The Blameworthy Gene

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With improved techniques of genetic analysis, medicine is becoming increasingly reliant on genes to diagnose and treat disease. However, a genetic diagnosis is a value-laden entity with significant potential to change the way we categorize people as ill or healthy, flawed or normal, and responsible for or a hapless victim of disease. Nowhere are these value judgements more prevalent than the field of mental health. This article will examine the meaning and implications of genetic diagnoses, and apply the theories to the example of alcoholism.

“Genetic Diagnosis” Defined and Clarified

“Help us find the gene for insert disease here.” Slogans such as this have become increasingly common since the human genome was sequenced. These slogans seem to imply that once we have found the gene, the cure will naturally follow. They also imply that we cannot be truly certain of a diagnosis until it is proven by genetic technology. These attitudes have the potential to modify the way we conceptualize both the disease and the patient. Is a person with a genetic diagnosis but no symptoms a patient? Is a patient with symptoms but no gene defect simply a malingerer? The answers to these questions have the potential to change the way the person (or patient, depending on the interpretation) is categorized, palliated, and stigmatized.

The term ‘genetic diagnosis’ is oft used in the literature, but requires several clarifications. The first is the timing of the diagnosis. One may have a disease and later discover that it has a genetic component. This impacts the patient by a process that has been termed ‘geneticization’: the patients are relieved of blame for their conditions, they are simply victims of faulty genes. Such a diagnosis, however, does not prevent susceptible individuals from looking ahead; there are implications for family planning as the condition could potentially be passed on to offspring. In contrast, one may receive genetic testing before the onset of symptoms. When the genetic preceding the clinical diagnosis, the individual may be prematurely thrust into the sick role. This broadened definition of the sick role carries with it societal repercussions, as it increases the number of patients requiring intervention. A genetic diagnosis can carry permanent stigma and may damage one’s feeling of personal control.

Secondly, it is important to keep in mind which disease is being examined with the ‘genetic diagnosis’. Certain diseases have more significant genetic contributions, thus affecting the potential for remedy. For instance, a genetic diagnosis of Huntington’s disease correlates well with clinical progression to the disorder whereas depression has approximately 40% heritability. Mental illnesses carry both a high social stigmatization and a low genetic determination, two conditions that Spriggs et al alleged require stringent justification for genetic testing. However, the low genetic determination means that identification of a gene carrier need not be a diagnosis, rather it may be an opportunity for prophylactic intervention. This has been termed the ‘genetic window’.

The final clarification is the distinction between pathology and variation. There will naturally be variations in genes, but these differences need not be correlated with disease. Therefore, a genetic diagnosis may inappropriately label an individual as “ill.” The antithesis of pathologizing the healthy is
legitimizing the plight of the ill. Mental health is often viewed as a disorder, something that is the responsibility of the affected person or the consequence of poor moral fibre.\textsuperscript{14} Once there is a genetic diagnosis, the disorder may qualify as a legitimate disease, one that is now the responsibility of the health care system.\textsuperscript{14,16} Thus, genetic diagnoses must be approached with caution: they have equal potential to inappropriately or appropriately cast individuals in the sick role.

**Nature Versus Nurture**

The distinction between a clinical and genetic diagnosis hinges on the multi-factorial nature of disease. Specifically, the expression of genes can be influenced by the individual’s environment. In a document classic in the realm of health promotion, Hancock summarized these multiple influences in the Mandala of Health (Figure 1). The individual is viewed as the centre of a web of influence, with each spoke having the potential to create or treat disease.\textsuperscript{15} Therefore, while genes themselves cannot be altered by promoting healthy living, there is the potential to mitigate disease through environmental and lifestyle interventions.\textsuperscript{12,14}

**Reactions to a Genetic Diagnosis**

With the meaning of “genetic diagnosis” clarified, the reaction to such diagnoses can be examined. A genetic diagnosis can decrease blame and stigma associated with disease.\textsuperscript{12} In an era of preventive health and personal responsibility, Minkler summarized these value judgements as “when ill is redefined as being guilty”.\textsuperscript{16} In the past, sufferers of mental illness were the victims of societal stigma largely because mental pathology does not fit into the classic Western biophysical approach as there is often not one easily delineated causal mechanism. If there is a causal gene identified, however, the disorder becomes a biophysical disease and the

![Mandala of Health](image)

**THE MANDALA OF HEALTH**

Figure 1. Hancock’s Mandala of Health. The individual is viewed as the centre of a series of spheres of influence, each providing an opportunity for health promotion interventions.\textsuperscript{15}
person now has legitimate claim to the sick role. Notwithstanding this positive reaction to a genetic diagnosis, there can also be negative ramifications. While there was nothing you could have done to prevent the disease, there is nothing you can do now. Minkler warns that a genetic diagnosis may be the same as labelling someone as fatally flawed. However, this fatalistic approach disregards the multi-factorial nature of most mental diseases. The same argument applies to those who receive their genetic diagnosis before the onset of clinical symptoms - the optimism of environmental modification should temper the doom of one’s biological lot.

Given this spectrum of possible reactions, the impact of a genetic diagnosis must be considered at three levels: the patient, the health care system, and society. An individual receiving a genetic diagnosis is now a candidate for early intervention and a legitimate actor in the sick role. Conversely, the genetic diagnosis may turn a healthy individual into a ticking time bomb, waiting anxiously for the onset of symptoms that now seems inevitable. This emphasizes the importance of not viewing a genetic diagnosis as a label of defectiveness nor as an immutable entity. These issues can now be considered in the context of a specific illness - alcoholism.

**Genetic Diagnoses in Practice: Alcoholism**

A number of studies demonstrate that children of alcoholics suffer from a variety of behavioural and psychopathological problems, such as substance abuse, anxiety, depression, conduct disorders and delinquency. This type of behaviour, however, is not consistent across all children of alcoholics. By developing a better understanding of these variations, clinicians can attempt to ensure that children of alcoholics will not succumb to potentially avoidable health problems.

The mode of inheritance of an alcoholic “gene” is far from established, yet there is evidence to suggest that alcoholism is indeed a disease of both genetic and environmental etiology. Past research has demonstrated an increased risk of alcohol abuse in the children of alcoholics. It is possible that this is due to “vertical cultural transmission.” This theory describes the transmission of disorders, or traits which lead to increased susceptibility to a disease, through parental-offspring learned behaviour. However, studies have demonstrated that behavioural transmission is not the only contributing factor when the children of alcoholics develop drinking problems of their own.

For example, a twin-family study by Kendler et al examined the mode by which alcoholism was passed from parents to daughters. The results indicated that a solely environmental etiology was insufficient to explain alcoholism. Furthermore, the researchers found that in the best-fitting model, susceptibility to alcoholism was due in large part to a genetic predisposition.

In a cohort study by Goodwin et al, the prevalence of alcoholism was compared in two groups of adoptees: a group whose biological parents were alcoholics, and a control group whose biological parents were not alcoholics. The researchers found that 18% of those adoptees whose biological parents were alcoholics suffered from the same disease; nearly four times the prevalence found in the control group. These findings offer further evidence of a genetic component of alcoholism.

As there has not yet been a single culpable gene identified, one cannot be “diagnosed” with the potential for developing alcoholism. Notwithstanding, family history may be utilized as a proxy by which to identify targets for health promotion interventions. For example, in a study of college students in the U.S., the children of problem drinkers (COPDs) were identified and compared to a control group, and COPDs were 17% more likely to engage in heavy episodic drinking than non-COPDs. Additionally, they were approximately three times more likely than non-COPDs with similar drinking habits to seek help for their drinking problems. This was particularly true of students who had previously consumed alcoholic beverages but had abstained
from drinking in the past year. In light of these results, it stands to reason that when possible, COPDs, particularly those who have recently attempted to curtail their own drinking, should be targeted for counselling and treatment in order to prevent future alcohol dependency.

Walker and Lee, however, caution that clinicians should not pathologize the children of alcoholics (COAs).\(^9\) They indicate that COAs who come from “fami[lies] with strong emotional bonds and … warm, supportive environment[s]”, are able to maintain caring and empathetic interpersonal relationships, and will not necessarily develop psychiatric disorders, as has been suggested to be typical of COAs. The authors elaborate, explaining that alcoholic families may have “reservoirs of strength”, which can come in a variety of forms, which clinicians should seek out and draw on in order to provide treatment. For example, sibling-sibling relationships within alcoholic families may be the only instance in which a family member is consistently emotionally available, and thus these relationships should be encouraged in an effort to promote healthy living and an avoidance of alcohol dependency.

Walker and Lee emphasize the plasticity of human development and the fact that it can drastically influence the qualities exhibited by COAs; specifically, the exhibition of resilient or maladaptive behaviours.\(^9\) The key to this process is to determine which relationships (i.e. marital, parent-child, sibling-sibling) within the family are resilient, and then encourage the fostering of these relationships, the affirmation of belief systems, and the improvement of communication. Resilient relationships within families may be the counterbalance to genetic predisposition.

Werner\(^11\) adds that COAs who do not develop serious problems tend to have good communication skills, are goal-oriented, have a positive concept of self, and believe in self-help. While these determinants may possess a somewhat innate component, they are also qualities which can be addressed in counselling. By encouraging the development of the aforementioned attributes, it is possible that susceptible individuals may alter their environments in such a way as to not fall victim to a genetic predisposition to alcoholism.

**Impact of a Genetic Diagnosis - Conclusions**

Genetic diagnosis can be costly not only in terms of the gene test itself, but in terms of treating the patients we have created. Before the advent of gene testing, these asymptomatic individuals did not consume health care resources. Ethical and political considerations arise of whether we have an obligation to treat those that we have identified as being ill or at risk, regardless of a lack of clinically recognizable disease. An additional societal consideration is the creation of the “other” - a group of genetically distinct individuals, almost a separate species. This us-versus-them attitude stems from the view of a genetic diagnosis as a fatal flaw and has the potential to perpetuate rather than reduce stigma.\(^13,14\)

On the other hand, early identification of at-risk individuals opens the possibility of health promotion and disease prevention interventions. Alcoholism is but one example of diseases which are all too frequently attributed to a lack of “will power” on the part of the ill. By acknowledging the scientifically demonstrated genetic component of this disease we can play a decisive role in the prevention of alcoholism in those who are most susceptible to the disease.

Being cognizant of the issues discussed in this article is an important step in improving our understanding of the individual and societal implications of genetic testing. Furthermore, recognition that phenotypic expression of genotype is environmentally mediated provides an opportunity for health promotion interventions.

**References**