Gender and genetic risk: Exploring conceptualisation and interface in the health care context by Sandra Taylor, PhD
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Abstract

Differential responses by men and women to genetic risk information and the availability of genetic tests are increasingly reported. Such apparent differences coincide with differences in broader health and illness contexts such as in mortality and morbidity rates and health behaviours like help seeking, coping and adaptation. As well as biomedical and psychological perspectives, sociological frameworks have been used to critically analyse ‘gender’ and ‘risk’ concepts, separately and intersecting, within the genetics health context. Such analyses have examined the relational and social domains of men and women with genetic risk information, gendered responsibilities, embodiment and gender identity as well as the inherent power dimensions of genetic and reproductive technologies, with their significant implications for women. This paper presents a brief overview of interdisciplinary engagement with concepts relating to gender, health and genetic risk. It concludes that a singular dichotomous construct of gender is unlikely to progress understanding and argues for an integrated and inter-disciplinary analysis in order to maximise the potential benefits of genetic testing technology for consumers, service providers and the broader community. Ongoing investigation of masculinity, femininity and identity discourses relating to genetic risk, particularly as they intersect with reproductive imperatives and life stage, is warranted.

Key words: Gender; genetic risk; genetic testing; theoretical; identity; life stage

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Introduction

There is increasing evidence that men and women who are at risk of inheriting genetic conditions and disorders are engaging differentially with genetic testing for adult-onset conditions that have no sex-based difference in heritability (Taylor 2005:152; Hayden 2003:142). This has been reported in Australia (Taylor 2005:155), Canada (Creighton et al 2003: 462) the United Kingdom (Harper et al 2000:567) and Europe (Tibben et al 1997:33) and in regard to different types of inherited disorders and different types of genetic tests.

This paper aims to examine gender-based differences in the genetic health context from a range of conceptual and disciplinary perspectives. Key concepts of gender, sex, genetic risk and genetic testing will be introduced with a brief critical analysis to reflect the contested nature of these concepts. An overview of the focus of biomedicine, psychology and sociological perspectives relevant to gender-based differences within health and genetic health contexts is undertaken. The paper concludes by discussing the implications of this analysis for clinical genetics services providers, supporting the merits of inter-disciplinary debate and analysis and recommending further research directions.

Defining concepts and parameters

Defining concepts like ‘gender’, ‘sex’, ‘risk’ and ‘genetic risk’ is not a straightforward matter as each definition is underpinned by implicit assumptions and emphases which are embedded in the disciplinary perspective from which the definition comes. While the
Gender and Sex

From a sociological perspective, gender is a fundamental construct within social and cultural life. According to Broom (2005:96), the concept of ‘gender’ is widely accepted as referring to the socially constructed categories of feminine and masculine, categories which dictate roles and behaviours for, and between, men and women in society. By contrast, ‘sex’ refers to the biological characteristics, genetically and hormonally-based, which are associated with people identifying as being either male or female; thus a man or woman’s sex is attributed to having either XX or XY chromosomes which then code for hormonal and physiological distinctions (Keleher & Murphy 2004:20). Sex and gender are intricately related. Lupton (1992:230) describes this in terms of gender being ‘how biological [sex] differences are interpreted and translated into social expectations in everyday life’ (Lupton 1992:23). Cattell (1996:87) puts this in another way: ‘Women and men are socially created [emphasis added]’; she argues ‘gender may have little to do with biological sex’.

The dichotomising approach to men/women, male/female has been increasingly challenged within sociological analysis of gender however as being simplistic and unhelpful in advancing our understanding (Broom 2005:96). Connell (2002:10) argues that the key focus for examining gender should not be upon the differences between men and women as...
fixed dichotomous categories but rather upon understanding how society and its structures deal with human bodies, whether they be male or female; for Connell, defining gender involves examining the ‘structure of social relations that centres on the reproductive arena’. As a result of such analysis and critique, the previously clear distinctions between ‘gender’ and ‘sex’ have become blurred and the terms are therefore becoming more likely to be used inter-changeably within sociological discussions (Broom 2005:96).

Within the domains of biomedical, clinical and genetics health care however, the intricacies, distinctions and debates about sex and gender remain largely under-emphasised and unexplored. Biomedicine is the dominant paradigm for understanding health and illness in Western countries like Australia and is a scientifically-based approach that focuses on the biological, physiological and genetic factors that determine health, illness and gender (Taylor 2008a:26). From this perspective, the body can be viewed as a machine with intricate workings and genetic, biological and physiological inter-relationships (Taylor 2008b:10). According to this perspective, a patient’s sex is biologically, hormonally and genetically determined and this is a fundamental and automatic ‘reality’ when viewing health status or diagnosing disease; the biological differences between men and women thus constitute a foundational context in which to examine health and disease status (Connell, 2002:30).

Risk and Genetic risk
Risk is a concept that has increasingly defined social and medical discourses in Western society (Germov 2005:43); in 1992 Beck coined the term ‘the risk society’ to describe this. Risk generally refers to hazards or dangers and the potential for individuals to run into danger or be exposed to injury or loss; within the risk society, focus becomes centralised on risks and how they are calculated and managed (Beck 1992).

Within the clinical genetics framework, genetic risk is a foundational concept and is defined as the probability that a trait, condition or disorder will occur or recur in an individual or family more frequently than it occurs by chance (Weil 2000:117). Genetic risk can be determined by examining a person’s family history and the transmission of the relevant inherited condition or by undertaking a genetic test to identify genetic markers or mutations in genes that are associated with the genetic condition in question (Weil 2000:117).

Within health care, health promotion and genetic medicine, the focus upon risk, the surveillance of risk and the individual’s responsible management of their risks is now ubiquitous (Petersen & Bunton 2002:56; Petersen & Lupton 1996). From a sociological perspective, the concept of risk has been strongly critiqued as it engages the potentialities of otherwise healthy people regarding future illnesses and engages all citizens in being ‘potentially ill’ (Petersen & Lupton 1996; Beck 1992). This critique has also been applied to the clinical genetics context wherein genetic risk is seen to have significant implications for how people regard themselves and their bodies and how they formulate self identities as genetic identities (Petersen & Bunton 2002:56). In a seminal paper in 1992, the American
geneticist and physician Paul Billings (Billings et al 1992: 476) warned of the dangers of stigmatisation and discrimination against the ‘asymptomatically ill’ that is, discrimination against healthy people on the basis of their genetic characteristics. Comprehensive research has since been undertaken in regard to the issue of genetic discrimination (Taylor et al 2007).

Since the mapping and sequencing of the human genome in 2001, genetic testing technology has expanded quickly and is becoming increasingly integrated into the health care system. Over 200 genetic tests are now available in Australia for clinical diagnosis of genetic disorders (Human Genetics Society of Australasia, 1999) and other tests can be accessed internationally (ALRC/AHEC 2003). As well as diagnosing genetic disorders, screening population groups for genetic risks or predispositions and identifying carriers of faulty genes for couples who may be at risk of having children with genetic disorders, genetic tests can be used to predict future disease prior to the development of symptoms by identifying variations in genes that cause, or predispose individuals to, the relevant illness (Haan 2003:459; ALRC/AHEC 2003).

For disorders like Huntington disease and inherited Alzheimer’s disease that are caused by single mutations, a predictive genetic test result can indicate with certainty that the relevant condition will develop in the future, or not, depending on the presence or otherwise of the faulty gene; for conditions like inherited breast, ovarian or bowel cancer that are caused by multiple factors including those which are genetic in nature, a predictive test will indicate increased risk, susceptibility or predisposition only regarding future development of the
condition. These two types of predictive genetic tests are described as *pre-symptomatic* or *predictive*, respectively. Predictive tests are expected to be increasingly utilized to inform people of their risks or predispositions in regard to common conditions such as heart disease, cancer, diabetes and mental illness (Haan 2003:460).

Documented reasons for undertaking predictive or pre-symptomatic testing include a desire for increased certainty in regard to genetic risk status, future planning including reproductive decisions and to help other family members like offspring clarify their own genetic risks (Taylor 2004; Chapman 2002; Evers-Kiebooms et al. 2000; Cox & McKellin 1999; Hallowell 1999). To undertake predictive testing is a complex decision however as a person might have to accommodate knowledge about increased risk or future illness for many years, sometimes also knowing that the condition is untreatable. The potential treatability of the condition in question is usually a significant consideration in an otherwise healthy person’s decision to undertake a predictive genetic test. In addition, concerns about such people being treated negatively because of genetic characteristics established through genetic testing or family history of an inherited condition, for example in regard to life insurance, have been widely documented and are not unfounded (Taylor et al, 2008; Otlowski et al. 2002).

**Gender, health and genetic risk**

It has been widely accepted that men and women have different health status profiles and outcomes, experience health and illness differently and respond to health and illness issues
differently (Broom 2005: 95). The latest report of the Australian Institute of Health and Welfare regarding the health of Australians [Australian Institute of Health & Welfare 2008:25-42] for example describes different life expectancies at birth for men and women, a greater likelihood of women in reporting a long term or chronic condition although men and women appeared to experience similar types of long term conditions, and different causes of disabling conditions as well as death for men and women in Australia. Further, women appear to experience greater psychological distress in relation to health issues but are more likely to proactively seek health-related information and to engage with medical systems, health professionals and positive health care behaviours (Broom 2005:98). By contrast, while males remain comparatively silent and invisible in their health-and illness-related behaviours, they have higher death rates and are more likely to commit suicide (Broom 2005:98). Statements such as ‘Women get sicker but men die quicker’ provide catchy, albeit blunt, assessments of the generalized differences between men and women in regard to their health experiences, profiles and outcomes (Broom 2005:98; Keleher & Murphy 2004: 21).

Similar trends are being increasingly reported in regard to apparent differences between men and women in genetic health and clinical genetics contexts: that is, women and men are described as differentially experiencing genetic risk and utilizing genetic health services such as genetic testing; these differences are being noted in spite of the genetic conditions in question conferring no differential risk to either sex. In 2000 for example, in regard to the serious inherited neurological disorder of Huntington’s disease, clinical geneticist Michael Hayden reported “[the] interesting finding that most people requesting predictive
testing [for Huntington disease (HD)] are women (about 60% worldwide)” (p.1945). Other researchers have reported similarly (Taylor 2005:152; 1994: 353; Hayden 2003:142; Creighton et al 2003:462; Harper et al 2000:567). In 2003 Creighton and colleagues reported findings from a comprehensive study of genetic testing for HD that was undertaken in Canada. Of 1061 predictive genetic tests undertaken across Canada between 1987 and 2000, 60.2% were undertaken by at-risk women, compared to 39.8% of at-risk men ($\chi^2 = 22.0$, $p<0.0001$). HD confers no differential genetic risk to either sex however. Harper et al. (2000) reported similarly within the United Kingdom as did Tibben et al. (1997) within Europe. Within the Australian context, Taylor (1994) had earlier described a gender differential in the uptake of HD predictive testing in Australia between 1987 and 1993 with significant differences in men and women’s engagement with genetic testing at different life stages.

Gender-based differences in behaviour regarding genetic risk, information and testing have also been documented with respect to other inherited neurological disorders (Hayden 2003.142), familial cancers (Gaff et al 2006:771; Evans et al 1997:748) and carrier status testing for cystic fibrosis (Marteau et al 1997:51). Further such differences appear constant in regard to different types of genetic tests, as indicated in 1998 by Marteau and Croyle (1998:693) who, after reviewing relevant literature, concluded “[W]omen are more likely than men to undergo [genetic] carrier tests, pre symptomatic tests and pre-dispositional tests”.

**Explaining gender-based differences in health and genetics contexts**
As indicated earlier, views about gender, sex differences, risk and genetic risk are complex and often contested. Possible explanations for apparent gender-based differences in health and genetics contexts are underpinned by the beliefs and assumptions that are implicit within the conceptual, theoretical and disciplinary frameworks that attempt to explain the phenomena.

From the biomedical and genetics medicine perspectives, sex-based differences are examined according to their biological or genetic features. Thus where women develop breast or cervical cancer and men develop prostate cancer, the biological basis of diseases is clearly sex linked. Similarly, biomedicine will focus upon sex-based differences where hormonal or physiological features of men and women are impacting upon or influencing the disease in question, its diagnosis or prognosis. Within the genetics context, there are inherited conditions that are specifically sex-linked such that the inheritance mechanism operates at a chromosomal level and the genetic conditions or carrier status are associated with one sex or the other; haemophilia, Fragile X syndrome and Duchenne and Becker types of muscular dystrophy are examples of sex-linked inherited conditions (Centre for Genetics Education 2008). The primary foci therefore of biomedicine and genetic sciences as they are traditionally taught and operationalised within health care contexts are upon sex-based differences in terms of underlying physiological and/or molecular processes.

Psychiatry, psychology and other bio-psychosocial disciplinary perspectives have had long-standing and significant engagement with gender in terms of examining sex-based
differences in individuals particularly in regard to health decisions and behaviours (Taylor 2008b:35; Sarafino 2006). Here differences between men and women’s decision-making, beliefs, assessment, perceptions and responses to risk and a multitude of other facets of individual behaviour that pertain to health and genetics contexts have been explored and examined (Sarafino 2006; Shiloh 1996:82). Psychological literature relating to risk perception, threat and appraisal suggests for example that men and women respond differently to information about risks to their own, and/or their children’s, health (Marteau et al., 1997). According to evidence reviewed by Marteau and colleagues (1997), women reportedly had a greater fear of hazards, and associated greater risks with technology, than did males (the latter appeared unrelated to differences in knowledge about the risk). Explanations involving differences in cognition, threat appraisal and self-efficacy have all been indicated as relevant as have attitudes towards the importance of reproductive risks (Marteau et al 1997; Shiloh 1996:82).

The bio-psychosocial perspective largely underpins the emerging specialty professions of clinical genetics and genetic counselling which are based upon the knowledge and practice interface between genetic science, risk and psychological functioning. Early trends indicating the apparently greater uptake by women of genetic testing than men were tentatively explained by reference to psychological concepts including women’s greater capacity to deal with and cope with emotional challenges such as those associated with genetic testing and their greater emotional proximity to reproductive issues and concerns (for example, Hayden 2003: Harper et al 2000). Such explanations have implicitly drawn from social psychology and its focus upon sex-role socialization of boys and girls, their
identification and internalization of roles and norms that help set their identities and subsequent behaviours including health-related behaviours (Sarafino 2006; Connell 2002:12).

Finally, sociology and many of its sub-specialties, undertake analysis from socio-cultural, political, historical and critical bases and have engaged extensively with questions about gender differences, risk, health and genetic risk. From a range of sociological perspectives, it appears to be accepted that continued focus on differences between men and women in terms of health and illness outcomes, experiences or behaviours including those relating to genetic risk and genetic testing, is unproductive. It is agreed that gender as a dichotomous and solitary category is unable to explain differences in men and women’s health experiences and health outcomes as it is intricately connected with, and significantly impacted upon by other key features of the social and cultural context, such as age, class, income and occupation (Charles & Walters 2008:117; Broom 2005:96; Keleher & Murphy 2004: 21). Sargent and Brettell (1996:93) propose that a life stage approach for analysing gender and health through the life course is particularly fruitful.

Men and women are thus differentially exposed to health risks across their life stages, within their employment, leisure or relationships as well as according to their income, education or geographical location; such differential exposures, while ostensibly expressed through different behaviours, are underpinned by the gendered organisation of social structures, roles, expectations, opportunities and the embodied experience. Broom (2004: 96-102) describes health and illness as gendered phenomena, with exposure to risk and
subjective experiences of health and illness being gendered and significantly impacted upon by interactions between gender and social organisation. She concludes (2004:96) ‘the relationship between gender and health is a complex interaction among material circumstances, physical entities, cultural processes and social organisation’.

Biomedicine and genetic science, their products and processes have also been critiqued from a range of sociological perspectives. Attention has been drawn to the pervasive aspects of an increasing geno-culture which is underpinned by gene-ist assumptions and practices and rapidly developing genetic technologies, and questions are asked about who will benefit from such technologies, who may be excluded and who may be oppressed through their use (Willis 2005:241; Petersen & Bunton 2002:1). Feminist scholars bring a strong analysis to such questions through analysis of the power and gender relations that are implicit within these fields and their professional activities, particularly in regard to reproduction and associated technologies (Ettore 2002:65; Stacey 1996:331). Such analyses draw attention to the rise of surveillance medicine, the importance of language, the greater engagement of biomedicine with women’s bodies which have been comparatively constituted as abnormal and the stronger imperatives for women to manage risk, not only for themselves, but for significant others (Lupton 2005:195; Armstrong 2002:112; Petersen & Bunton 2002:35).

These themes resonate within genetics contexts where Richards (1995) for example describes women as the ‘genetic housekeepers’ and ‘kinkeepers’ within families, primarily responsible for initiating contact with genetics clinics, collecting information within
families and informing significant others about risks. Recent important work by d’Agincourt-Canning (2006:462) and Burgess and d’Agincourt (2001:361) presents extensive analysis of the increasingly gendered and relational nature of responsibility for managing genetic issues and the potential impact of genetic information upon the embodied self, the relational self and the social self. Earlier, Taylor (2004:137) and Hallowell (1997:597) discussed the moral imperatives and expectations that are embedded within the responsible management of genetic risks within families.

Assumptions about the objectivity, rationality and certainty of genetic risk have also being challenged by examining the subjectivity and significance of lay beliefs in relation to behaviour and by locating genetic science, and technology within socio-cultural contexts. Cox and McKellin (1999, p.641) for example, after conducting extensive ethnographic work, argue that “while Mendelian genetics may provide a coherent framework for calculating the odds of inheriting a disorder … they do not recognise the liveable framework … for understanding risk as it emerges within everyday life”. Similarly Richards and Ponder (1996) demonstrate how “lay” understandings and subjective experience of concepts like genetic risk are significantly influenced by kinship and family beliefs about inheritance and can vary considerably from scientific explanations. Rees et al. (2001:1433) and Sanders et al (2003:53) concluded that the perception of genetic risk is a subjective experience which can vary considerably from ‘actual’ family history and be influenced by beliefs about relationships within a family or physical resemblances to affected relatives.
Finally important theoretical work continues to be undertaken in subject areas relating to identity, embodiment, masculinity and femininity and the move away from dichotomous notions of differences between men and women based upon sex and gender (Connell 2002; Pease 2001). Connell (2002:28) systematically dismantles evidence regarding sex differences that are underpinned by biological factors and sex roles and introduces notions of multiple masculinities and femininities which constitute identities. Pease (2001:16) argues against generalising about a traditional and singular notion of masculinity that is characterised by ‘emotional stoicism, homophobia, emphasis on work and achievement, competitiveness, distant fathering, neglect of health needs and mistrust of women’; instead he also argues for the importance of recognising multiple masculinities. Such views have been strongly influenced by post modern analysis which challenges the dominant notion that gender defines individuals and their identities in a fixed way (James in Pease 2001:39).

As gender is socially constructed, it is argued therefore that its influence upon the identities of individuals is contextualised and intricately related with other key characteristics including class, age and life stage, ethnicity and personal history. Self identity, a construct influenced by both gender and embodiment, is not static but fluid, dynamic and contextualised across history, family and personal experience. Thus a person’s sense of being masculine or feminine will change accordingly: from situation to situation and also influenced by structural factors such as class, life stage and ethnicity. Men and women are therefore not singularly homogeneous groups of people whose main differentiating characteristic is their gender or sex but people who potentially embody multiple masculinities and femininities, influenced, determined and enacted through complex social
processes and contexts. Connell (2002:47), engaging with sociological concepts of structure and agency and their interconnection, describes such processes as social embodiment. Thus, how men and women encounter and engage with health, illness, risk and medical systems is significantly influenced by socially organised structures that are enacted and experienced by gendered bodies. Connell argues (cited by Watson 2000:41) ‘gender analysis needs to go beyond gender’ while Watson (2000:140) advocates greater understanding of ‘the lived experiences of all healthy and unhealthy bodies [emphasis added]’.

The application of these ideas within the genetics context implies that the engagement, or not, of men and women with genetic risk and genetic testing will vary, not simply according to whether they are male and female per se, but in conjunction with their gender and social embodiment at that time as well as other characteristics such as relationships with others, education, rurality, disadvantage, access to genetic services and information, parenting status, age and life stage, ethnicity and cultural values and so on. This has important implications for genetic clinicians for whom it may be productive to consider such defining features of their clients’ experiences and identities as well as to challenge embedded assumptions that gender is fixed and defining of client identity.

**Conclusion**
This paper has undertaken a brief conceptual overview of the concept of gender and its inter-relationship with health, risk, genetic risk and genetic testing. The perspectives of different disciplinary frameworks including biomedicine, psychology and sociology
contribute significantly to our understanding of these concepts, the apparent differential engagement of men and women with health and genetics issues and the complex inter-relationships between them. These concepts, while being foundational and ubiquitous in health, risk and health behaviour domains, are contested however. The idea that men/women, male/female, masculine/feminine are fixed, dichotomous and difference-based concepts are being challenged and alternative understandings are being proposed which instead promote multiple and contextualised masculinities, femininities, gendered relations and social practices. Thus, from a range of sociological perspectives, gender becomes meaningful and useful as a concept primarily when considered in relationship with other structural and contextualised features of individuals’ lives, identities and experiences. Age and life stage seem particularly relevant to consider in this context. Much is to be gained from a reflective and inter-disciplinary approach to the analysis of gender in the health and genetic health contexts and from engaging with cross disciplinary analysis, debate and critique. Within the genetics context, further research and application of these ideas and concepts is indicated in order to elucidate our understanding and to maximise the potential benefits that new genetic technologies can deliver.

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