Communication of information about genetic risks: putting families at the centre

ABSTRACT

Genetic information is a family affair. With the expansion of genomic technologies, many new causal genes and variants have been established and the potential for molecular diagnoses increased, with implications not only for patients but also their relatives. The need for genetic counselling and intrafamilial circulation of information on genetic risks grew accordingly. Also, the amount and, particularly, the complexity of the information to convey multiplied. Sharing information about genetic risks with family members, however, has never been an easy matter and often becomes a source of personal and familial conflicts and distress. Ethical requisites generally prevent healthcare professionals from directly contacting their consultands' relatives (affected or still at-risk), who often feel unsupported throughout that process.

We discuss here the communication of genetic risks to family members. We first consider genomic testing as a basis for family-centred health care, as opposed to a predominant focus on the individual. We reviewed the literature on sharing genetic risk information with family members, and the associated ethical issues for professionals. Some clinical cases are presented and discussed, and key issues for meeting the needs of individuals and families are addressed. We argue that genetic information is inextricably linked to the family, and that communicating about genetic risks is a process grounded within the broader milieu of family relationships and functioning. We conclude for the need for a more family-centred approach and interventions that can promote sensitive attitudes to the provision of genetic information to and within the family, as well as its inclusion in educational and training programmes for genetic healthcare professionals.

KEYWORDS: genetic information, genetic counselling, genetic testing, genetic risk, genomics, family communication, family-centred care.

INTRODUCTION

Genomic technologies offer a great potential for improved diagnosis, treatment and prevention for a growing number of genetic diseases. As genome-based testing (multigene panels, whole-exome or whole-genome sequencing) becomes routine, the number of persons known to be affected or at-risk for inherited conditions increases. Genetic testing and genetic information (Sequeiros et al, 2012) have implications not just for the individual, but also their relatives. This often becomes a challenge, as families may feel unsure about that information, and how, when and whom to tell (Gaff & Byllund, 2010). Genetics health professionals (GHPs) and services are thus confronted with the need not just to provide care to the individuals tested, but also to help families understand and cope with genetic information (McDaniel, Rolland, Feetham, & Miller, 2006; Rolland, 1999).

Whilst many scholars and clinicians have long claimed the importance of paying attention to family relationships in the dissemination of genetic information (Feetham, 1999; McDaniel, 2005; Peters, Djurdjinovic & Baker, 1999; Rolland, 2006a), existing literature mostly focuses on what prompts (or hinders) individual patients to disclose that information to their relatives (McClellan et al., 2013). This fails to acknowledge the intricacies of the communicative processes that operate within the family and how they are entwined with other domains of family functioning. Additionally, genetic health care is mainly designed to address individuals, and more family-centred approaches are still scarcely embedded into routine care, which may leave families ill-supported while they seek to integrate genetic information into their lives.

In this article, we discuss the communication of genetic risks to family members. First, we set genetic knowledge as a basis for family-centred health care, as opposed to its predominant focus on the individual. Then, we present a systemic lens for understanding genetic diseases, based mostly on the Family Systems Genetic Illness (FSGI) model

(Rolland & Williams, 2005). Next, we discuss the sharing of genetic risk information within the family, including instructive clinical cases, and ethical dilemmas faced by GHPs. We address some of the key issues for meeting the needs of individuals and families; in doing so, we frame genetic information as a family affair and present communication about genetic risks as a process that encompasses and impacts the family all along. Finally, we highlight the need for family-centred care and interventions designed to help families managing genetic risk information, and a call is made for an appreciation of the family as the biopsychosocial unit of care.

GENOMICS: ORIENTING CARE TO THE INDIVIDUAL OR TO THE FAMILY?

Genetic information differs from most other disease-related information because of its familial character. Disclosure of genetic information to relatives gives them the opportunity to make informed choices regarding treatment, prevention or changes in life-styles. Depending on the type of disease (mode of inheritance and penetrance, age-at-onset, clinical severity and psychosocial burden, treatments available, and potential for early detection and follow-up) and its associated level of uncertainty, this may pose a difficult challenge. For example, knowing that an individual carries a mutation in BRCA1/2 (linked mainly to breast, ovarian and prostate cancer) will be relevant for biological relatives, who may or may not wish to consider genetic testing and appropriate preventive options, as well as reproductive and other life-planning choices. This intrafamilial communication, however, is not always straightforward, and consultands (whether already affected, presymptomatic carriers or still at-risk) often feel unsupported in managing it (Gaff et al., 2007).

Traditionally, there has been a great emphasis on individualized healthcare. The burgeoning *precision medicine* movement insinuates that genomics will tailor prediction,

diagnosis and treatment to every person (Tutton, 2012). In stark contrast with this dominating narrative, the converse is that genome-based tests have far greater implications for family relations than does directed, conventional genetic testing. Furthermore, genomic testing increases the need to involve family members to clarify incidental findings and inconclusive test results (newly-discovered variants and variants of unknown significance) (Hallowell, Hall, Alberg & Zimmern, 2015). This entails processing information and involvement across and within generations of a family. GHPs work mostly with individuals or nuclear families, and often have limited access to the wider family network. Help offered by these professionals is usually information-based and focuses predominantly on the individual, rather than the family unit (Mendes, Paneque, Sousa, Clarke & Sequeiros, 2016). GPHs also indicate they do not always address issues of family communication in their practice (Forrest, Delatycki, Curnow, Skene, & Aitken, 2010). Additionally, privacy requirements prevent healthcare professionals from directly informing their patient's relatives (Parker, 2001). This can lead patients to feel frustrated, as they are given indications as to which relatives to inform, but often no support to proceed with doing so. How will families respond as more information is generated via genomic analysis?

A family view on genetic illnesses

The experience of a genetic illness in the family has been well described in the family-systems literature (Brouwer-DudokdeWitt, Savenije, Zoeteweij, Maat-Kiewit & Tibben, 2002; Kessler & Bloch, 1989; Sobel & Cowan, 2003). A conceptual framework setting out likely psychosocial demands of families with various genetic conditions was provided by Street & Soldan (1998). Based on Rolland's Family Systems Illness (FSI) model (Rolland, 1984), they proposed to expand the 'illness stages' to include the 'pre-illness phase'. Subsequently, Rolland & Williams (2005) extended the earlier model to include genetic

disorders (the FSGI model), especially the presymptomatic phases of genetic diseases, aiming to understand their impact on the coping processes of family members. Coping with awareness of a possible genetic risk, consideration and active decision-making regarding testing, and the incorporation of genetics knowledge into personal and family life are important challenges.

Developments in genetics increasingly blur the boundaries between health and illness, creating “patients in waiting” (Timmermans & Buchbinder, 2010). Living with uncertainty, the quantity of medical information presented, sharing risks with relatives and the psychosocial impact of those choices are other fundamental issues outlined in the FSGI model (Rolland & Williams, 2005). This includes a psychosocial typology, based on four biological variables: likelihood of developing the genetic condition (penetrance), clinical severity, age of onset, and availability of treatment or preventive measures. The FSGI model acknowledges the interface between temporal phases of a genetic condition through the individual and family life-cycle and the influence of anticipatory loss, i.e., living with possible, probable or inevitable future losses, after the diagnosis of a hereditary disease (Rolland, 1990; 2006b). Intergenerational effects have a greater impact with dominantly inherited or sex-linked disorders. In sum, this model suggests a framework to organize the complexity of genetics into a common meta-language, to help health practitioners viewing genetic diseases longitudinally, as an ongoing process, with transition points and changing demands.

FROM THE CLINIC TO THE FAMILY: COMMUNICATING GENETIC RISK INFORMATION TO FAMILY MEMBERS

Family issues that may arise from the communication of genetic information are commonly discussed in genetic counselling, including identification of at-risk relatives. Test results

are often given in person to the consultand. Traditionally, professionals rely on patients (or, if minors, their parents) to pass on that information to other family members, and reinforce the importance of this. However, consultands may feel unsure about the responsibility of professionals; some believe that professionals should be the ones to inform relatives rather than themselves (Mesters, Ausems, Eichhorn, & Vasen, 2005). The explicit refusal to share relevant information with relatives is rare (Clarke et al., 2005), but failure of communication may occur for many other reasons. For those requiring support or showing difficulties in this process, professionals may use multiple strategies, particularly psycho-educational guidance and written materials to be given to at-risk relatives (Mendes et al., 2016).

Genetics healthcare professionals' ethical dilemmas

Ethical and professional guidelines advocate that professionals should not contact family members directly (Forrest, Delatycki, Skene & Aitken, 2007). Guidelines also state that professionals should encourage consultands to transmit risk information to relatives and support them throughout the communication process, although it is not clear how this should be done. Possible dilemmas involve the balance of the patient's autonomy and right to privacy with potential for harm to relatives. Professionals' responsibility and proactivity to ensure awareness of relatives about their risks have long been debated and still need further clarification (Clarke, 1997; Dheensa, Fenwick, Shkedi-Rafid, Crawford, & Lucassen, 2016). Some countries (e.g., Australia and France) bind patients to inform family members and provide professionals the right to override their patients' confidentiality.

Traditionally, biomedical models see patients as the "owners" of their personal medical information; however, genetic information is shared with blood relatives and thus

challenges the meaning of these assumptions (Widdows, 2013). Relational approaches to autonomy emphasize that it may be enhanced through solidarity, engagement and social embeddedness (Dove et al., 2017). For example, the joint account model conceptualizes clinical information as personal, but genetic information as "belonging" to all pertinent relatives; this way, disclosure of information within this circle would not represent a breach of confidentiality (Dheensa, Fenwick & Lucassen, 2016). Genetic information, however, is also viewed as ultimately private, and there are difficulties in clearly defining which relatives have equal right-of-access to it (Clarke, 2007), and whether and how they may preserve their right not-to-know (Chadwick, 2009).

Sharing genetic risk information with family members

Empirical research shows that, although consultands of genetics services (or their parents) are generally willing to transmit relevant information to their relatives, sharing genetic risk information with family members is sometimes problematic (Seymour Addington-Hall, Lucassen & Foster, 2010). Estimates suggest that only 15-20% at-risk relatives become aware of important information, and test uptake by relatives is generally low (Fehniger, Lin, Beattie, Joseph & Kaplan, 2013). Also, Daly and colleagues (2016) reported that 82% of the first-degree relatives to whom probands (the first in the family to get tested) had reported their test results for BRCA1/2 had correctly understood the results; nevertheless, their intention to pursue genetic testing was substantially lower. This, however, is just a proxy measure of family communication and depends also of other variables. Qualitative studies show that this transmission, itself, may be withheld, delayed or incomplete. Indeed, patients may have a poor understanding of it, or they may anticipate their family members would not want to know or are unable to understand or cope with it (Featherstone, Atkinson, Bharadwaj, & Clarke, 2006).

Family dynamics, including patterns of rules and boundaries, influence preferences about sharing and knowing health information within the family. This is impacted by the range of biological variables of the disease that are important in a clinical evaluation (Rolland & Williams, 2005). While some relatives act as proactive gatherers and disseminators of genetic information, others block the communication process (Kohely et al., 2009).

Two anonymised examples, from real cases, are presented next to illustrate some of the relevant issues in sharing genetic information in families.

Rita's guilt: gathering extended family members for predictive testing

In a family with Huntington disease (HD), Rita was the youngest and the only of three sisters not carrying an expanded HD gene. HD is a progressive, fatal neurodegenerative condition, with onset most often by age 35-60 years, with a *a apriori* 50% risk to sibs and offspring. It leads to involuntary movements (chorea), motor impairment, behavioural symptoms and cognitive decline. Her father had had the disease, as well as several of her uncles and aunts. Rita was 49 years-old at the time of presymptomatic testing; she has three adult sons and had divorced when they were young children. She had never kept a long-standing relationship thereafter and was living with her younger son. In a very enmeshed family, she structured her life as a caregiver for both her affected sisters, until they died. During genetic counselling, it became clear she was having difficulties dealing with her non-carrier result and became severely depressed. She also developed a very strong sense of responsibility towards her seven nephews/nieces (all in their mid-to-late twenties), and was very proactive, even forceful, in trying to persuade and bring every single one of them for genetic testing.

Genetic information has the potential for altering family relationships. As in this case, altruistic intentions and behaviours may emerge, as Rita experienced "well-sibling", survivor guilt in relation to her sisters. This first became apparent as she assumed the role of caregiver for them, and, later on, when she made all efforts to bring her nephews/nieces

for presymptomatic testing. The testing experience can be most intense for support persons and may extend to subsequent caregiving responsibilities (Williams et al., 2000). Rita had difficulties finding a balance between relief (as a non-carrier) and the concern for her at-risk relatives, as if awareness of her risk-free future prompted a ruptured loyalty towards her family legacy of HD. Additionally, the pressure she exerted on her nephews/nieces potentially disturbed their own autonomous decision-making process for genetic testing and thus increased the risk for family conflicts. This underscores the importance of providing long-time engagement with families (including the extended family) and also with those who are non-carriers.

Communication is one aspect of family processes that includes systems of belief and family organization, transgenerational patterns of coping and life-cycle challenges (McDaniel et al., 2006). These processes are embedded in the everyday reality of families, and the communicative style of the family reveals how family members manage closeness and distance while they deal with difficult topics, such as genetic risk, illness and healthcare (Atkinson, Featherstone, & Gregory, 2013; Featherstone et al., 2006; Forrest et al., 2003), including genetic testing.

Cultural factors also shape family communication. Ethnicity and religion may lead some families and communities to feel more reluctant to generate and disclose risk information in the family, because of its potential negative impact on *honour* and marriage prospects (Shaw & Hurst, 2009). These decisions are also affected by concerns regarding privacy, stigmatization and discrimination (Mendes, Sousa, Sequeiros, & Clarke, 2017). Open communication allows family members to develop a shared understanding and support one another, becoming more likely to cope and adapt well to living with the genetic condition or its risk and establishing more resilient relations (Hoskins, Roy, Peters, Loud & Greene, 2008). Open communication tends to occur more frequently among women, first-

degree relatives or spouses, while those under 18 years are less likely to be given information (Oliveira, Mendes, & Sousa, 2017; Van Oostrom et al., 2007). Approximately 40% of parents do not talk to their children about their family's condition, although most would like to be able to do so (Metcalfe, Coad, Plumridge, Gill & Farndon, 2008). Communication styles vary in a continuum ranging from disengaged to enmeshed patterns of communication, and can evolve over time as family members go through their life cycle (Brouwer-DudokdeWitt et al., 2002; McDaniel et al., 2006). For example, parents may decide not to inform their children about the genetic condition, as they lack confidence in deciding when and how to talk; they may change their mind as offspring approach adulthood and information can have implications for life planning, such as selecting partners and procreation.

Eugénio's burden with his ataxia test results

Eugénio was aged 48 when his pre-symptomatic test for spinocerebellar ataxia type 2 (SCA2) showed him to carry his family's disease mutation. As this is a fully-penetrant single-gene disorder, he is certain to develop it sometime in the future, but also that his 16 year-old son has a 50% risk of carrying it too. SCA2 is a late-onset, incurable neurological disease, progressing to severe motor impairment. Eugénio has two older brothers, both non-carriers. He has been aware of his risk since early adulthood, when his uncles became affected. He then decided to live a carefree life, without the burden of knowing his genetic status. After his mother became affected a few years ago, he began to actively consider being tested. As the bread-winner in a middle-class family, Eugénio has been running the family business for several years and fears having to stop this when symptoms become severe. He shared the test result with his wife, brothers and father. His brothers urged him to keep it a secret from their mother, in order to protect her from alarm and blame. Eugénio, himself, is reluctant to inform his son, as he fears the effects this might have on his wellbeing, while his wife thinks they should tell him. This disagreement intensified marital stress, and problems emerged when Eugénio started to show early symptoms of the disease. At this point, the couple was clearly having difficulties adapting to this new stage of their lives together.

This case highlights how the sharing of genetic information can be affected by secrecy in the family system. In this case, Eugénio seem to accept family values around genetic knowledge through alignment with his siblings, though colliding with how his spouse prefers to manage it with their son. This case also illustrates how predictive testing, as well as chronic illness, can impact on the balance of partner relationships. Indeed, couples may have to deal with issues of anticipatory loss, as test results challenge their expectations about the future (Rolland, 1999). This includes rescheduling joint life-planning, as well as optimising the timescale for passing information about risk to their children. The crisis arising at the transition to overt disease is a further burden to the family system. This case underscores the relevance of pre- and post-test family consultations, exploring the implications of secrecy, as well as the right of access to information, and providing ongoing support to the family system.

HOW TO MEET THE NEEDS OF INDIVIDUALS AND FAMILIES?

Genetics services have traditionally emphasised the need to prepare a person for genetic testing and its results, but have placed less emphasis on the secondary impact on other members of the family. Indeed, the reflective process commonly used by professionals when supporting consultands through predictive testing will not always address the communication processes operating within the family. Consistent with this, interventions aimed at facilitating communication to relatives are mainly centred on the need of the patient to disclose information about him/herself, so that relatives may come to know their own risks (Mendes et al., 2016). Information is typically delivered as a single transaction, often in a transmitter-receiver fashion, highlighting what is communicated and with whom. In general, research focuses on the “disclosure” of test results to at-risk relatives and how

this is recalled. These simplistic models of communication regard individuals as equals, who are well placed to absorb what they are told, and treat information as something that can be passed, unaltered, from person to person in the family.

However, disclosing genetic risks is more than the transmission of discrete parcels of 'information'. It goes beyond the consultand's motivations regarding "disclosure" and the actions promoted by professionals to improve dissemination of information. In fact, tension may arise in families when professionals use heavy-handed prompts to encourage it (Carrieri, Farrimond, Kelly & Turnpenny, 2016). Concern for the continuity of family life may lead some to avoid the potential disruption caused by transmission of genetic risk information and, thus, families may choose to act in ways at odds with medical advice (Geelen, Van Hoyweghen & Horstman, 2011).

Using multifamily discussion groups (MFDGs) to support families

Systemic family-centred approaches have a long tradition in assisting families to deal with complex biomedical situations, often more clinically effective and cost-efficient than individually-based care (Weihs et al., 2002; Proulx & Snyder, 2009). Two groups, in Portugal and the UK, have independently begun developing a more systemic approach to family care for those affected by genetic diseases, particularly using psycho-educational MFDGs: these have been established as family-centred interventions for chronic medical illnesses (Steinglass, 1998; Steinglass et al., 2011); pilot MFDGs have been implemented as a way of promoting families' adaptation to cancer genetic risks (Mendes, Chiquelho, Santos & Sousa, 2010; 2015), and also to facilitate family communication about a range of genetic conditions (SPRinG, 2016a). Families participated in creative and experiential exercises about building a sense of identity, belonging and self-esteem. Preliminary evaluations suggest a therapeutical effect; families reportedly find MFDGs highly beneficial

in facilitating bonding and extra-familial networks, and improving psychological wellbeing (SPRinG, 2016a; Mendes et al., 2010; 2015). MFDGs educated family members about the genetic disease and facilitated families talking to and learning from each other about it, while coping with and adapting to its impact.

A UK team also began investigating the possibility of training genetic counsellors and nurses to deliver this intervention (SPRinG, 2016b). This training took longer than anticipated; although they consider their work to be family-centred, they are not trained in family evaluation and interventions. This is a shortcoming in educational programmes for genetic healthcare professionals that should be considered when designing them.

TOWARDS A FAMILY-CENTRED APPROACH?

Communicating about genetic risks to and within the family is a longitudinal process. It challenges traditionally established assumptions about the belonging of medical information and right-of-access to it, and may depend on local legislation and professional guidelines. A systemic, family-centred approach can enhance the understanding of this processes of communication and the care of those involved. This requires more training for GHPs in family systems evaluation and consultation (e.g. family assessment and interpersonal counselling skills), so they may feel more confident in working in the context of families and running family-centred interventions such as MFDGs.

Medical family therapy (MFT) has long provided a suitable framework for family-centred health care (McDaniel, Doherty, & Hepworth, 1992). The inclusion of professionals trained in MFT in genetics services has been suggested (Feetham, 1999; McDaniel, 2005). Its applications may also include the supervision of practitioners and interdisciplinary collaboration, and the training of professionals to bridge specific gaps in the care of

patients and their families. Besides MFDGs, family consultations, couple and family therapy, and individual or group psycho-education are other interventions that might enable genetics services to engage with consultand's strengths and vulnerabilities and key relationships (McDaniel et al., 2006).

Longitudinal family systems consultations, before and after testing, may help identifying core aspects of family functioning with a genetic disease, including processes of communication. Of course, involvement of extended family members should not pre-empt individual autonomy. To include everyone who might be relevant denies the consultand the opportunity to keep any secrets at all, so that the question of disclosing or not an information item never arises, as it will have already been disclosed. A space needs to be designed, within which these considerations can be articulated and weighed.

Careful consideration is needed about who in the family would be relevant for discussions around privacy, secrecy and "responsible" communication about genetic risks. This includes nuclear and extended family members, as well as non-biological relatives and not only those at-risk or with carrier test results (Rolland, 1999). However, GHPs may have only limited access to the wider family network. They need to promote the consultand's adjustment, while gaining insight into processes of communication within families and its potential consequences for the consultands. This exploration should contribute to the consultand to consider his/her options and preferences, and to anticipate management of challenges and concerns underlying genetic/genomic information. Additionally, it could help building a comprehensive plan to support planning communication with family members.

CONCLUSION

We consider the process of communicating information about a genetic disease and associated risks as a family affair. We discussed difficulties families face when sharing information with relatives and acknowledged practical and ethical issues commonly met by GHPs. We then reflected on how current clinical ethics standards may not be completely apt for genetics/genomics information, and questioned efficacy of intra-familial communication. Finally, we introduced MFDGs as an example of systemic-inspired interventions to help families effectively communicating genetic information to relatives, and reflected on the need for family-centred care in genetics healthcare services.

Whereas GHPs have always emphasized the shared nature of genetic information, including preparing consultands to convey that information within the family, far less attention is generally paid to supporting them in that process. Family systems interventions could be seen as a natural approach to be adopted towards family-centred care. This could facilitate handling of genetic information within the family, minding the need to avoid “one-size-fits-all” solutions and to acknowledge the complexity and plurality of family life.

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