Discovering the gene for schizophrenia: Analysis of the presentation of molecular genetics in *The Australian and New Zealand Journal of Psychiatry*

Julie Henderson
School of Nursing & Midwifery
Julie.Henderson@flinders.edu.au

Abstract:
This paper explores the presentation of information about molecular genetics in the *Australian and New Zealand Journal of Psychiatry*. Content analysis of the journal reveals that the proportion of articles citing genetics as a cause of mental illness has increased within the last 10 years, in line with an increase in articles about biological psychiatry in general. A causal link between genes and behaviour has yet to be established. The current wisdom is that mental illness arises from a combination of, as yet unidentified genes, which increase susceptibility to environmental pressures. Despite a failure to identify the genes responsible for mental illness, psychiatry demonstrates an ongoing commitment to, and faith in molecular genetics. This paper explores the framing of information about molecular genetics in *The Australian and New Zealand Journal of Psychiatry* highlighting the tension between faith in molecular genetics and a failure to establish a genetic cause for mental illness. This tension will be explored in relation to the dominance of a biological paradigm within psychiatry and negation of social and environmental factors in the development of mental illness.

> "Just as the 1990s is the 'decade of the brain', the next decade promises to be the 'decade of the gene'" (Levy, Barr & Sunohara 1998: 99).
>
> "The traditional genetic aetiology for schizophrenia is that it is 'multifactorial'… It is a word used when geneticists do not know" (Turner 2000: S207)

Introduction

Molecular genetics has been touted as providing a "'magic bullet' to alleviate human problems" (Conrad 1997: 142) or "the first draft of the book of humankind" (Smart, 2003: 25). This understanding is reflected in the public perceptions' of genetics. Novas & Rose (2000: 487) argue that the popular imagination has been captured by "molecular optics": a vision of life in which we see and explain life at a molecular level "in terms of

the molecular structure of bodily components, the molecular processes of life functions and the molecular properties of pharmaceutical products" (Novas & Rose 2000: 487). This understanding of life is associated with the belief that illness and behavioural disorders have a genetic basis; that the genes responsible for these illnesses and behaviours can, and will be identified in time and that the identification of these genes will allow for the screening of those at risk of developing disorders (Novas & Rose 2000: 486).

There has not been uncritical public acceptance of genetic technologies. There is disquiet about the manner in which these technologies may be used to "taper with nature", with the creation of "designer babies" or with the possibility of creating a "genetic underclass" (Smart, 2003: 33). Others argue for consideration of the ethical implications of genetic screening. Concerns include the creation of an obligation to be screened and to act upon medical advice to manage the risk to future family members (Novas & Rose, 2000) and that genetic screening may make "families pedigree….the object of expert gaze" (Koch et al. 2005). For Hilgartner (1990: 520) scientists manage public criticism of science through demarcating the boundaries between popularist knowledge about science and "genuine" scientific knowledge. Scientists can label negative representations of science as 'distortion' through arguing that popularist perceptions of science are based on simplification of scientific work. 'Genuine' science in contrast, is presented as value-free and neutral due to the objectivity of the scientific method allowing Scientists to protect their work from outside challenge (Kerr, Cunningham-Burley and Amos 1997; Petersen, 2001).

This paper explores the presentation of information about molecular genetics in *The Australian and New Zealand Journal of Psychiatry.* Content analysis of the journal demonstrates a growing interest in molecular genetics reflected in an increasing proportion of articles citing genetic causes of mental illness. Psychiatry has yet to identify the genes associated with mental illness. Molecular Geneticists, unlike earlier Geneticists, have to demonstrate a causal relationship between genes and mental illness. They assume that this relationship exists on the basis of earlier family, twin and adoption studies (Joseph, 2003). Implicated genes are identified through linkage studies which explore the statistical likelihood of a genetic marker or biological difference present in

those with the condition, occurring in consanguineous family members with a disorder (Joseph 2003; Russell 1995). At best, linkage studies can only identify the area on the chromosome which may be implicated in mental illness (Joseph, 2003). The current wisdom therefore, is that mental illness arises from the interaction of multiple genes, each of which have a "small effect that increase susceptibility" to disorders (Joseph 2003: 284). This approach is reflected in a 'multifactorial threshold model' which associates the risk of developing mental illness with a critical mass of genes which increase mental fragility which may become mental illness when triggered by environmental stresses (Hallmayer 2000; Jablensky 2000).

Despite widespread recognition of the limitations of molecular genetics, it is firmly established as a legitimate model of aetiology in *The Australian and New Zealand Journal of Psychiatry*. Analysis of articles about molecular genetics elicited four themes relating to the potential of molecular genetics; the limits of current knowledge; the impact of molecular genetics on mental health prevention; and the social and ethical implications of genetic technologies. Underlying these themes is a tension between a continuing belief in the possibilities of molecular genetics, a lack of knowledge about the genes causing mental illness, and the limitation of genetics in preventing and curing mental illness. This paper explores these issues in relation to the dominance of biological psychiatry and devaluing of the role of social and environmental factors in causing mental illness.

Methods

The Australian and New Zealand Journal of Psychiatry was published quarterly during 1967-1995, six times a year from 1996-2003 and ten times a year from 2004. The sample for this paper consists of the 777 articles published between 1980 and the end of 2005 containing a discussion of the aetiology of mental illness. Content analysis of these articles was undertaken to determine the number of articles which adopt biological and in particular, genetic explanations for mental illnesses. This data was used to map changes in the number and proportion of articles adopting a biological and genetic focus.

Thematic analysis was also undertaken of articles published between 1996 and 2005 which identify genetics as a cause of mental illness. This period was chosen as it was

when molecular genetics first appeared in this journal. Fifty-one articles cited molecular genetics as cause of mental illness during this time. Of these, in the majority (29 articles), genetics was discussed in passing. The remaining 22 articles had a primary focus on molecular genetics. These articles were subject to thematic analysis using a grounded theory approach. Thematic analysis elicited four themes. These themes address the potential of molecular genetics; the limits of current knowledge; the role of genetics in mental health prevention and social consequences of genetic technologies.

Molecular Genetics: Paradigm change?

Table 1 reports the results of the content analysis of the journal. It demonstrates the proportion of articles in the *Australian and New Zealand Journal of Psychiatry* which cite biological, and in particular, genetic causes of mental illness. Prior to 1996 the journal published articles exploring the relative merits of genetic and environmental reasons for the higher incidence of mental disorders within families. The period 1996-2000 witnessed a burgeoning interest in molecular genetics (n=33) accounting for 12.7% of all aetiological articles. While there were less articles about molecular genetics published between 2001 and 2005 (n=18) the proportion of genetics articles increased to 16.1% due to a reduction in total number of articles addressing the aetiology of mental illness.

Table 1: Number and proportion of articles that use genetics to explain mental illness in the *Australian and New Zealand Journal of Psychiatry* 1981-2005

	1981-1985	**1986-1990**	**1991-1995**	**1996-2000**	**2001-2005**
Total articles with an aetiological focus	98	121	139	259	112
Articles with a genetics focus	6	7	5	33	18
Genetics articles as a proportion of all aetiological articles	**6.1%**	**5.8%**	**3.6%**	**12.7%**	**16.1%**
Total articles with a biological aetiological focus	34	51	64	137	86
Proportion of articles with a biological aetiological focus	**35%**	**42%**	**46%**	**53%**	**77%**

This trend reflects the extent to which biological psychiatry has gained ascendancy.

Kuhn (1996) argues that new sciences experience stages in which there is competition between diverging scientific paradigms. A paradigm gains ascendancy when it is viewed as having a greater capacity to explain a social or natural phenomenon than the previous paradigm. Once a paradigm has gained general acceptance it becomes accepted as 'normal science' (Kuhn 1996: 24). Normal science becomes the framework for determining the problems to be addressed and the apparatus to solve them allowing some conditions and problems greater visibility than others through directing attention to "the articulation of those phenomenon and theories that the paradigm already supplies" (Kuhn 1996: 24). The proportion of articles adopting a biological focus has increased from 35% of all articles about the aetiology of mental illness in 1981-1985 to 77% of all aetiological articles in 2001-2005 reflecting the extent to which biological psychiatry has become normal science. This change reflects the number of articles citing the role of either brain structure; levels and receptivity to neurotransmitters such as serotonin or dopamine or genes in causing mental illness. The total number of articles addressing aetiology has fallen from 137 in the period 1996-2000 to 86 in the period 2001-2005. This may indicate the extent to which debate about aetiology has been resolved in favour of biological psychiatry.

The Potential of Molecular Genetics

The dominance of a biological paradigm is also evident in the reporting of the potential of molecular genetics in managing mental illness in *The Australian and New Zealand Journal of Psychiatry*. Conrad (2001) found that popular media reporting of molecular genetics is framed in the language of discovery and advancement. While genetic scientists claim to be objective and value–free they often adopt the imagery or metaphors operating in wider society, framing discussion of molecular genetics around genetic discoveries and the potential for elimination of disease (Petersen 2001). For Conrad (2001: 230) this results in 'genetic optimism' reflected in the framing of news stories around the ideas that "a gene for the disorder exists; it will be found; and it will be good."

Thematic analysis demonstrates that this approach is evident in *The Australian and New Zealand Journal of Psychiatry*. Molecular genetics is presented as "beginning to show exciting results" (Blair, Mitchell & Schofield 2005: 542) and as "one of the most active

and promising areas of modern psychiatric research" (McGuffin & Southwick 2003: 657). Discussions of the potential of genetic research in psychiatry often refer to success in discovering the genes associated with behavioural disorders with an identifiable biological basis such as Huntington's Chorea and Alzheimer disease. Jablensky (1999: 137) notes, for example, that "molecular genetics has been successful in identifying genes that play a role in the susceptibility to Alzheimer's disease." This is presented as evidence of the capacity of genetic advancements to enable the discovery of the genetic causes for mental illness. Thus Jablensky reports that:

> The genes predisposing to disorders such as schizophrenia, bipolar affective illness, autism and other developmental disorders, a well as genes contributing to temperament and character traits, will eventually be identified (1999: 137).

Limits of Current Knowledge

Reporting of the potential of genetic advancement is balanced by discussion of the limits of current knowledge. While most articles (n=21 [95%]) accept the proposition that mental illness has a genetic component, a number of articles highlight how little is currently known. Cloninger (1999: 176) states, for example, that "single gene hypotheses were pursued for schizophrenia and bipolar disorders…[but that] no such discoveries have been made" while Jablensky (2000) notes that the genes currently understood to be involved in the transmission of mental illness are found relatively commonly within the population as a whole. A number of articles also point to a failure to replicate genetic linkages. McGuffin and Southwick (2003: 659) describes the results of linkages studies as "inconsistent and confusing" while Hallmayer (2000) indicates that genomic scans for schizophrenia have resulted in the implication of regions on more than half of the 23 chromosomes but that none of these findings have been consistently replicated

Despite reservations, the same articles provide evidence for the existence of a genetic cause of mental illness. McGuffin and Southwick (2003: 659) note that there is "persuasive evidence of linkage regions in schizophrenia" while other articles view a failure to identify the specific genes associated with mental illness as support for polygenic and 'multifactorial threshold models' which associate mental illness with a critical mass of genes (Cloninger 1999; Hallmayer 2000; Jablensky 2000). This trend

demonstrates the extent to which biological psychiatry and genetics have become normal science. Faith is such, that the failure to identify the genes associated with specific disorders, has led some psychiatrists to conclude that the current disease categories should be abandoned in favour of nosologies based upon genetic phenotypes (Berrios, 1999). Current classifications are seen as "fundamentally flawed and ineffective...[in so far as they enshrine] categorical labels that imply the presence of natural boundaries between major syndromes" (Cloninger 1999: 174). The assumption is that research will soon yield clearer understandings of the causes of many major mental illnesses resulting in "these disorders being diagnosed on the basis of cause rather than descriptions [of behaviour]" as occurs in the current disease nosology (Frances and Egger 1999: 162).

A consequence of the widespread acceptance of genetic causes for mental illness is the designation of the social environment to a secondary role. Environmental factors are viewed as risk factors with exposure increasing the susceptibility to developing mental illness, a correlational rather than casual relationship. Hallmayer (2000) for example, states that mental illness arises from a combination of genes which create the predisposition to develop mental illness, a liability that can be modified by environmental factors. Mental illness is seen as arising from "gene-environment interactions" (Hallmayer 2000: S51) as a result of "genetic vulnerability, coupled with environmental and psychosocial stressors" (Raphael 2000: 9).

Genetics and Mental Health Prevention

Novas & Rose (2000: 489) argue that genetic determinism evident in the belief that "genes 'cause' disorders" negates social solutions to social problems and promotes research designed to identify and manage individual rather than social pathologies. Funding for mental health prevention is currently targeted towards indicated preventative strategies which consist of early intervention with those identified as being "at risk of developing more severe disorders with the aim of taking appropriate action to lessen the risk" (Australian Health Minister 1998: 14). Indicated strategies target people who, by virtue of exposure to risk factors, are identified as having a higher relative risk of developing a condition than the general population (Herman 2001). Generally, those designated as being genetically at-risk are encouraged, through genetic counseling, to

adopt behaviors to lessen the risk of developing a condition through lifestyle changes (Koch et al. 2005).

A multifactorial understanding of disease causation impacts upon strategies for preventing mental illness. Six articles explore mental health prevention. All question whether genetics can provide a basis for preventative strategies in mental health. Level of genetic risk of mental illness is determined statistically on the basis of twin, adoption and linkage studies, and is a correlational rather than causal relationship (McGrath 2000). Yet those who are designated as being statistically at-risk only account for three out of 20 people developing the condition (Jablensky 2000). The majority of people developing mental illness do not have a history of a family member with the disorder (Raphael 2000). Further, as mental illness is viewed as polygenic, genetic screening is an ineffective as each gene is understood to have a small impact, making it difficult to screen for all relevant combinations (O'Toole 2000).

These deficiencies lead to calls for alternate prevention strategies, however recognition of the impact of environment factors does not translate into mental health strategies which manage that environment. Mental heath prevention strategies cited in the *Journal* include: improved prenatal nutrition and better antenatal care for women with a family history of mental illness; education about the risks of marijuana use; immunization against prenatal infection; social skills training for withdrawn children; and early intervention with those demonstrating symptoms of mental illness (McGrath 2000; Raphael 2000).

Social consequences of molecular genetics

Just as the role of the social environment is largely absent from the *Australian and New Zealand Journal of Psychiatry*, so is reflection upon the social and ethical implications of genetic technology. Kerr, Cunningham-Burley and Amos (1997) found that Geneticists manage concerns about genetic technologies through appealing to the objectivity of the scientific method. For Scientists, those issues which are viewed as pertinent are those which can be regulated through research protocols or through legislation against discrimination (Smart 2003: 27). Only three articles address ethical issues, two of which highlight the ethical implications of screening children and adolescents. Morley et al.

(2004) identify three issues to be considered: whether there are any benefits to be gained from early detection; the stigma associated with being labeled as at-risk of developing a disorder and the impact of screening on family dynamics in the form of reduced parental expectations. Yeh et al. (2004) view the rights of parents to make decisions which may have a long-term impact on children as problematic and voice concerns about how this information may be used by schools and insurance companies. Both argue that these issues can be addressed, in part, through the maintenance of confidential case records.

Conclusion

This paper has explored the presentation of information about molecular genetics in *The Australian and New Zealand Journal of Psychiatry*. It argues that Psychiatrists adopt as form of 'molecular optics' that is reflected in a commitment to, and faith in genetic explanations of mental illness, in face of a failure to identify the implicated genes. This is seen as being secondary to the dominance and widespread acceptance of biological psychiatry which leads to research and interventions which devalue the impact of environmental factors in causing mental illness, reinforcing the role of biological psychiatry as normal science.

References

Australian Health Ministers (1998) *Second National Mental Health Plan.* Canberra: Mental Health Branch, Commonwealth Department of Health and Family Services.

Berrios, G. (1999) 'Classifications in psychiatry: A conceptual history', *Australian and New Zealand Journal of Psychiatry* 33: 145-160.

Blair, I., Mitchell, P. and Schofield, P. (2005) 'Techniques for the identification of genes involved in psychiatric disorders', *Australian and New Zealand Journal of Psychiatry* 39: 542-549.

Cloninger, C. (1999) 'A new conceptual paradigm from genetics and psychobiology for the science of mental health', Australian *and New Zealand Journal of Psychiatry*, 33: 174-186.

Conrad, P. (1997) 'Public eyes and private genes', *Social Problems* 44(2): 139-154.

Conrad, P (2001) 'Genetic optimism: Framing Genes and mental illness in the news', *Culture, Medicine and Psychiatry* 25: 225-247.

Frances, A. and Eggar, H. (1999) 'Whither psychiatric diagnosis', *Australian and New Zealand Journal of Psychiatry* 33: 161-165.

Hallmayer, J. (2000) 'The epidemiology of the genetic liability for schizophrenia', *Australian and New Zealand Journal of Psychiatry* 34 (supplement): S47-S55.

Herman, H. (2001) 'The need for mental health promotion', *Australian and New Zealand Journal of Psychiatry* 35: 709-715.

Hilgartner, S. (1990) 'The dominant view of popularization: conceptual problems, political uses', *Social Studies of Science,* 20,: 519-539.

Jablensky, A. (1999) 'The nature of psychiatric classification: Issues beyond ICD-10 and DSM-IV', *Australian and New Zealand Journal of Psychiatry* 33: 137-144.

Jablensky, A. (2000) 'Prevalence and incidence of schizophrenia spectrum disorders: Implications for prevention', *Australian and New Zealand Journal of Psychiatry*, 34 (supplement): S26-S34.

Joseph, J. (2003) *The Gene Illusion: Genetic research in psychiatry and psychology under the microscope*. Ross-on-Wye: PCCS Books.

Kerr, A., Cunningham-Burley, S. and Amos, A. (1997) 'The new genetics: professionals' discursive boundaries', *Sociological Review* 45(2): 279-303.

Koch, L. and Nordah Svendsen, M. (2005) 'Providing solutions-defining problems: the imperative of disease prevention in genetic counseling', *Social Science& Medicine,* 60: 823-832.

Kuhn, T. (1996) *The Structure of Scientific Revolutions 3rd ed.* Chicago: The University of Chicago press.

Levy, F., Barr, C and Sunohara, G. (1998) 'Directions of aetiological research on attention deficit hyperactivity disorder', *Australian and New Zealand Journal of Psychiatry* 32: 97-103.

McGuffin, P. and Southwick, L. (2003) 'Fifty years of the double helix and its impact on psychiatry', *Australian and New Zealand Journal of Psychiatry* 37: 657-661.

McGrath, J. (2000) 'Universal interventions for the primary prevention of schizophrenia', *Australian and New Zealand Journal of Psychiatry* 34 (supplement): S58-S64.

Morley, K., Hall, W and Carter, L. (2004) 'Genetic screening for susceptibility to depression: can we and should we?', *Australian and New Zealand Journal of Psychiatry* 38: 73-80.

Novas, C. and Rose, N. (2000) 'Genetic risk and the birth of the somatic individual', *Economy and Society 29*(4): 485-513.

O'Toole, B. (2000) 'Screening for low prevalence disorders', *Australian and New Zealand Journal of Psychiatry* 34 (supplement): S39-S46.

Petersen, A. (2001) 'Biofantasies: genetics and medicine in print news media', *Social Science & Medicine* 52: 1255-1268.

Raphael, B. (2000) 'Prevention in psychiatry: Australian contributions', *Australian and New Zealand Journal of Psychiatry* 34 (supplement): S6-S13.

Russell, D. (1995) *Women, Madness and Medicine.* Cambridge: Polity Press in association with Blackwell Publishers.

Smart, A. (2003) 'Reporting the dawn of the post-genomic are: who wants to live forever?', *Sociology of Health and Illness* 35(1): 24-49.

Turner, G. (2000) 'Presymptomatic screening for schizophrenia: a geneticist's perspective', *Australian and New Zealand Journal of Psychiatry* 34 (supplement): S204-S207.

Yeh, M,. Morley, K. and Hall, W. (2004) 'The policy and ethical implications of genetic research on attention deficit hyperactivity disorder', *Australian and New Zealand Journal of Psychiatry* 38: 10-19.