role in not only supporting families but also setting the research agenda. To identify and describe common psychological and reproductive challenges of carriers and explore the role of social support in the context of a patient support organization, we conducted semi-structured telephone interviews of 28 adult carriers recruited through the BSF. Interviews were recorded, transcribed, double-coded, and analyzed for common themes. Half of participants were American, one-third from other English-speaking countries, and the remainder non-native English speakers. While guilt was experienced by most, but not all, mothers and grandmothers, relationships among carriers via the BSF ameliorated distress by normalizing guilt. In contrast, participants held starkly different views of advanced reproductive technologies. Many expressed a strong desire for additional information and reported uneven experiences with genetic counseling. For a few, any prenatal testing was unacceptable, but for many considering reproductive options was both morally and practically/financially challenging. While practical considerations varied based on nationality, most mothers who considered pre-implantation or pre-natal diagnosis after having an affected child described a difficult decision-making process, likening affected embryos/fetuses to their sons. A few reached decisions that conflicted with long-held beliefs. In contrast to the strong mutual support carriers reported regarding the medical and practical aspects of BS, nearly all were wary of discussing reproductive planning within the BSF. While nearly all stressed that their choices should not be normative for or influence others, most feared damaging relationships by broaching these personal and potentially political topics. Several described fears of imperiling the success of nascent clinical trials should relationships be damaged. These results suggest enhanced social support for reproductive decision-making may best come from sources outside patient-based support groups. Our data also highlight potential fault lines leaders may encounter as members confront potentially polarizing issues surrounding reproductive technology.

The information and emotional support needs of Grandparents with Pompe Disease

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Grandparents of children with special needs have unique family roles and complex emotional experiences. Previous research has studied grandparents of children with X-linked diagnoses that are untreatable. Treatable, inherited conditions, such as Pompe disease (PD) where inheritance is recessive have not been studied. The availability of treatment and heritable nature present the possibility for unique grandparent roles, experiences and needs. Newborn screening for PD in the USA makes this study timely. The study described grandparents' roles and involvement, identified grandparents' information and emotional support needs and explored the psychosocial impact of having a grandchild with PD. An online survey containing forced choice and open-ended questions was distributed by various PD organizations. Data were analyzed with descriptive statistics, statistical measures, and thematic analysis. Twenty-one grandparents of children diagnosed with PD participated. Grandparents provided emotional support more frequently than financial support. Grandchildren's parents were the primary sources of information for grandparents. Information about treatment was most important to grandparents. Most participants learned about the genetics of PD (n=16) and understood PD's genetic etiology (n=15). Grandparents identified family and religion as the most valuable sources of emotional support, but also commonly received emotional support from friends (n=16) and Internet resources (n=¬15). Psychosocial impacts included altered travel and employment plans, increased awareness of grandchildren's limitations and medical needs, and the experience of double-grief. While grandparents are large sources of support for their families, they need considerable support themselves, yet resources for grandparents beyond their grandchild's parents are limited. These results warrant genetic counselors' consideration of extended family members' support needs surrounding a genetic diagnosis and facilitation of familial communication of complex medical information.

Up-skilling genetic counsellors to provide additional findings from genomic sequencing.

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There is an increasing expectation that patients having genomic sequencing should have "additional findings" returned. The Melbourne Genomics Heath Alliance is planning a proof-of-concept additional findings service for adult participants who have had whole exome sequencing for a clinical indication. In preparation to deliver this service, we designed a workshop to a) enhance skills in counselling patients who present for additional findings b) explore their views about service delivery models.

Development of the half-day interactive training workshop was guided by the principles of adult learning, experiential learning and reflective practice. It had three components:
1. a didactic presentation from an international expert genetic counsellor with experience providing a multi-disease panel test to healthy individuals.
2. reflection on two live counselling simulations performed by experienced genetic counsellors.
3. case-based role plays by participants followed by group debriefing/reflections.

A survey to assess confidence, knowledge and views about service provision was administered before and after the workshop. Thirty-eight genetic counsellors attended (89% female). Thirty five (92%) returned survey 1 and 30 (79%) returned survey 2. All respondents rated the workshop as useful. After attending the workshop:

- 75% of participants indicated they were confident/very confident to provide counselling for additional findings compared to 34% before the workshop.
- 81% of respondents indicated that gaps in their knowledge were addressed.

In regard to service provision for additional findings, 56% of respondents felt that additional findings should be offered some time after diagnostic genomic test results were returned and respondents rated genetic counsellors and clinical geneticists as the most preferred health professionals to provide pre- and post- test counselling.

Genetic counsellors have the core skills and knowledge to provide counselling for additional findings. A half day workshop was effective in increasing genetic counsellor confidence and knowledge in this setting.

Practical Tools for Managing Uncertainty in the Genomics Clinic

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Genomic medicine is fast evolving and making its way into the genetic counselling clinic. It is important for Genetic Counsellors to have to hand a psychosocial toolkit of counselling skills that can be employed to deal with "known unknowns" associated with Genomic test results. Some studies suggest that pre-test discussions about the possibility of uncovering such variants are paramount to prevention of psychological harm 1. Other studies state that adapting to and developing resilience towards existing uncertainty is important 4. Colleagues have considered the different types of uncertainty in healthcare and genomics 2, 3. We consider the application of particular psychological approaches that might be adopted by the genetic counsellor to aid a client's management of uncertainty and we apply this to case specific examples. Techniques considered include mindfulness practice, cognotive behavioural therapy and acceptance and commitment therapy.

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Celebrating molecular screening of over 10,000 families with inherited cardiac conditions: reflecting on past practice and future challenges

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In 2003, when the Oxford Medical Genetics Laboratory (OMGL) introduced an NHS diagnostic genetic testing service for inherited cardiac conditions (ICC), the main challenge facing laboratories was variant detection. By 2016, when the OMGL cardiac service reached the milestone of testing its 10,000th family, technology had not only evolved, but had been revolutionised by massively-parallel sequencing. This meant that the issue of variant detection had virtually been conquered, only to be replaced by new challenges of data handling and variant interpretation.

The demands on the cardiac service in Oxford are drivers for improvement and development. Data presented will demonstrate the effects of technological and procedural advances on the service, plus associated patient benefits. The introduction of bioinformatics tools and pipelines was also driven by the burden of data handling, which continues to increase with expanded gene panels and progression towards exome/genome sequencing for diagnostic services. In addition, we will reflect on the way that our genetic counselling practice changed to adequately prepare patients for uncertain test results as well as clearer ones.

Close collaboration with the clinical service delivering specialist ICC care in Oxford has facilitated and improved variant interpretation. In celebrating this milestone, the patients to whose care we have contributed shared their stories with the media, explaining the effects that genetic testing and counselling had on their families. The team were humbled by their openness in sharing their experiences, several of which included the sudden loss of relatives. We believe that this truly multidisciplinary approach between cardiac, clinical genetics and expert molecular testing services continues to deliver excellent specialist care to patients and families living with ICCs.

The Oxford cardiac model has shown that depth of knowledge and collaborations formed during analysis of large patient cohorts is vital in facilitating high quality variant interpretation. This model could be enhanced through sharing of data and knowledge between laboratories, which will be vital for the translation of the 100,000 Genomes Project into routine genomic testing services.

The G2NA: A global genomics nursing alliance to accelerate integration of genomics into everyday professional practice

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As the largest single healthcare professional group worldwide nurses have a pivotal role in: routine family history assessment; identifying people who would benefit from a consultation with a genetic specialist; facilitating referrals; and beginning the first conversations about genomics. However global effort is needed to transform nursing policy, practice, education, and research. The Global Genomics Nursing Alliance (G2NA) has been established to accelerate the integration of genomics into everyday practice. G2NA is not targeting the genetic specialist, but is aimed at everyday nursing practice and education through the sharing of resources, expertise, and mobilization of organizations that can help influence nursing leaders and policy directions.

The inaugural G2NA interactive meeting was held in early 2017 with delegates representing 19 countries and 7 organisations. All delegates either agreed (26%) or strongly agreed (74%) that the G2NA should collaborate with the interprofessional community. Only 3 countries indicated existence of genetic/genomic competencies applicable to all nurses regardless of clinical role, level of training, or specialty: Japan; UK and US. Six countries reported visible leadership driving developments in nursing to incorporate genomics. The top three priority areas for future action included: raising awareness; education; and resources to support genomics in nursing.

Outputs from this meeting so far have included: Meeting Reports to Wellcome Trust and Health Education England Genomics Education Programme, Resource Summary Report to the National Human Genome Research Institute (NHGRI), Genomic HealthCare Branch, the G2NA website www.G2NA.org , video, list-serve and resource repository for G2NA members and a Landscape Analysis of Global Genomic Healthcare Services and Nursing (submitted).

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NEXT GENERATION PHENOTYPING: A PERFORMANCE ANALYSIS

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FDNA

Introduction
While sequencing technologies are continuously progressing, diagnostic yield remains limited without use of patient phenotype data. Facial analysis provides a promising advancement in this area. In this study, we assessed the performance of the FDNA technology that automatically identifies facial patterns associated with genetic syndromes by analyzing two-dimensional facial photos.

Method
To train the system, images of tens of thousands of patients diagnosed with over 2,200 different genetic syndromes have been collected from users of Face2Gene, analyzed, and the de-identified visual information extracted from the images is stored in a secure database. Once the system has been sufficiently trained to recognize the visual markers of a particular syndrome, a new "syndrome-classifier" is deployed in the system. Any new image is analyzed and ranked based on similarities to each specific syndrome-classifier. To measure the classification success, we used the area under the curve (AUC) of the receiver operating characteristic (ROC) curve. An AUC of 100% indicates perfect accuracy, and an AUC of 50% indicates accuracy no better than the flip of a coin. In this study, we assessed the accuracy of all Face2Gene syndrome-classifiers.

Results and conclusions
The top 100 performing syndrome-classifiers demonstrated an averaged AUC of over 98%. The top 200 and top 300 syndrome-classifiers demonstrated average AUC's of over 96% and 94%, respectively. The high performance across hundreds of syndrome-specific classifiers show that FDNA 's technology can be useful for medical professionals in the clinic and to facilitate gene-targeting and variant interpretation.

Experience with genetic counseling: the adolescent perspective

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Adolescence is a complex period of development that involves creating a sense of identity, autonomy, relationships and values. This stage of adjustment can be complicated by having a genetic condition. Genetic counselling can play an important role in providing information and support to this patient population; however, resources and guidelines are currently limited. In order to appropriately establish genetic counselling approaches and resource development, we investigated the experiences and perspectives of adolescents with a genetic condition with respect to their genetic counselling interactions. Using a qualitative exploratory approach, eleven semi-structured interviews were conducted with adolescents diagnosed with a genetic condition who received genetic counselling between the ages of 12 and 18 years at The Hospital for Sick Children. Transcripts were analysed thematically using qualitative content analysis, from which three major interrelated themes emerged: 1) understanding the genetic counsellor's role; 2) increasing perceived personal control; and 3) adolescent-specific factors influencing adaptation to one's condition. Additionally, a list of suggested tools and strategies for genetic counselling practice were elucidated. Our findings can contribute to the development of an adolescent-focused framework to enhance emerging genetic counselling approaches for this patient population, and can also facilitate the transition process from paediatric to adult care within patient and family-centred contexts.

A review of symptoms observed in patients with low penetrance HD alleles

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The literature suggests that individuals with low penetrance HD alleles (36-39 repeats) will remain asymptomatic until a very late age. Our local ancedotal experience is that we have a number of cases with low penetrance alleles which show the same disease course as an individual with a complete penetrance allele (40 and over repeats). As such we are cautious when counselling individuals as to whether they will remain asymptomatic until older age or if indeed they may never develop symptoms. The review looks at the HD population in the Northern Genetic Service region, specifically at the individuals repeat size, if they are manifesting any symptoms and at what age these symptoms developed. The purpose of the review is to inform our genetic counselling practice and we present our findings.

Patient decision-making and the role of the prenatal genetic counsellor

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Objective: When a prenatal screening result indicates an increased risk for aneuploidy in a pregnancy, parents face the difficult decision of whether or not to have an amniocentesis. There is much theory about how genetic counselling facilitates prenatal decision-making, yet little research, and limited data regarding patients' perceptions of the process and the role of the genetic counsellor.

Methods: We performed a qualitative study to explore patients' decision-making regarding amniocentesis after prenatal genetic counselling related to increased risk for Down syndrome or Trisomy 18, with a focus on the perception of the role of the genetic counsellor (GC) in the decision-making process. Semi-structured interviews were conducted with patients, transcribed verbatim, and qualitatively analyzed using a constant comparative method. To complement the qualitative data, a secondary quantitative measure was employed - the 6-item Satisfaction With Decision-making (SWD) scale. The patients' GCs completed a questionnaire about the encounter comprising open-ended questions to provide some limited source triangulation.

Results: Eleven patients participated and four predominant themes emerged: 1) being unprepared; 2) accepting responsibility for decision-making; 3) the burden of responsibility; 4) the impact of support through affirmation. Despite the underlying tension within some of these themes, patient SWD scores indicated they were highly satisfied with their decision (mean score = 28.5/30, range: 26-30). The participating GCs reported not directing or influencing their patients, which was consistent with overall patient reports of the GC.

Conclusion: Patients felt affirmed but not swayed, and perceived the GCs to be non-biased yet supportive of their emerging decision. At least some elements of models that have been proposed to describe genetic counselling practice (shared decision making, reciprocal engagement) appeared to be present in this naturalistic study. However, the theme of 'being unprepared' did emerge much more strongly than anticipated and therefore is likely to have significant implications for the decision-making process. Further investigation is needed.

“An Empowering Encounter”: Exploring how the process of genetic counselling influences outcomes for individuals with mental illnesses

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Genetic counselling (GCing) for individuals with mental illness (MI) has been shown to improve patient outcomes, such as internalized stigma and perceived control; however, what is not clear is how GCing influences outcomes. In this qualitative study, we explored participants' experience living with MI, their experience receiving GCing, and their perceived impact of GCing on their lives.
Adult individuals with a diagnosed MI were recruited from two sources: the ADAPT clinic, which provides psychiatric GCing, in Vancouver, Canada; and a quantitative study led by our research group on the effect of GCing on medication adherence. Ten individuals were interviewed prior to receiving GCing and one month following counselling. Interview transcripts were analyzed using grounded theory methodology to generate a theoretical model on the process and outcome of GCing for individuals with MI.
Participants described GCing to be an "empowering encounter" that reportedly had an immediate and positive impact on their lives. Participants shared that they gained a "new perspective" on their MI due to the support (conceptualized as "being heard", "feeling normal", and "being validated") and information (defined as knowledge, tools, and resources) they received during counselling. Participants identified attributes of the session (described as "engaging", "presenting accessible information", "offering personalized information", "addressing questions") and the genetic counsellor (characterized as empathetic, nonjudgmental, trustworthy, knowledgeable) that contributed to their new outlook. Consequently, individuals felt empowered and reportedly more able to manage and talk about their MI. Participants also seemingly experienced a reduction in self-stigma and feelings of shame and blame.
The theoretical model generated in this novel study provides a better understanding of how GCing impacts patient outcomes and highlights aspects of the GCing process that most effectively and positively influence outcomes. The findings support the standard provision of GCing to all individuals with MI and can be used to inform the development of psychiatric GCing guidelines. Clinical guidelines would provide direction to both mental health and medical genetics care providers on how to provide appropriate support, education, and counselling and ensure the greatest benefit for patients. Such guidance is critical since many mental health care providers have limited knowledge on genetics and few genetic counsellors have received training in psychiatric GCing.

Prenatal chromosomal microarray analysis- what results do parents want to receive, and who should decide?

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Chromosomal microarray analysis (CMA) has become the first-line genetic test in pregnancies with fetal abnormalities detected via ultrasound. A growing number of centers worldwide are offering CMA to all women undergoing invasive prenatal testing. The major advantage of CMA is its higher detection rate of abnormalities, which is helpful for parents in managing present and future pregnancies. Nevertheless, the higher resolution at which the genome is examined means that chances are higher to identify: (1) variants with uncertain clinical significance (VUS); (2) susceptibility loci (SL); and (3) Copy -number-variants associated with adult-onset conditions. Implementation of CMA for prenatal testing is debated and no international consensus has been reached. In our centre, CMA is the first-tier test offered to all women undergoing invasive prenatal testing, for all indications, including advanced maternal age and parental anxiety. As of June 2017, women are asked to choose whether, in addition to diagnostic results, they wish to receive the following findings: VUS, SL, and risks for adult-onset conditions. Pre-testing, a group slide-presentation is shown to the women and their partners about the potential findings from CMA. A questionnaire is then delivered, aimed at evaluating the satisfaction from the explanation given; satisfaction from the choices given to them; and their understanding of the test's potential findings.
Eleven percent of women did not wish to know any of the findings for which choices were given. About half of the women chose to be informed of VUS; and three quarters (75%) wished to learn about SL and adult-onset conditions.
Two thirds (64%) of respondents to the questionnaire were satisfied with the explanation, whereas a third (36%) were either unsure, or had additional questions.
The majority of the respondents (78%) were satisfied with the choices given to them. All respondents replied correctly to the knowledge questions about the possible findings from CMA.
Our findings suggest that a group explanation is efficient in preparing women for CMA testing in pregnancy, and that couples can make informed choices regarding what findings are reported back to them. The possibility of face-to-face genetic counselling should be given to couples who feel uncertain about their choice, or wish to have additional information.

To review histology of patients tested for MAP to improve understanding of the MAP phenotype.

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Aim
To review histology of patients tested for MAP to improve understanding of the MAP phenotype.

Methods
We obtained a list of patients in who we requested MAP testing from January 2009 to December 2014. We reviewed the number and type of polyps and age of polyp/bowel cancer diagnosis.

Results
Common mutation testing or a full screen was completed in 122 families; 8/122 (6.6%) patients were MUTYH compound heterozygotes for pathogenic mutations or likely pathogenic variants of unknown significance, 2/122 (1.6%) were carriers and 105/122 (86%) had no mutations identified.

Six of the eight patients with two mutations/variants had at least one rare mutation. These 8 patients had 6 to ~30 polyps identified; most were low grade adenomas. The average age of polyp diagnosis was 54 and cancer diagnosis was 55.

Those without a mutation had a broader range of polyp type. Average age of polyp/cancer diagnosis was 45. Mutations were not found in any patients with serrated polyps or with isolated bowel cancer <35 years without polyps.

Conclusion
We have stopped testing isolated colorectal cancer cases <35 years or patients with <5 adenomas. We will offer a full screen if phenotype is classical. Serrated polyps appear to represent a phenotype distinct to MAP.

Pretest Counseling and Patient Autonomy in the Genomic Age

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Given the current state of medical genetics with rapid advancement in testing technology and a scarcity of qualified genetic counselors, our profession must adapt to meet growing and changing needs. However, as we evolve, it is important to remain cognizant of the history that the genetic counseling profession was born from and work to maintain the core values that the profession was founded on. Master's level genetic counseling emerged in parallel with the field of biomedical ethics in the late 1960's and early 1970's. Multiple forces including reproductive rights for women, advances in genetic testing technology, and the not-so-distant memory of atrocities of violations of human rights in the name of genetics, crystallized the purpose of the genetic counselor in the reproductive setting: to support patient autonomy and informed decision making, free from coercion. The current environment challenges our ability to maintain these priorities. The commercialization of reproductive genetic testing has led to intense and persuasive marketing to both patients and providers which can skew the balance of information and *promote testing* rather than support *individual choice*. Additionally, testing is increasingly being routinized through primary obstetrical care where patients often do not have access to pretest counseling by qualified professionals and may not appreciate that they have an active choice to make about whether to accept testing or not. The timing of genetic counseling is shifting to be offered only "post-test", if test results are abnormal. These factors may lead to unwanted information and outcomes that are inconsistent with the needs and values of individual patients. This presentation will discuss strategies for adapting with the rapidly changing landscape of reproductive genetics that will allow genetic counselors to continue to work effectively as patient advocates. Innovative tools and programs to improve education and provide patient support will be discussed including group presentations and outreach, educational videos, interactive decision support tools, telehealth and the use of social media. While maintaining a focus on our profession's primary core values we can optimize use of existing tools and develop new ones to deliver evidenced-based and patient centered genetic counseling in the genomic age of medicine.

Familial Communication of Positive BRCA1/2 Results: A Relational Dialectics Theory Approach

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Individuals who receive a genetic testing result revealing a pathogenic or likely pathogenic variant in the BRCA1 or BRCA2 (BRCA1/2) gene are encouraged to disclose this result to their biological family members. Various factors are known to influence the disclosure process and variations in the perceived importance of these factors make familial communication complex. The use of a theory grounded in the discipline of communication helped to explore the communicative processes involved in the familial communication of positive BRCA1/2 genetic testing results. By specifically focusing on the parent-adult child relationship, we gathered important knowledge on the unique dynamics that influence the BRCA1/2 experience in the individual and within the relationship. Semi-structured, dyadic interviews were conducted with an individual who received a positive BRCA1/2 genetic testing result together with his/her adult child. A total of fourteen dyadic pairs participated. Among the adult children, seven tested positive, three tested negative, and four had not pursued testing. Adult children seek parental input on testing and management options, but want to make their decisions autonomously. Family cancer history can impact how a parent and adult child contextualize personal and/or familial risk, sometimes inaccurately. Within the parent-adult child relationship, conversations related to the BRCA experience continue beyond the first disclosure. We discuss how our findings serve as a resource for clinicians to guide conversations with patients about the challenges and complexities of sharing results with family members, particularly with children.

Need for multidisciplinary approach in Ehlers-Danlos syndrome: the University Hospital Ghent experience

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Ehlers-Danlos syndrome (EDS) is a group of heritable disorders which affects the connective tissue that supports and provides tensile strength to the skin, bones, blood vessels, ligaments and many other organs and tissues. The latest classification recognizes six subtypes, including the hypermobility type, which is an autosomal dominantly transmitted disorder with variable expressivity and reduced penetrance. Lifelong follow-up in a multidisciplinary team is needed since the underlying genetic defect has thus far not been identified and hence laboratory diagnosis and curative therapies are currently not available. The hypermobility type of EDS may affect as many as 1 in 10.000 people. In 2014, the Ghent University Hospital has launched its multidisciplinary EDS clinic. The team consists of a geneticist, a genetic counsellor, physiotherapists, an occupational therapist, podiatrists, a psychologist and a social worker. After having been diagnosed by the geneticist, patients receive appointments with members of the team, according to their needs. An individual yearly follow-up scheme is offered to each patient.
Since the start of the clinic approximately 300 patients were recorded. Each patient has an individual file where the overall health status is registered and that is evaluated in a monthly multidisciplinary meeting.
The EDS hypermobility type comprises a clinically heterogeneous group of connective tissue diseases. Since there is no cure, a multidisciplinary approach is required to provide a preventive and symptomatic care. Organizing an EDS clinic where patients are seen on a regularly individual base, at one moment by several experienced specialists, is an added value in optimizing the care. This work aim to present the structure of our multidisciplinary team and the longitudinal follow-up of patients with EDS.

The current UK Genetic Counselling workforce

N.V. Taverner, on behalf of the Association of Genetic Nurses and Counsellors committee

All Wales Medical Genetics Service and Cardiff University

As genetics and genomics increasingly integrate into mainstream healthcare, the role of the genetic counsellor is changing. Genetic counsellors are uniquely placed to help families understand genomic information about their health and support them to use this information as part of their decision making. They also provide education and support for other healthcare professionals who are using genetic and genomic information in their practice. This changing role has implications for genetic counsellor workforce planning and training. The Association of Genetic Nurses and Counsellors (AGNC) committee, along with Health Education England, is looking into these issues. We have carried out a survey of the current UK genetic counselling workforce, including predictions of likely future demand, by requesting information from the regional genetic/genomic centres and from genetic counsellors working in other settings. Data from this survey are presented here, along with implications for the UK genetic counselling profession.

Handling of emails to the Cancer Genetics Unit (1st April – 31st May 2017)

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Patients are using email more frequently nowadays. Increasing use of email may be associated with information governance breaches or clinical risks. Time spent dealing with patient emails is not formally recognised and reimbursed financially.
The Cancer Genetics Unit conducted a retrospective audit to determine the volume of emails sent to the departmental email address over a two month period, work generated by these emails and any associated information governance breaches or clinical risks.
A total of 320 emails were received, 72(22.5%) of which were from patients. Of these, 35(49%) required clinical input (in the majority of cases this was giving clinical advice) and 43(60%) required administrator input. Of the total 320 emails, 264 (from patient, clinician or administrator) related directly to a Royal Marsden Hospital (RMH) patient. Of these 264, 32(12%) emails that contained important clinical information were not scanned to the patients' electronic patient record (EPR). The mean time to deal with an incoming email relating to an RMH patient - as assessed by the number of working days between email receipt and scanning to EPR - was 1.1 days. Information governance breaches were identified in 27/320(8.4%) incoming emails and/or the associated response, all of which involved transfer of person identifiable sensitive data between email routes that are not secure (e.g. NHS.net to NHS Trust). All patient emails were, by their nature, also associated with an information governance risk. Two emails were clearly associated with a clinical risk, both relating to a clinic letter being sent to the wrong address. The failure to scan 32 emails to EPR may also be considered a clinical risk.
This audit shows that emails were dealt with in a timely manner and required significant clinical and administrator input, making the case for recording email processing as clinical activity. It also highlights the need for a formal protocol to ensure emails are scanned to EPR where relevant and a departmental information governance email policy, including communicating to patients the risk of using email.

Loss, grief and bereavement within the spectrum of genetic counselling

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Genetic counsellors frequently meet with individual patients, couples and families who are currently grieving a loss or who have historically experienced a loss related to the inherited condition within the family. Loss, grief and bereavement are inherent aspects of the clinical genetics consultation, therefore, it is helpful for Genetic counsellors be familiar with grief theory, so we can identify when it may be impacting on the genetic counselling process. As well as the traditional loss through the death of a loved one there are multiple other ways in which loss is experienced by patients seen within the genetic counselling spectrum. Loss and therefore grief can be seen within many situations including predictive testing, in decision making around prenatal cases and the loss of the 'normal' family unit as a family member affected by the familial condition progresses through its natural course. I will draw on literature from traditional loss, grief and bereavement, apply it to case studies and present interventions genetic counselors could use within our time limited framework to help educate patients on the psychosocial issue of loss, to help patients recognise where they are in the grief process and how this may impact on their decision making in the genetic counselling process.

Informing relatives at risk of inherited cardiac conditions: a qualitative study with health care professionals, patients and relatives

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Inherited cardiac conditions (ICCs) can lead to sudden cardiac death at young age, but often remain undetected. Cardiac monitoring and/or predictive genetic testing is advised to relatives at risk. In current practice, index patients are asked to inform their relatives, supported by a family letter. This study investigated experiences with and attitudes towards this family-mediated approach in ICCs and explored whether and how improvements can be made. A qualitative study design was used. Two online focus groups with 27 healthcare professionals (HCPs), including cardiologists, clinical geneticists, genetic counsellors and psychosocial workers, and 20 face-to-face semi-structured interviews with index patients (n = 8) and relatives (n = 12) were conducted. Data were independently analysed by two researchers using a thematic approach. The findings show that HCPs as well as patients and relatives agree that in most situations, it is preferred that index patients inform relatives about genetic risks in ICCs. However, several barriers are perceived regarding the family-mediated approach. Both HCPs and index patients struggle with the dependency and the psychological and practical burden on index patients to inform their relatives. HCPs think they should take a more active role in informing relatives at risk to overcome these barriers. Index patients and relatives are of the opinion that ideally a tailored information provision strategy to inform relatives at risk should be used, adjusted to family dynamics and personality characteristics of relatives. In contrast, HCPs prefer uniformity in procedures to restrict the work load. In conclusion, these results show that although in most cases it is preferred that index patients inform relatives, several barriers are perceived. Whether the information should be uniform or tailored, the opinions of HCPs, patients and relatives differ. Further research is needed to assess the best suited approach to inform relatives at risk of ICCs.

Referred for genetic counseling, but no appointment: barriers or deliberate choice?

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Patients who are referred for genetic counseling to the Department of Clinical Genetics at the Erasmus MC, University Medical Center Rotterdam in the Netherlands, receive an invitation to make an appointment. However, more than 30% of these patients do not respond to the invitation. Because of health risks among this group of patients and/or family members, the question arises what the reasons are for not responding.
The objective of this study was to gain insight into motivations of referred patients who do not apply for an appointment for genetic counseling and to determine whether and which interventions are needed to reduce possible barriers.
The study was based on a cross sectional survey among 331 patients who were referred for genetic counseling but never applied for an appointment. The survey was performed in January/February 2016 at the department of Clinical Genetics at the Erasmus MC Rotterdam, the Netherlands, and showed a response rate of 31,4% (n=104). Results of the questionnaire were analyzed with the statistical software package SPSS.
Results showed that the majority of the patients (82,3%) who did not apply for an appointment, is interested in receiving genetic counseling. The enrollment procedure itself forms an important barrier as indicated by more than 50% of the respondents. Ambiguities in the invitation letter and troubles with filling in forms were important barriers. Of the respondents 66,4% indicates also other reasons, the most important being that the moment of referral was not convenient to the patient because of illness or treatment of the patient or family members at the moment of referral. Finally the study showed that patients were insufficiently informed about the nature and usefulness of genetic counseling.
In conclusion: Utilization of Clinical Genetic care was not optimal. Adjustment of the enrollment procedure is needed and referring physicians should improve and personalize the information they give to patients about genetic counseling.

Ethical considerations in returning genomic sequencing results in newborns

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The BabySeq Project is a randomized control trial assessing the medical, behavioral, and economic impacts of genomic sequencing (GS) in the neonatal period. Infants that receive receive GS have a disclosure session with a genetic counselor and physician to review identified monogenic childhood-onset disease risk and recessive carrier status results. Prior to disclosure, all potentially returnable results are evaluated by the study team according to an objective framework. Variants are assessed on three criteria: 1) childhood-onset disease association of the gene, 2) pathogenicity, and 3) penetrance. When a variant occurs in a gene with childhood-onset disease risk, is likely pathogenic or pathogenic, and is highly penetrant, it is reported. Complication arises when the variant meets some, but not all, of these criteria, necessitating expert case-by-case analysis. For example, when a pathogenic variant in a childhood-onset disease gene with moderate penetrance is identified, the actionability of the condition is assessed. If actionability is high (i.e. there is available screening/management), the benefit to parents is generally felt to outweigh the uncertainty that accompanies reduced penetrance. These conversations on the intersection of penetrance and actionability focus on considerations such as paternalism, duty to warn, and nonmaleficence. We present three cases of inherited variants in KCNQ4, ELN, and RB1 that highlight these ethical complexities. When a heterozygous variant meeting the above criteria is identified in a gene associated with recessive inheritance, it is returned as carrier status. As our study is limited to returning childhood-onset disease risk, genes associated with both recessive childhood-onset disease and dominant adulthood-onset disease pose a dilemma. In such cases, group considerations include the aforementioned ethical principles, but consensus on how to proceed is often guided more strongly by protocol limitations. We present case examples of returning heterozygous variants in GBA, MUTYH, and BRCA2. Our encountered ethical challenges highlight the need for and limitations of an evaluative framework for variant assessment in GS in the neonatal period.

Anticipation in Swedish Lynch syndrome families

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Lynch syndrome (LS) is one of the most prevalent hereditary colorectal cancer syndromes, representing between 2-5 % of all cases of colorectal cancer. LS is mainly caused by mutations in the DNA mismatch repair genes MLH1, MSH2, MSH6 or PMS2. Patients primarily have an increased risk of early onset colorectal and endometrial cancer, as well as an increased risk of tumors e.g in the small bowel, upper urinary tract and brain. Age at cancer onset may vary between family members and a decreasing age at onset in successive generations, called anticipation, has been reported. In addition, anticipation has been suggested in other heritable cancers such as familial melanoma, pancreatic and breast cancer. The purpose of this study is to determine whether anticipation can be shown in Swedish LS families referred to the regional departments of clinical genetics in Lund, Stockholm, Linköping, Uppsala and Umeå between the years 1990-2013. Our long-term goal is to enable better prediction of age at onset in different family members, which is highly dependent on if anticipation is part of the clinical picture in LS.

In total, 239 families (96 MLH1, 90 MSH2 including one family with an EPCAM-MSH2 deletion, 39 MSH6, 12 PMS2, and 2 MLH1+PMS2 families) comprising 1003 individuals with available follow-up information that could be included in the study. Using a normal random effects model (NREM) we estimate a 2.1 year decrease in age at onset per generation. An alternative analysis using a mixed-effects Cox proportional hazards model (COX-R) estimates a hazard ratio of exp(0.171), or about 1.19 for age at onset between consecutive generations. Gene-specific anticipation effects are significant for MSH2 (2.5 years/generation for NREM and hazard ratio of 1.33 for COX-R) and PMS2 (7.3 years/generation and hazard ratio of 1.85) while the evidence is equivocal for MLH1 and MSH6. This indicates that gene-specific dynamics influence age at cancer onset and encourages further studies of the mechanism behind anticipation and the complex relationship between genotype and phenotype, to facilitate the management of families with LS.

A Case Study: Reflecting on ABC vs St Georges; a case of advanced genetic counselling skills facilitating family communication.

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The legal case of ABC v St George's Healthcare NHS considered whether the team responsible for the care of a patient with Huntington's disease (HD) had a duty to disclose genetic risk information to adult children without their patient's consent. The claim was initially struck out because there was "no reasonably arguable duty of care" owed to the claimant. However, a recent appeal court judgment has allowed appeal and remit of the case to trial. This raises the ethical and legal issue of our duty to disclose genetic information to the family.

Clinical Case Study: A 70 year old man was referred by his GP with a family history of HD. The main concern was that the patient now appeared to be symptomatic of HD. There had been no disclosure and no intention of disclosure of the familial risk. A granddaughter was currently pregnant. At the appointment it was revealed that a decision not to determine his own genetic status was made at the time of his mother's diagnosis. A joint decision was also made by the family not to inform future generations about the familial risk. The motivation was to protect the family from the burden of this knowledge. The genetic counsellor reflected with respect and unconditional positive regard on the patient's prior decision not to disclose. Advanced empathy and immediacy skills established rapport and a trusting therapeutic relationship. Challenging skills were used for consideration of the ethical implications and relevance of this information for the family today. Barriers to disclosure were identified and potential solutions offered. The couple were given time to adjust to the clinical discussion before re-contact was made. The couple disclosed the family history to their children shortly after their appointment.

The ABC vs St Georges case has prompted genetics services to reflect on current practice. This case explores the ethical and legal issues surrounding genetic counselling and the principles that underpin practice. In doing so it highlights the role of the genetic counsellor as a facilitator to family communication. The counselling skills employed empowered the family to communicate offering an ideal resolution without having to address our duty to disclose genetic information to the family.

Using Solution Focused Approaches to Enhance Genetic Counselling: A Reflection on My First Steps

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Keen to enhance my skills as a genetic counsellor, I undertook a series of short training courses in Solution Focused (SF) practice, also known as Brief Therapy. As the name suggests, SF work encourages clients to shift their attention from their problem/s and to consider their own solutions. This is achieved by asking the client a series of questions to which the facilitator does not have the answers. SF sessions generally consist of some introductory 'problem free' talk, establishing the clients best hopes from the session, exploring in detail what the future would be like if the best hopes were realised, then using a simple linear scale (e.g. 1-10) to establish ways in which success is already being achieved. The client is then encouraged to describe how progress along that scale would be identified and the difference this would make to their situation.

I found the experience of learning about SF work to be refreshing and energising but when I considered the application of this to genetic counselling I had some reservations: genetic counsellors have clear objectives from consultations such as addressing the reason for referral, creating a family tree, carrying out a risk assessment, discussing inheritance and offering testing or information as necessary. Using an exclusively SF approach did not seem appropriate for most genetic counselling cases. I will describe a case where I did use a SF approach; in this case the patient had already been given a genetic diagnosis and understood inheritance patterns, sources of support and future medical issues. The referral was for 'more support'. I will also discuss other ways SF approaches can be integrated into genetic counselling, such as in telephone consultations and mentoring a colleague.

The South West Thames Regional Genetic Service Hereditary Cancer Carrier clinic: the experience from the perspective of the patients and health professionals involved

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We established a hereditary carrier clinic for any individual who is a carrier of an inherited cancer predisposition condition (BRCA1/2 carrier, Lynch syndrome, familial adenomatous polyposis, MUTYH-associated polyposis, Li Fraumeni syndrome, Peutz Jeghrs Syndrome Juvenile polyposis Syndrome) in January 2015. This clinic is run by a genetic counsellor (GC) and a breast physician. The role of the GC in this clinic is to provide ongoing psychosocial support to carriers while they adjust to their result and help facilitate decision-making around management options. The role of the breast physician is to provide detailed information about the different risk-reducing breast surgery and reconstruction options in addition to educating about breast self-examination.

In our department all carriers are seen by the GC or Clinical Geneticist that has taken them through genetic testing for a detailed discussion about the implications of their result. Only carriers who would like/need additional support or would like to consider risk-reducing surgery are seen in the Hereditary Cancer Carrier Clinic at St George's Hospital or the specific BRCA carrier clinic at The Royal Surrey County Hospital.

We wanted to find out the patients' views about the Hereditary Cancer Carrier Clinic at St George's Hospital. In particular we wanted to know how helpful they found their appointment and their satisfaction with the clinic. In September we will be asking all the carriers that have been seen in the clinic since 2015 to complete a patient satisfaction survey via monkey survey. We will report the findings from this survey.

From my perspective as the GC running this clinic, the consultations have required me to focus on the counselling and psychosocial educator aspects of my role. I have found it invaluable to be able to draw upon the counselling skills and tools that I have gained through various counselling courses (Alan Phillips) I have undertaken. I will discuss some of the tools and skills I have found particularly helpful.

Evaluation of a centralised national telephone genetic counselling service that facilitates BRCA1/2 testing for women with relapsed high-grade serous ovarian cancer

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Women with recurrent high-grade serous ovarian cancer (HGSOC) who have a germline BRCA1/2 mutation benefit from treatment with poly (ADP-ribose) polymerase (PARP) inhibitors. In order to guide treatment decisions prompt access to genetic testing is essential for these women. However, the rates of genetic testing for such women in Australia remain low despite recent changes to BRCA1/2 genetic testing criteria and efforts to mainstream BRCA1/2 testing. In an attempt to address barriers to genetic testing, in 2016 a national centralised telephone genetic counselling service was established in the Parkville Familial Cancer Centre at Peter MacCallum Cancer Centre. Women with recurrent HGSOC are referred by their medical oncologist. Pre and post test genetic counselling, including facilitation of BRCA1/2 testing and delivery of test results, occurs via telephone with a genetic counsellor.

A mixed-methods evaluation of the telephone genetic counselling service commenced in August 2016. The aim of the evaluation is to examine the acceptability and feasibility of the telephone genetic counselling to facilitate BRCA1/2 testing in women with recurrent HGSOC. The evaluation consists of three stages: 1) a survey of women who received the telephone genetic counselling service; 2) interviews with the referring medical oncologists; and 3) a measurement of the cost effectiveness of the telephone genetic counselling compared with face-to-face genetic counselling. This presentation relates to the first stage only: women's experiences of receiving telephone genetic counselling to facilitate BRCA1/2 testing.

Reproductive decision making process, medical concerns and special needs of preimplantation genetic diagnosis (PGD) users:

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PGD technology can increasingly detect genetic disorders and traits. This procedure may eliminate some of the obstacles related to conservative options of prenatal diagnosis, but can raise personal, social and moral questions. The psychosocial aspects of the technology have been discussed in the genetics, bioethics and sociological communities and were evaluated among various population groups worldwide. However, only scant empirical data focused on PGD users' expectations, concerns, decision-making and experiences, involved in the process.
In order to evaluate PGD's implications regarding perceptions, attitudes and reproductive decision making process involved in the procedure and to assess their experiences and future needs, a combined methodology was used: Qualitative analysis of semi-structured in-depth face-to-face interviews with 43 PGD users for medical reasons (carriers of autosomal recessive, dominant and X-linked disorders, and HLA-matching), representing variety of population's sub-groups. On the basis of the interviews, a detailed closed web-based questionnaire was developed. Univariate and multivariate adjustment was performed on data obtained from 155 subjects involved in various stages of PGD procedure.
PGD is considered a preferable diagnostic procedure for 139 (95%) subjects. Nevertheless, 71 (47 %) reported a complex decision-making process. Perceived advantages are: assurance of the embryo's unaffected status from the beginning of the pregnancy, thus avoiding the need for pregnancy termination and invasive prenatal tests. Perceived disadvantages focused on the medical actions involved, and the delay in time between the first counselling and the PGD procedure itself. Other future needs included improving the communication with medical staff and implementing emotional support. The study indicates special needs of respondents from groups with distinct genetic and socioeconomic backgrounds.
PGD users are coping with both genetic disease and load of the PGD procedure. Taking into consideration their opinion is important since it reflects the gains and burdens of these procedures alongside the demand for future optional services.
This interdisciplinary qualitative and quantitative analysis of a large sample of PGD users can form a basis for development of counselling and guidance programs of future PGD users and will also help foster a public debate concerning medical, ethical, sociological and economic aspects of the technology.

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