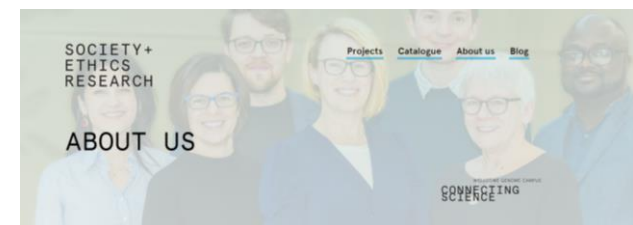


Genomics, patients and families; the practice of genetic counselling

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Aims

- Show the impact of genetic conditions and risk information has on families.
- Recognise the important role of health professionals in supporting families' adaptation and coping with genetic conditions and risk information.
- Discuss examples of research

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

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

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

- More standardisation
- More information
- More complexity
- More change
- More uncertainty



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

(1) understand the medical facts of the disorder;

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

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

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

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

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

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

http://www.eurogentest.org/professionals/info/public/unit3/final_recommendations_genetic_counselling.xhtml

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- What is the impact of genetic information
 - Individuals, families, health systems, society
- How do we support individuals and families
 - With thanks to Professor Alison Metcalfe

What do we know from research about outcomes from genetic counselling

Need for an evidence base

commonly measured

- knowledge,
- anxiety or distress,
- satisfaction,
- perceived risk,
- genetic testing (intentions or receipt),
- health behaviours, and decisional conflict.

Genetic counselling improves patient knowledge, risk perception and autonomy in decision-making, decreases stigma experienced and generates high patient satisfaction

Communicating genetic information in/for families is difficult

- Systematic review in 2012-families with inherited genetic conditions
 - Disclosure of genetic information within families is a highly complex and affective process often resulting in delayed disclosure.
 - This can lead to family tensions generated by misunderstandings, blame and secrecy.
 - Children and young people want their parents to engage in open and honest discussions about the genetic condition
 - Rowland and Metcalfe IJNS 2012

Five key barriers for parents:

1. Emotional

2. Communication skills

3. Care related

4. Information and support

5. Family relationships



Parents' and children's communication about genetic risk: a qualitative study, learning from families' experiences

Alison Metcalfe,
Gill Plumridge,
Jane Coad,
Andrew Shanks
& Paramjit Gill

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

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

Parents and children's experiences revealed:

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

- Genetic science is having a socio-psychological effect on individuals and their families.
- Patients and families need time to assimilate and cope with 'new risk' information.
- Many families need support to make sense of and use the genetic information - and this will change across the family lifespan with additions and losses.
- Developments in genetics exposes the flaws in service provision about the 'care of the family'.
- Better educational provision for health professionals caring for patients across the lifespan about the use of genomics in clinical practice.
- Family Systems Theory says 'good communication is essential for families to cope and adapt to living with difficult or adverse situations.'

RESEARCH



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

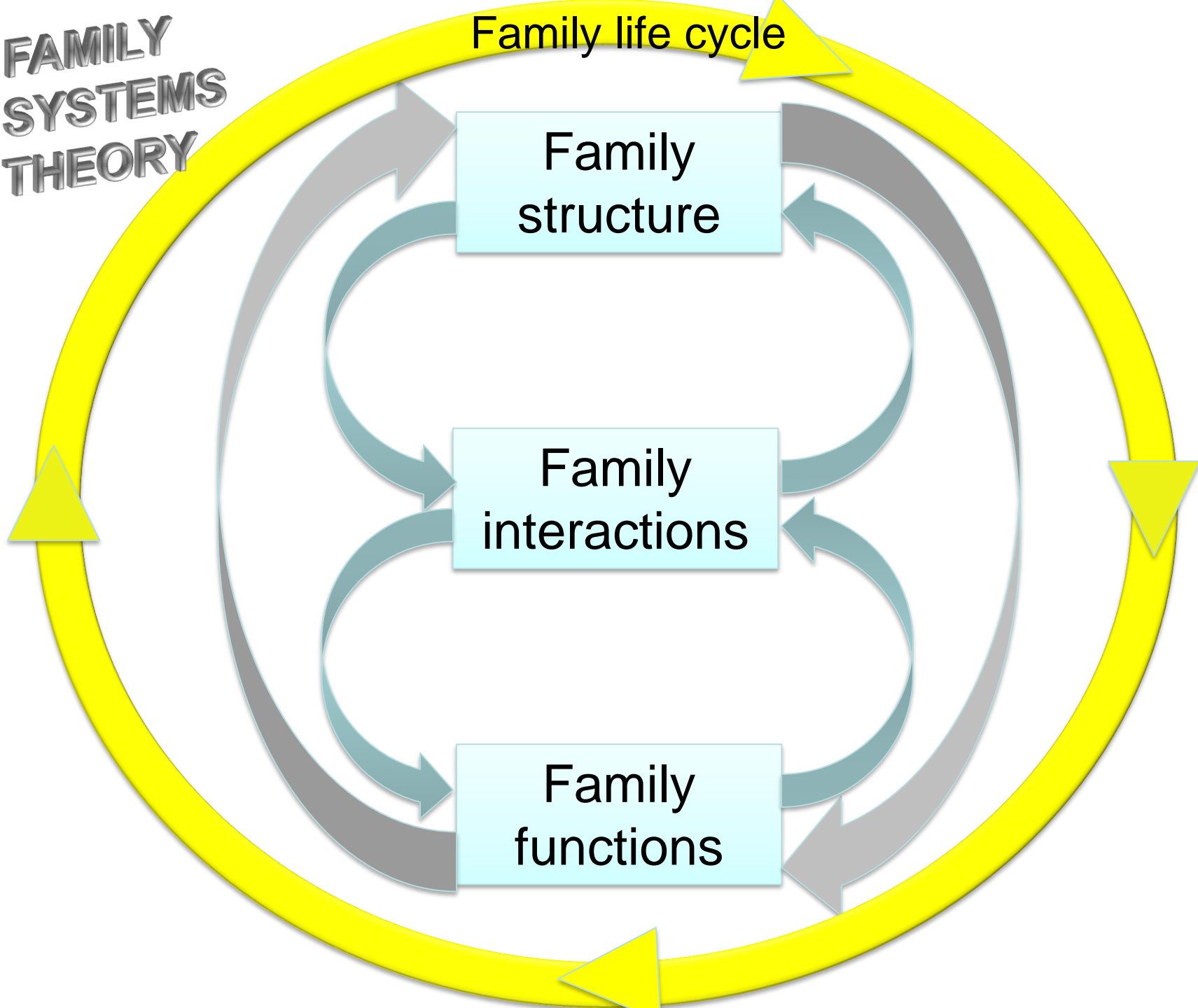
**FAMILY
SYSTEMS
THEORY**

Family life cycle

Family
structure

Family
interactions

Family
functions



Family Transitions and Neuromuscular Disease: Implications for Practice and Research

Dr. Veronika Waldboth, PhD, RN; Prof Dr. Christine Patch, PhD, GCRB, RN;
Prof Dr. Romy Mahrer-Imhof, PhD, RN; Prof Dr. Alison Metcalfe, PhD, RN

- Effects on families:
increasing caregiving needs,
economic,
psychosocial and
emotional hardships
- Stress during life
cycle transitions;
e.g. transition into
adulthood



(chreisgleis.ch, retrieved 01.06.15)

Methods

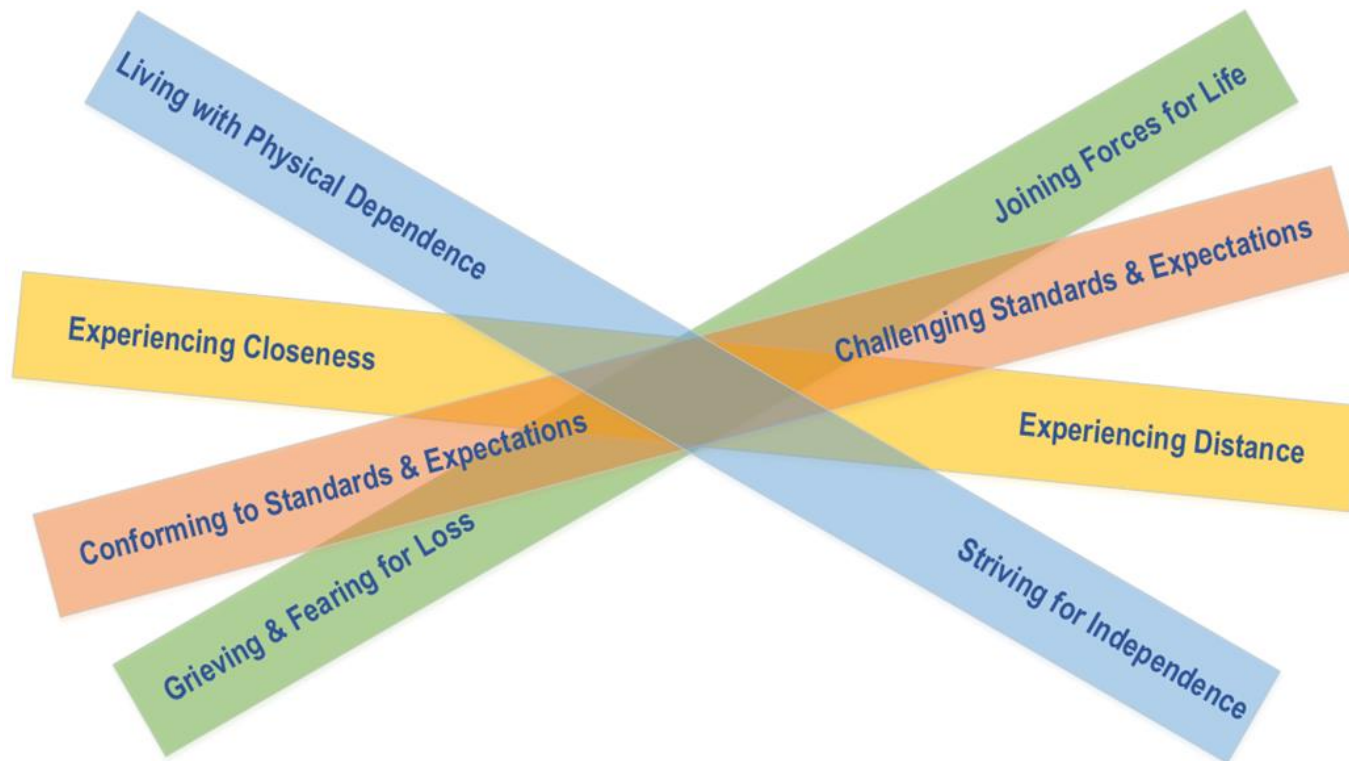


- Qualitative study of the families' transition experience
- Theoretical framework: family systems theory
- Study design: Constructivist Grounded Theory
- Setting: German-speaking Swiss cantons
- Data collection: One-on-one interviews with affected young people (14-30 years) and their families
- Data analysis: coding, memo writing, theoretical sampling, constant comparison, theory development

(Charmaz 2014, Rolland & Williams 2005)

Findings - Theory

«Living in a field of tension between
development and degeneration»



Findings - Functional Domain



Living with physical dependence

...and striving for independence

- Adapting to growth and sexual maturation
- Assessing the situation
- Negotiating roles and distributing tasks



“I tell him [affected person], what you can do by yourself, you do by yourself. I am neither your maid, nor your nurse or anything else. Um, what you can do, you do. And where you need help, I am very happy to help you.” (Parent, 3:187)

Findings - Relational Domain



Finding balance between closeness ...and distance

- Being involved in private matters and family boundaries
- Balancing between freedom and protection
- Effects of distance and closeness

“Yes just simply because in addition to being disabled, he was also going through normal puberty and totally annoyed me and um, was kind of not able to distance himself, like it would have been healthy for his age, because at the same time he simply was totally dependent on me. And that was sometimes really a bit much.” (Parent, 30:25)

Findings - Social Domain

Conforming and ...challenging social standards and expectations

- Being responsible for each other
- Being different is normal
- Having a social life



“I notice that from the outside a person sees, she is simply sitting, she has a tube into her throat and that is somehow the picture of the disability where distance between me and others is created. The moment they let themselves get to know me, then it changes quickly, then people also see what’s behind the disability, but the first contact is always difficult.” (Affected young person, 14:68)

Findings - Emotional Domain



Grieving for loss and a future ...joining forces for life

“You cannot really even not be sad” (Sibling, 7:56).

- Understanding the consequences
- Experiencing recurring health crises
- Coping and adaptation
- Being full of hope and joining forces for life

Implications for Practice



- **Family centred approach to care**
- **Focus on illness and development-related challenges**
- **Assessment and monitoring of the life situation**
 - Multigenerational assessment (genograms)
 - Meaning making, coping and adaptation
 - Resources and particular vulnerability
 - Social and cultural expectations and beliefs
 - Information and caregiving expectations
 - Family traditions e.g. role transcripts



Implications for Practice

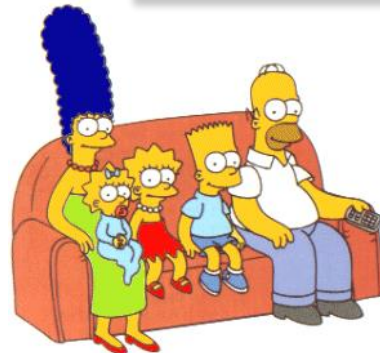
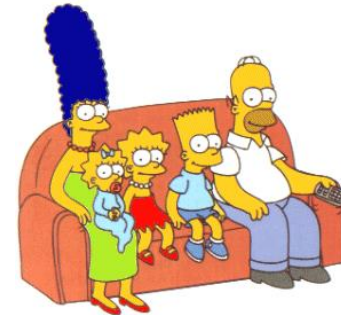
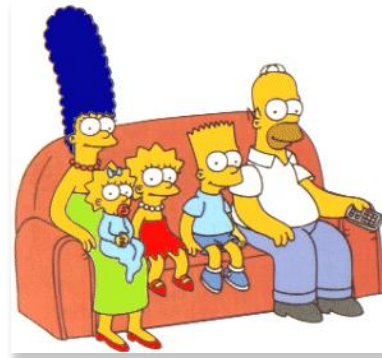
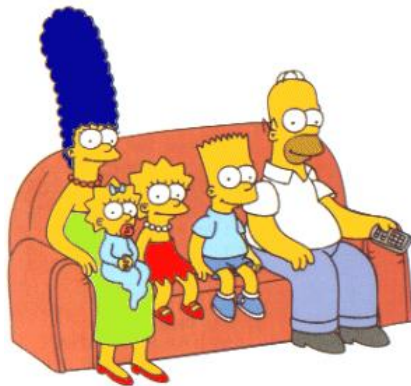


- **Multimodal, psycho-educative and relationship-oriented family interventions**
 - Family education, support and skill building
 - Family communication, problem solving and conflict management
 - Sexual maturation, boundaries and integrity
 - Management of stress and role overload
 - Facilitating coping and adaptation
 - Supporting the grieving process

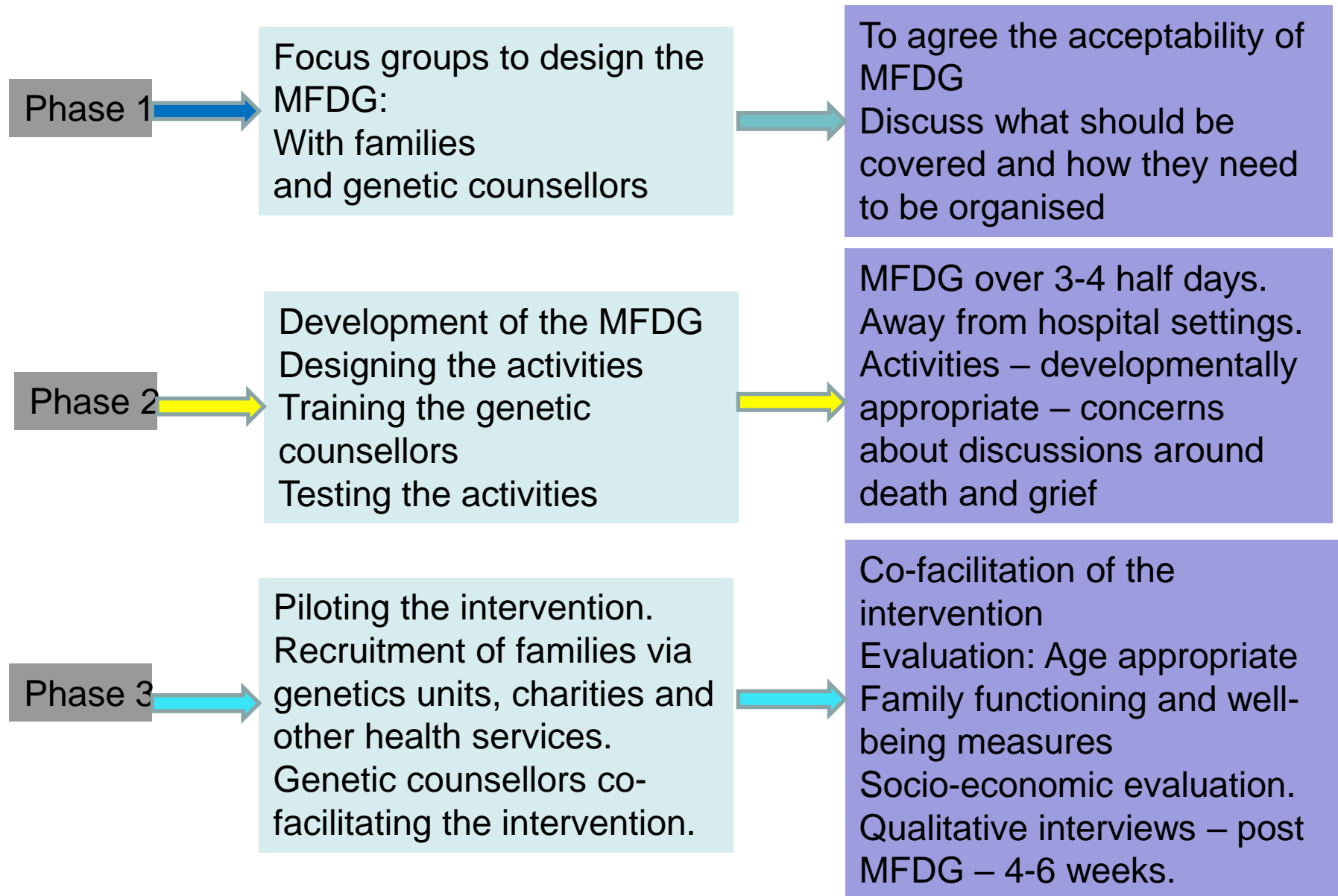


Co-design of family centered interventions

Introducing Multi-Family Discussion Groups (MFDG)



Overview of the MFDG intervention's design



Findings

- Focus groups with a total of 11 families (self selected) represented – different genetic conditions – mainly Autosomal Dominant.
- Families mainly UK British but not all.
- 6 genetic counsellors took part in focus groups

Findings from the Focus Groups

2 key themes emerged showing

i) the need for the intervention

ii) the intervention's design:

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

- Therapeutic value of focus groups!

Developing Pilot MFDG with Genetic Counsellors Co-Facilitating.

Genetic counsellors trained over 9 months – a range of practice sessions held.

The full MFDG pilot ran with 6 families, mixed age ranges including grandparents.

It focused on a range of MFDG activities run over 2 days to:

- Facilitate communication between parents and children.
- Increase parents confidence in discussing difficult information and managing their own and children's emotions.
- Create a wider focus on family life – not just the genetic condition.
- Reduce parents' feelings with regard to stigma and guilt.
- Enable sharing and learning from other families.

Activities included:

- Parents survival kits
- Tree of life.
- Family shields.
- Family sculpts.
- News interviews.
- Goldfish bowl
- Family therapy techniques – therapeutic questions.



Outcomes

MFDG evaluated very highly 4-6 weeks post intervention:

- After some uncertainty genetic counsellors saw large benefits for families.
- Parents, children and young people reported better family life, more open discussions, reduced feelings of stigma, confidence in dealing with difficult conversations.
- Families did not like completing many of the standard family functioning measures – too ‘pathological’ in their focus.
- Volunteers for ‘Graduate Families’ scheme.



Discussion and Conclusion

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

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

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

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

Objectives:

- Train and support researchers to work with patients, families and healthcare staff (Manchester; Prague) to deliver the EBCD method to co-design interventions or services to support the sharing of genomic sequencing information.
- Perform a process evaluation of the EBCD process and the implementation of the intervention or service using interviews and surveys (translated into member countries languages) in the initial two participating ERNs.
- Publish the EBCD method used within the initial two ERNs in order to provide the opportunity for the other 22 ERNs to adopt the findings or follow a similar process to design their own intervention or services.



Summary

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



Genetic Counsellors could play a vital role in:

- Facilitating families coping with and adapting to the genetic condition.
- Facilitating family communication about genetic risk – promoting cohesion and relationship management.
- Assisting parents in providing the right level of information for their children.
- Promoting family cohesion and resilience
- Working with other health professionals to do all of the above

If we don't then genetics could threaten families rather than give them the health benefits it promises.



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