Title of Paper Attitudes of medical genetics practitioners and psychiatrists towards communicating with patients about genetic risk for psychiatric disorders

Short running title Communicating with patients about psychiatric genetics

Yi Zhou Zhou, 1 Alex Wilde, 2,3 Bettina Meiser, 1 Philip B Mitchell, 2,3,4 Kristine Barlow-Stewart, 5,6 Peter R Schofield 5,7,8

1 Prince of Wales Clinical School, University of New South Wales, Sydney; 2 School of Psychiatry, University of New South Wales, Sydney; 3 Black Dog Institute, Sydney; 4 Brain Sciences UNSW, Sydney; 5 Centre for Genetics Education, Sydney; 6 Sydney Medical School, University of Sydney; 7 Neuroscience Research Australia, Sydney; 8 School of Medical Sciences, University of New South Wales, Sydney.

Correspondence details
Dr Alex Wilde, School of Psychiatry, University of New South Wales, Black Dog Institute Building, Hospital Road, Prince of Wales Hospital, Randwick, New South Wales, 2031.
Email: alex.wilde@unsw.edu.au

Conflict of Interest statement
Y. Zhou Zhou: None declared, A. Wilde: None declared, B. Meiser: None declared, PB Mitchell: Editorial board member, Psychiatric Genetics K. Barlow-Stewart: None declared, PR Schofield, Editorial board member, Psychiatric Genetics.

Abstract
Objectives: To examine the self-rated competencies and perceived roles of medical geneticists, genetic counselors and psychiatrists in the communication of genetic risk for psychiatric disorders to patients and families at increased risk for schizophrenia, bipolar disorder or major depressive disorder, and their perspectives on training needs in this field.

Methods: Clinically active members of the Human Genetics Society of Australasia and the Royal Australian and New Zealand College of Psychiatrists were invited to complete the online survey. One hundred and fifty-seven responses were included in data analysis: 17 medical geneticists, 36 genetics counselors and 104 psychiatrists.

Results: 34.4% of respondents disagreed that their professional training had prepared them to discuss genetic information about psychiatric illnesses with patients. Medical geneticists perceived significantly higher levels of self-rated competency to discuss with patients and families genetic information regarding psychiatric disorders, compared with genetic counselors and psychiatrists ($t=-0.61, p=0.001$) ($\beta = 0.33, 95\% \text{ CI: } 0.16, 0.49, p<0.001$).

Conclusions: Findings suggest deficiencies in education and training programs concerning how to best communicate psychiatric genetic risk information to patients, suggesting that specialist programs are needed to better support health professionals. As self-rated competencies differed among the professional groups, training programs need to be tailored to participants’ professional backgrounds.

Key words: Genetic counseling, psychiatric disorders, genetic risk information, genetics training, medical education.
Introduction

Major depressive disorder (MDD), schizophrenia and bipolar disorder are among the leading causes of disability globally (Organisation, 2008). These conditions have strong genetic origins, with heritability estimates of 85% for bipolar disorder, 81% for schizophrenia and 37% for major depressive disorder (Bienvenu et al., 2011). Accumulating evidence indicates that the mode of inheritance is most likely complex with multiple genes of small effect interacting with environmental factors (Stefansson et al., 2009), though rare structural variants are responsible for a yet-to-be-determined minority of cases for some mental illnesses (Consortium, 2008). Some of the specific small effect genes are now being identified by major international genome wide association studies (GWAS) consortia (Ripke et al., 2011, Sklar et al., 2011).

Genetic testing for psychiatric illnesses is currently unavailable in clinical settings, but several commercial companies are presently marketing several genetic tests direct-to-consumer (DTC) via the Internet that purport to predict susceptibility to MDD, schizophrenia and bipolar disorder and psychotropic medication response (Mitchell et al., 2010). Individuals at an increased genetic risk of developing these disorders and relatives of affected individuals have reported interest in genetic testing, despite the lack of evidence of a role played by genetic testing in therapeutic decision-making (Mitchell et al., 2010). Preliminary data suggest individuals have shown a preference for undertaking testing for genetic susceptibility to MDD, if it were available, via ‘trusted health professionals’ (Wilde et al., 2010). For psychiatric illnesses, such health professionals would most likely include psychiatrists or genetics professionals. Austin et al. (2006) anticipated that the implications of DTC genetic testing for psychiatric disorders would justify making psychiatric genetic counseling routinely available. In consideration of this, health professionals must be ready to
respond to potentially strong interest in psychiatric genetic counseling among patients, relatives and the public.

Several studies have investigated the attitudes of psychiatrists (Hoop et al., 2008b, Finn et al., 2005, Peay et al., 2002) and genetic counselors (Martin et al., 2012, Monaco et al., 2010) towards discussing genetic information related to psychiatric disorders. Psychiatrists’ attitudes have been reported to be generally positive towards clinical uses of genetic testing for psychiatric illnesses (Hoop et al., 2008a). However, several areas of knowledge deficits have been described (Finn et al., 2005). A disparity between perceived responsibility and actual competency in managing genetic testing for psychiatric illnesses has also been reported for psychiatrists (Hoop et al., 2008b, Finn et al., 2005). These are particularly urgent issues, especially in light of the increased accessibility of commercial DTC genetic testing. Insufficient knowledge and competency may hinder the ability of psychiatrists, medical geneticists and genetic counselors to provide adequate and appropriate genetic counseling.

Data are needed to inform the development of future education and training programs to improve expert counseling and risk communication for people at increased genetic risk for psychiatric disorders. The current study is the first to investigate medical geneticists’, genetic counselors’ and psychiatrists’ perceived roles and competencies in psychiatric genetic risk communication, as well as their preferences for further training in this field. Medical geneticists are generally physicians with specialist training in medical genetics, while genetic counselors are typically trained in genetic counseling at the masters degree level (Lobb et al., 2001). This study aimed to ascertain potential differences in perceived roles in the psychiatric genetic counseling setting by testing the following hypotheses. First, there will be differences between medical geneticists, genetic counselors and psychiatrists in terms of their
levels of self-rated competency in providing genetic risk communication, and the extent to which they perceive psychiatric genetic counseling as pertaining to their roles. Second, the period of time since most recent professional genetics training will be associated with differences in levels of self-rated competency and perceptions that psychiatric genetic counseling pertains to one’s role. Finally, health professionals’ endorsement of a genetic component in psychiatric illnesses will be associated with perceptions that increased public knowledge about genetics in mental illness would have a psychosocial impact.

Methods

This cross-sectional study was conducted via a web-based survey hosted by the University of New South Wales KeySurvey program. The study was approved by the Medical and Community Human Research Ethics Advisory Panel, University of New South Wales.

Sample

A group e-mail was sent to all members of the Human Genetics Society of Australasia (HGSA) and the NSW state branch (the largest branch) of the Royal Australian and New Zealand College of Psychiatrists (RANZCP). Individuals were invited to follow the link posted in the email to consent and commence the survey. An advertisement was also placed in the fortnightly electronic newsletter of the RANZCP. Inclusion criteria were working at least part-time in a clinical setting and past or current training towards qualifications in genetics or psychiatry. Eligible individuals who completed the survey had the option of receiving a book about psychiatric genetic counseling (Peay et al., 2011). For the purpose of this report, the term ‘health professional’ refers to psychiatrists, medical geneticists and genetic counselors.
Measures

A copy of the survey items is included in Supplementary Table 1. In the introductory part of the survey, the term ‘psychiatric illness’ was defined as referring to MDD, schizophrenia and bipolar disorder. Items within scales and subsections were randomized in order to minimize possible ordering effects. The survey instrument included both items used in previous related surveys by Finn (2005) and Hoop (2008a, 2008b) as well as purposively developed items.

Demographics

Data on sex, age group, professional background, field of practice, period since last professional genetics training and location of training (if outside of Australia) were collected.

Beliefs about heritable components of psychiatric and non-psychiatric illnesses

One item asked respondents to rate their perceptions of the degree of influence that genetics had on a person’s general mental health from ‘none at all’ to ‘strong influence’. Seven 4-point Likert-type items were used to rate participants’ perceptions of the degree of risk attributable to genetics for Huntington disease, cardiovascular disease, breast cancer, colon cancer, bipolar disorder, schizophrenia and depression, with response options ranging from ‘none’ to ‘strong’.

Perceived competency regarding psychiatric genetic risk communication

Four 5-point Likert-type items were used to assess the level of agreement with five statements (Figure 1) relating to competencies in a range of aspects concerning psychiatric genetic counseling for these disorders. Cronbach’s alpha for the five items was 0.72, and a summary mean score was calculated for analysis with possible values ranging from 1 to 5, with larger values indicating greater perceived competency.
Perceived role in psychiatric genetics risk communication

One item asked respondents to rate their level of agreement with the belief that it was their role to discuss genetics information regarding psychiatric disorders with patients and families; response options ranged from ‘strongly disagree’ to ‘strongly agree’. Six items presented hypothetical clinical scenarios concerning psychiatric disorders and required respondents to consider whether they would address the issue themselves or whether they would refer to a colleague if they knew a medical geneticist, genetic counselor or psychiatrist who could provide expert genetic counseling to patients at increased genetic risk (Figure 2). Cronbach’s alpha for the six items was 0.48; a summary score calculated for analysis had possible values of 0 to 6. One item also assessed whether respondents were aware of another professional (including a medical geneticist, genetic counselor or psychiatrist) who provided expert genetic counseling to patients at increased genetic risk for psychiatric disorders in the geographic area in which they worked (yes or no).

Perceived importance of different components of psychiatric genetics risk communication

One item assessed whether respondents routinely took family histories of psychiatric disorders during the evaluation of patients, with response options ranging from ‘strongly disagree’ to ‘strongly agree’. In addition, six 5-point Likert-type items were used to rate the
importance of six components of expert genetic counseling from ‘not at all important’ to ‘very important’.

Training and education needs
Participants were asked to rank seven components of genetic counseling from least important (rank order = 1) to most important (rank order = 7) to be addressed in training programs. Five formats in which additional training materials may be presented were provided for ranking from least useful (rank order = 1) to most useful (rank order = 5) when used in training programs (shown in Supplementary Table 2).

Perceived impact of genetics knowledge
Eleven 4-point Likert-type items assessed respondents’ perspectives on the impact of genetic knowledge on a range of psychosocial and clinical factors (Supplementary Table 3) from ‘no influence’ (1) to ‘strong influence’ (4). The seven items that measured positive views yielded a Cronbach’s alpha of 0.76; a summary score was calculated for analysis (range 7 to 28). The four items that addressed negative views yielded a Cronbach’s alpha of 0.64 and a summary score consisting of possible values from 4 to 16.

Data analysis
Data were analyzed using the Statistical Package for the Social Sciences 18 (SPSS Inc., Chicago, IL). Non-normally distributed continuous outcome variables were re-coded into binary variables, using a median split. To compare attitudes between medical geneticists, genetic counselors, and psychiatrists, multivariable analyses were conducted, while entering potentially confounding variables (age, sex) as covariates. Multiple linear regression was
used for normally distributed outcome variables and logistic regression for the re-coded binary outcome variables.

**Results**

**Respondent Demographics**

Of the 157 individuals who were included for analysis, 17 were medical geneticists, 36 were genetic counselors, and 104 were psychiatrists. Table 1 shows participants’ sociodemographic characteristics and professional backgrounds.

[Insert Table 1 about here]

**Beliefs about the Heritable Components of Psychiatric and Non-psychiatric Illnesses**

Most medical geneticists, genetic counselors and psychiatrists believed that genetics or heredity had a moderate to strong influence on a person’s mental health (94.1%, 86.1% and 96.2% respectively). The majority of health professionals believed Huntington disease had a strong genetic component, mean(SD)=4.0(0.2). The majority also attributed a moderate genetic component to cardiovascular disease, mean(SD)=3.0(0.5), breast cancer, mean(SD)=3.1(0.6), colon cancer, mean(SD)=3.0(0.7), bipolar disorder, mean(SD)=3.2(0.6), schizophrenia, mean(SD)=3.1(0.6) and MDD, mean(SD)=2.9(0.6).

**Health professionals’ self-rated competency to communicate with patients about genetic risk for psychiatric disorders**

Fifty-two percent (52.2%) of respondents agreed or strongly agreed to feeling competent to discuss genetic information with patients and their families, mean (SD) =3.3(0.9). Figure 1 shows health professionals’ rating of their perceived competency to deliver different aspects
of psychiatric genetic counseling for MDD, schizophrenia and bipolar disorder. Thirty-four percent (34.4%) of respondents disagreed or strongly disagreed that their professional training had prepared them to discuss genetic information with patients and their families. In multivariable analyses, medical geneticists (mean competency score=3.8) were found to rate their perceived competency regarding psychiatric genetics risk communication at a significantly higher level than genetic counselors (mean competency score=3.3) and psychiatrists (mean competency score=3.2), when simultaneously controlling for age ($p=0.39$) and sex ($p=0.007$) ($\beta=0.33$, 95% CI: 0.16, 0.49, $p<0.001$). The rating of perceived competency in genetic communication about psychiatric disorders was not significantly associated with the time passed since most recent training in genetics ($r=-0.13; p=0.11$) for the three groups combined.

[Insert Figure 1 about here]

**Perceived roles in the communication of genetic risk for psychiatric disorders**

Most respondents (91.1%) agreed or strongly agreed that they perceived it was their role to discuss genetic information regarding psychiatric illness with patients and their families, mean (SD)=4.17 (0.80). In multivariable analyses, compared to medical geneticists, there was a strong trend for psychiatrists to believe it was their role to discuss genetics information with patients and families (OR 0.32, 95% CI 0.104, 1.000, $p=0.05$). Similarly genetic counselors were significantly less likely to believe that it was their role to address genetic information for such patients (OR 0.16, 95% CI 0.04, 0.62, $p=0.008$), compared to medical geneticists. The period since most recent training in psychiatric genetics and perceived role in genetic communication in psychiatric illness were not significantly associated ($r=-0.72; p=0.37$) across the three health professional groups combined.
Figure 2 shows the number of psychiatrists, medical geneticists and genetic counselors, who would personally address issues raised in a range of hypothetical scenarios as opposed to referring to a colleague. Only 26.8% of respondents were aware of another health professional (including a medical geneticist, genetic counselor or psychiatrist) in the geographic area in which they worked who provided expert genetic counseling to patients at increased genetic risk for a psychiatric disorder.

Perceived importance of different components of communicating genetic risk for psychiatric disorders

More than half (77.7%) of all respondents agreed or strongly agreed that they routinely took detailed family histories of psychiatric illness during the evaluation of their patients. All respondents reported that taking a detailed family history was important or very important. The majority also believed that identifying patient beliefs (96.2%) and conveying genetic risk information (96.8%) was important or very important. A large proportion (90.4%) believed that it was important or very important to convey recurrence risk information to such patients. Participants considered that emphasizing to patients the role of both genetic factors (82.8%) and environmental factors (94.9%) in the development of psychiatric disorders was important or very important.

Training and education needs

Table 1 shows that 64.7% of medical geneticists and 55.6% of genetic counselors reported not having received any training in psychiatric genetics, compared to 22.1% of psychiatrists. The three target health professional groups ranked the mean importance of seven components
of genetic counseling to be potentially addressed in training programs. The most highly ranked skill was ‘exploring patient concerns’, mean(SD)=5.1(2.0), followed by ‘effectively communicating genetic information to patients’, mean(SD)=4.6(1.6); ‘taking a detailed family history’, mean(SD)=4.6(2.1); ‘providing advice to assist in patient decision-making processes’, mean(SD)=4.1(1.9); ‘effectively presenting risk values to patients’, mean(SD)=3.7(1.6); ‘calculating risk information for an asymptomatic person with a family history of psychiatric disorder’, mean(SD)=3.0(1.0) and ‘calculating recurrence risk values from patient history’, mean(SD)=2.96(1.73).

Supplementary Table 2 displays the mean rankings that respondents assigned regarding the usefulness of five formats in which further training may be provided. ‘Face-to-face workshop groups’ was ranked as most useful (mean ranking 3.7, SD=1.3) whilst ‘paper-based materials’ were ranked as least useful (mean ranking 2.4, SD=1.3).

**Health professionals’ perceptions about psychosocial and clinical impact of increased genetics knowledge amongst the public**

The highest ranked positive and negative psychosocial or clinical impacts ('moderate influence' or 'strong influence') of increased public knowledge about the genetics of psychiatric disorders was the 'discovery of new and better treatments' (75.0%) and 'potential denial of insurance to patients with high-risk genes for psychiatric illness' (68.1%), respectively (Supplementary Table 3). Health professionals’ endorsement of a genetic model for psychiatric disorders was not significantly associated with perceptions that greater knowledge of genetics amongst the public would have a positive impact ($r=0.83; p=0.30$) or negative impact ($r=0.07; p=0.38$).
**Discussion**

Over 90% of participants in this study agreed or strongly agreed that it was their role to discuss genetics information regarding psychiatric illness with patients and their families. These findings are comparable to those from a similarly sized sample of U.S. psychiatrists, where 83% reported perceiving it as their role to communicate genetic information about psychiatric illnesses (Finn *et al.*, 2005). In another US study, 78% of psychiatrists considered themselves to be the most appropriate professional to counsel psychiatric patients about genetics in psychiatric illnesses (Hoop *et al.*, 2008b). Few psychiatrists in the U.S. study felt that it was the role of a genetic counselor or medical geneticist (13% and 7% respectively). In the present study, medical geneticists were more likely to perceive it to be their role to discuss genetic information regarding psychiatric disorders when compared to psychiatrists and genetic counselors. This may be due to the multidisciplinary approach adopted by genetics services in Australia. For example, a study which focused on specialised familial clinics for breast cancer in Australia suggested that genetic counselors often facilitate the majority of counseling sessions, including gathering medical and family histories (Lobb *et al.*, 2001). The study found that medical geneticists seem to have a more specialised role in providing risk assessments or answering specific questions (Lobb *et al.*, 2001). Given the perceived differences in clinical roles among groups of health professionals, future training programs need to address communication skills and teamwork models that allow a multidisciplinary team approach to genetic counseling for psychiatric illnesses.

Thirty-four percent of participants in the present study disagreed that their professional training had prepared them to discuss genetics information about psychiatric illness with patients and their families. This is comparable to figures reported by Peay and McInnerney (2002), where 32% of 41 practicing genetic counselors felt ‘very unprepared’ or ‘somewhat
unprepared’ to raise the issue of psychiatric illness with patients, and 44% felt similarly unprepared to answer patients’ questions about psychiatric disease. Similarly, only 23% of psychiatrists surveyed in a US study reported themselves as competent to communicate genetic information about psychiatric illnesses (Finn et al., 2005). In the present study, however, medical geneticists showed a significantly higher level of self-rated competency than both psychiatrists and genetic counselors.

The results of the present study further underscore previous findings (Finn et al., 2005) that there are perceived deficits amongst health professionals in conveying genetics information about psychiatric illnesses to patients. The findings reported here reflect a disparity between perceived role and perceived competency among the three target health professional groups. This perceived deficiency should be addressed by future training programs.

The study also found that the proportion of health professionals who would personally address psychiatric genetics issues as opposed to referring to a colleague with expertise varied widely depending on the particular hypothetical scenario being considered. In the scenario of counseling a pregnant couple with a family history of psychiatric illnesses, who are considering terminating the pregnancy, more than 80% of the three health professional groups would refer to a colleague. However, over 70% of respondents were not aware of another professional within their geographic area who could provide expert genetic counseling for psychiatric disorders. The difference between health professionals’ willingness to refer patients and awareness of colleagues with suitable expertise to refer such patients indicate deficiencies in current professional networks. If there were a demand for genetics services in psychiatry stimulated by increased accessibility to genetic testing, such a demand would not be met by the current health system.
Sixty-five percent of medical geneticists and 56% of genetic counselors reported not having received any training in psychiatric genetics, compared to 22% of psychiatrists, suggesting that training of genetics practitioners have the greatest unmet training needs. Specialist training will assist health professionals to determine appropriate and efficacious methods for the communication of genetics information about psychiatric disorders to patients, whilst also providing the basis for the development of a non-directive, patient-oriented approach to risk communication.

In terms of the preferred format of future training programs, face-to-face workshop groups with other health professionals and experts and talks or seminars with experts were perceived as most useful. This is possibly due to these training formats being able to provide a more experiential learning environment that is able to directly address participants’ needs, as suggested by a previous study that examined the effectiveness of professional development workshops for genetic counselors in oncology (Dunlop et al., 2011).

There was no significant association between health professionals’ endorsement of a genetic component in psychiatric illness and their perception that greater public knowledge about genetics in psychiatric illnesses would have a positive or negative impact. This suggests that health professionals may view genetic research into psychiatric disorders as relatively benign in terms of public impact at this stage. This finding is comparable to the perspectives of a group of genetics researchers in another study who believed that expectations regarding the impact of genetics research on clinical practice were unrealistic, given the limited evidence available on the genetics of psychiatric illnesses (Erickson et al., 2011).
Limitations of the present study include the opt-in nature of the survey, which may have generated participation bias. This could not be evaluated as details on individuals who did not participate were not available. Health professionals’ rating of competence in communicating about genetic risk for psychiatric disorders to their patients was highly subjective. Future studies are needed to investigate actual, objectively-rated competence by documenting the process and content of communication about psychiatric genetic risk using rigorously designed studies of communication behaviors (Meiser et al., 2008). This study reflects the views of a small group of health professionals in Australia. Perceived roles, competencies and training deficits in psychiatric genetics may differ elsewhere and should be considered on a region by region basis.

The findings provide a starting position from which to guide the development of future training and education courses in psychiatric genetics for health professionals charged with patient care in this field. The study has revealed major deficiencies in the training of health professionals, who will be increasingly called upon to provide genetic risk information and counseling to susceptible patients and their families about psychiatric disorders. Such deficits will become increasingly critical with increasing public awareness about the importance of family history in determining genetic risk for psychiatric disorders and as more individuals access DTC genetic testing that requires professional advice and guidance.

Acknowledgements

The authors are grateful to the health professionals who completed the survey. The authors also thank Dr Michelle Peate for her guidance in using the KeySurvey program. A/Prof Bettina Meiser was supported through Career Development Awards (ID 350989 and ID
1003921) from the National Health and Medical Research Council (NHMRC). Professor Philip B. Mitchell’s research is supported by an NHMRC Program Grant (510135).
References


Figure legends

Figure 1. Percentage of medical geneticists, genetic counselors and psychiatrists who agreed or strongly agreed with statements relating to self-rated competency regarding psychiatric genetic risk communication for major depressive disorder, schizophrenia and bipolar disorder (N=157).

Figure 2. Percentage of medical geneticists, genetic counselors and psychiatrists who would personally address issues raised in some hypothetical clinical scenarios (N=157).