Genetic medicine: the balance between science and morality

A. Surbone*

Department of Medicine, New York University School of Medicine, New York, NY, USA

This article explores the relationship between science and morality with respect to the major changes that genetic knowledge has induced in medicine, as well as in many other spheres of our lives. The following themes are treated: (i) the influence of genetic knowledge on the concepts of normalcy and diversity with respect to health; (ii) the influence of genetic knowledge on the concept of responsibility; (iii) the reciprocal influence of pre-existing biases and genetic knowledge; (iv) the influence of genetic knowledge on the concept of community; and (v) the influence of genetic knowledge on autonomy and trust in the patient–doctor relationship. The article does not wish to be prescriptive, but rather to raise open questions. The key philosophical question is to what extent human beings benefit from predictions about the future. The role attributed to genetics is largely overestimated. Genetic knowledge can be perceived as enhancing the control that individuals have on their lives, or as paralyzing the decision process of an individual who may feel predestined to a serious disease. In the case of breast and ovarian cancer, the probabilistic nature of genetics is especially relevant, given the relatively low penetrance of \( BRCA \) mutations. “Future things: not our domain. But in this today which unravels in front of us, what shall we do?” (Sophocles, Antigones).

**Key words:** breast cancer, familiarity, moral aspects

**Introduction**

According to the Oxford Dictionary of Philosophy, the central questions of epistemology—the theory of episteme or knowledge—include the origin of knowledge; the place of experience and of reason in generating knowledge; the relationship between knowledge and certainty; the relationship between knowledge and error; the possibility of universal scepticism; and, finally, the changing forms of knowledge that arise from new conceptualizations of the world [1]. Knowledge is not immutable as some ancient philosophers used to believe. Genetics reminds us that knowledge is subject to change in its content as well as in its forms. In contemporary times, genetics has in fact induced profound changes in our way of knowing, by uprooting the traditional sharp distinction between certainty and uncertainty. By being individual, predictive and probabilistic, genetic knowledge is somewhat at variance with traditional medical knowledge [2]. Genetics has suddenly modified some of our long-established medical conceptions, including the key definitions of health and illness. The question that genetic knowledge poses is whether genotypic or phenotypic information should be privileged in defining the interrelated concepts of health and illness. In other words, is the carrier of a genetic predisposition to a disease healthy or sick? Is there a difference between being carrier of a major versus a minor disease? Who is in charge of making this determination, and how does this reflect on our lives?

Genetics seems to have created a new category of the ‘asymptomatically ill’ [3]. This is extremely important, since illness is not only a value-free pathological entity, but rather it can determine deep existential and social changes when the symptom experienced by the patient is recognized as a disease [4]. A simple example may clarify the complex changes that disease may induce. Consider the case of a person experiencing profound fatigue. At first, such a person may ignore the symptom for a given amount of time, but on a certain day that person decides to make an appointment with a physician. Already there has been a change in the attitude of the person experiencing the symptom. After the medical encounter, with an examination and likely with some tests, the symptom experienced at a subjective level will be given a name, i.e. it will be labeled as a disease. The person may thus discover, for instance, that his/her fatigue was due to a transient viral syndrome, or to a more or less profound degree of anemia, or to depression or to a form of leukemia. Each diagnosis will affect each person differently, not only because of the impact that it has at the subjective level, but also in view of the different ways in which any illness is perceived and more or less accepted by the entire society. The influence that each possible diagnosis has on different persons varies in different cultural and social contexts.

The epistemic changes that have followed the development of human genetics are thus clearly accompanied by equally relevant changes in the moral and ethical spheres. By unravelling
the secrets of our genome, we accomplish two major and unexpected tasks; we find out about genetic diversity and we are in the position of making predictions about the future. The repercussions at the social, ethical and moral levels are profound.

The epistemic relationships between knowledge and certainty and between knowledge and error find a correlate in medicine in the two moments of diagnosis and prognosis [5]. Diagnosis is where the objective dimension of disease lies and where the epistemic responsibility of the doctor is first founded. The moment of diagnosis carries deep psychological and existential implications for the patient, and a correct diagnosis traditionally corresponds to the highest moment of certainty in the patient–doctor relationship. Prognosis, as the ability to predict the likely course of a patient’s disease, is less objective, although it is also a distinctive responsibility of the medical profession. Modern medicine aims at making prognosis as accurate and as precise as possible, but there always remain elements of uncertainty. Stressing certainty over uncertainty in prognosis betrays the complexity of life. Diagnosis and prognosis are formidable tools in the hands of the physician. Both have acquired special meaning and relevance in the field of genetic testing, where not only the interplay between certainty and uncertainty (both at the epistemic and at the ethical level) is particularly evident, but also the intertwining of scientific knowledge and normativity is strikingly manifest.

The following discussion focuses on the identification of BRCA mutations involved in hereditary breast and ovarian cancer. BRCA testing is different from other instances of genetic testing, where the gene mutations have a higher penetrance and the ethical issues relate to certainties more than to risks. Furthermore, penetrance varies with different BRCA mutations and in various ethnical groups, and it is presently unknown for sporadic cases of BRCA mutations. This accentuates the probabilistic nature of the knowledge derived from genetic testing for BRCA mutations.

Primary issues in genetics and morality

The questions that genetics has raised are situated between science and morality [6]. Not everything in life necessarily carries a moral value, and many of our daily actions are indeed morally neutral. Science, on the contrary, always carries values, sometimes with deep moral implications and consequences. Ethics is first a matter of identifying and posing the right questions about those issues that affect our lives also at the moral level. Ethics has been defined as ‘a condition of the world, like logic’ [7]. This is why we benefit from a ‘logical analysis’ of all possible ethical ramifications of genetic testing for breast and ovarian cancer predisposition, in order to identify, clarify and possibly anticipate the moral issues that are or can be involved. This is also why this paper is in the form of raising questions. Providing prescriptive and normative solutions to the social, ethical and moral dilemmas of genetics is only possible through an open, honest, continuous debate that involves society in its entirety.

The main areas where genetic testing and, in particular, genetic testing for predisposition to breast and ovarian cancer, seem to hold profound moral implications are: (i) the influence of genetic knowledge on the concepts of normalcy and diversity with respect to health; (ii) the influence of genetic knowledge on the concept of responsibility; (iii) the reciprocal influence of pre-existing biases and genetic knowledge; (iv) the influence of genetic knowledge on the concept of community; and (v) the influence of genetic knowledge on autonomy and trust in the patient–doctor relationship.

(i) Influence of genetic knowledge on the concepts of normalcy and diversity with respect to health

The risk of ‘geneticization’, the identification of a person with his/her genes, parallels the development of human genetics. Geneticization is the widespread tendency to overemphasize the role of genes in disease causation, in medical practice and in social attitudes toward disease. The original definition was provided by Lippman in 1991 [8]. She coined the word geneticization to describe ‘an ongoing process by which differences between individuals are reduced to their DNA codes, with most disorders, behaviors and psychological variations defined, at least in part, as genetic in origin.’ Philosophically, geneticization is a direct consequence of two underlying and seemingly contradicting assumptions: that knowledge and control over nature are strictly related and that knowledge is value-neutral. Francis Bacon first equated genuine knowledge with the power of manipulative control [9]. Bacon also extended the equation ‘knowledge and power’ to the sphere of human security and hope [10]. By so doing, he attributed moral worth to knowledge. Yet a contemplative view of science has been perpetuated in the myth of scientific neutrality, whereby scientists pursue their research without any direct responsibility toward its application [10]. In the clinic, geneticization originates from an underestimation of the role of environmental factors in pathogenesis and of an overestimation of the causative role of genetic factors. Finally, geneticization arises from social and normative practices.

One of the consequences of geneticization is the mistaken interpretation of genetics as a source of diversity, while on the contrary more similarities than differences are shared by living organisms. Diversity, whether genetic or not, is completely compatible with human equality, and this should give us little incentive to misuse genetic data [11]. Yet, misuse of genetic information for more or less subtle discriminative purposes is possible and it has been reported in many fields, including in health insurance, at the workplace, in the processes of adoption and of education, and in the microenvironment of carriers of genetic mutations [3,12–15]. Women carriers of a BRCA mutation are potentially exposed to this risk. Minority and underprivileged women are likely to be at increased risk of discrimination [16].

(ii) Influence of genetic knowledge on the concept of responsibility

The debate with respect to genetic responsibility is very heated, and carries important consequences. Some of us believe that
genetic responsibility exists, but does not differ from any other type of responsibility [12], while others hold the view that the concept of responsibility should change on the basis of genetic information [17]. According to the latter view, there would be an additional responsibility that we owe to society, based on our genome. The appeal to genetic responsibility has been used, for instance, to question the right of a high-risk individual to refuse to be tested and/or to reveal the results of a genetic testing. Because of the possible vertical transmission of a genetic predisposition or illness, the member of a high-risk family or community has moral responsibilities toward other members that go beyond his or her own personal interest [18]. This potentially raises the question of whether a person’s rights to privacy and confidentiality, also with respect to her decision-making process, cease to exist because of a genetic responsibility. In the clinical setting, it is often very difficult to strike a balance between the rights of one person and those of others. Ideally, such a balance should be reached together by the patient, the doctor and the family.

At the level of the single individual, however, the right not to be tested would logically seem to be the inevitable counterpart of the right to obtain genetic testing, as well as an essential component of a person’s autonomy and self-determination [12]. Those who strongly endorse the concept of genetic responsibility would rather claim that the right not to be tested equates to a ‘right to genetic ignorance’. Following this view, autonomy is only possible when based on full information, and thus refusing to be tested (when otherwise considered appropriate from a medical point of view) is an abdication of one’s own autonomy [17].

The claim to a special type of genetic responsibility may also impact on whether we have less or more personal, social and environmental responsibility. The responsibility to improve the level of education or to keep a safe environment or to foster preventive measures does not cease if we know when that child is predisposed to become a bright student, or when that worker is predisposed to be more susceptible to damage from heavy metal exposure, or when that woman is predisposed to develop breast and ovarian cancer. Insisting on genetic responsibility may shift our attention from other responsibilities in our life [12].

Finally, there is concern that any excessive emphasis on genetic responsibility may open the doors to subtle ‘backdoor eugenics’, a very important risk of genetic knowledge in general [19]. Humanity often needs to go through the tragedy of many of our fellow human beings to achieve higher levels of moral understanding [20]. History has made us aware of the reality and absurdity of eugenics and of the risk of more subtle forms of eugenics even in today’s democratic societies [21]. Eugenics stems from social and political decisions and practices, rather than from the scientific discoveries themselves, and arguments against eugenics have always been social and ethical, rather than scientific.

Defining what genetic responsibility is and how far it reaches requires a proper balance between science and morality. The task of finding this balance involves the entire society, and cannot be left only to the political or scientific elite.

(iii) Reciprocal influence of pre-existing biases and genetic knowledge

Discrimination in disguise of a health concern has notably occurred also in the recent history of health care. An example is sexual discrimination under the excuse of public concerns for the HIV infection—a disease with a horizontal transmission. Sickle-cell anemia in the African–American population in the USA is a known example of discrimination based on genetic predisposition [16].

Potential risk exists that predisposition to BRCA-associated breast and/or ovarian cancer may increase already existing gender biases. An employer may in fact not welcome a woman, whose health care expenses, whether in terms of surveillance, prevention or possible treatment, may be substantially higher than those of a non-carrier woman or of a man. In many societies women are still not considered equal to men, and even in our Western societies women undergo subtle forms of discrimination. Most women are still offered lower salaries, even when performing the same duties as men. Poverty is higher in women. Focus on women and on gender justice is necessary [16].

(iv) Influence of genetic knowledge on the concept of community

Among the moral and social repercussions of genetics, there are several that are very positive and most welcome. The reappraisal of the sense of belonging to a community that genetic knowledge has brought about is one of them. Even in those Western societies that privilege individual autonomy over the values of family and community, there seems to be a renewed sense of connectedness. The example of BRCA mutations is one of the most striking: members of families or communities, which had been scattered over the world, are now coming together to face the possible risk of being predisposed to a disease, or to help other members interested in their family history, or just to ‘be there’ for others.

Thus genetics also has the potential to remind us of our similarities much more than of our diversity [22], and to foster a novel appreciation of morality based on relational rather than on individual values.

(v) Influence of genetic knowledge on autonomy and trust in the patient–doctor relationship

There is a strong need for and appreciation of the component of ‘trust’ in the patient–doctor relationship within the context of any form of genetic counseling. The patient–doctor relationship is at the same time a contractual relationship among persons of equal rights and dignity, and a covenant between two fellow human beings based on the vulnerability of one partner and the special knowledge of the other [23]. The vulnerability induced by any illness renders the relationship
between the patient and the doctor an asymmetrical relation of help and care [24]. In recent decades, however, the patient–doctor relationship has suffered from growing economic and legal pressure, becoming increasingly similar to an exchange between provider and consumer.

In the case of BRCA mutations, women and men seeking to be tested or undergoing counseling after genetic testing feel and express the need to trust the experts, whether doctors or genetic counselors. Potential or actual mutation carriers trust the experts’ honesty, truthfulness, and ability and willingness to advocate against any social abuse of genetic information. Trust also goes beyond the individual relationship between the doctors or genetic counselor and the patient or client—a word often preferred by genetic counselors. Trust involves also technology and institutions. Trust involves the media.

Reaffirming the role of trust or the value of community and family ties does not mean abdicating one’s autonomy. It only means redefining it in a more contextual and relational perspective based on the connectedness of human beings.

**Genetics and control**

Ancient Greeks realized that human beings can be haunted by their past, but also by their future. The sense and the fear of predestination have dominated Western philosophy for many centuries. The relationship between human freedom and determinism has been at the center of theology. The era of modern science and technology seemed to have distanced these questions from us. Genetics dramatically reopened them.

The contribution of genetics to our lives is likely to be much more limited than we tend to believe. Yet, being a carrier of a genetic predisposition to cancer has a dramatic impact on a person’s life. In the case of BRCA carriers, genetic knowledge may either increase the sense of control on a woman’s life or it may paralyze the decision-making process by shedding a dim light on the woman’s future. Genetic predisposition and fate can be easily equated, and one of the responsibilities of the physician is to avoid overemphasizing the predictive power of genetic knowledge and to remind his/her patients of the intertwine of genetic and environmental factors. Physicians should respect their patients’ different attitudes toward genetic information. Possible carriers of BRCA mutations may consider with great apprehension the risk of developing breast and ovarian cancer, especially since they generally belong to families with a high incidence of cancer. For other women, the ethical and social risks potentially inherent in genetic testing are an overwhelming consideration. Some women worry considerably about the repercussions that a positive result may have on themselves, as well as on their family dynamics. A woman might decide to contribute to research and science and to be tested even in the absence as yet of definitive preventive measures. A woman may decide to do so for her close or extended family. She may decide to inform her young relatives or she may be afraid of the psychological repercussions. A woman who discovers that she is a BRCA mutation carrier may opt for prophylactic surgery or choose to enter a chemo-prevention trial or simply to be closely monitored. Finally, a woman may refuse to undergo genetic testing and ask to be followed and monitored as closely as if she were a BRCA mutation carrier.

I believe that we, as physicians or genetic counselors or health-care workers in general, should reflect on our patients’ feelings and opinions, and also ask ourselves to what extent we benefit from any degree or form of anticipation of the future. Not, certainly, with the intent to influence our patients’ decision-making process, but rather with the aim of being honest and helpful with them. When discussing the final impact of a predictive test with one of our patients, knowing our own personal standpoint seems to me an essential step in bridging science and morality in the clinic. The answers given when facing a quandary only in theory can be very different from the ones given when faced with the same quandary in reality [25]. Yet, investing time to explore these issues can make us more complete persons and physicians.

What makes genetics so different from other equally important branches of science and medicine is the call to rethink our understanding of what ‘moral agency’ is. The discussion in medical ethics has long been centered and framed around principles that presuppose a traditional understanding of ‘personal moral agency’. The issues now raised by genetics seem to call into question whether moral agency is centered around a ‘me’ or an ‘us’. As an example, we need to understand whether the (limited) control that we may gain from genetic knowledge is going to be achieved at a personal or at the more extended level of a community. The key philosophical and moral issue at stake in discussing genetic predisposition to a serious disease such as breast and ovarian cancer is, in fact, that of ‘control’. This is a moral issue traditionally approached within the context of the ‘free-will versus determinism’ discussion, which assumes that most objective situations and/or life chances are to be seen as circumstances, rather than causes of a certain moral choice, and which generally leaves aside the role of ‘moral luck’ [26]. The concept of ‘genetic abnormality’, on the contrary, is one where scientific and philosophical understandings might usefully intersect, relating statistical distribution and projections to normative standards of acceptability, health and well-being. Within this new context, the normative standards for what could be ‘controllable’, and hence for what is within one’s moral agency, may be extended from a simple individual responsibility to a more collective one. The latter would be embedded in a shared moral understanding, supported by various social practices of labeling, treating and even manipulating [27]. We should then try to speak of ‘control’ within the context of a relational view of our moral agency, by concentrating not only on the individual consequences of finding out who carries a genetic predisposition to a certain disease, but also, and primarily, by finding and defining acceptable and non-discriminatory shared understandings of what a genetic abnormality means for the individual part of a community.

This is the ‘moral revolution’ that genetic knowledge has introduced into our Western morality: genetics has reminded our Western societies, increasingly privileging individualism.
and scientific progress \textit{per se}, of the importance of human connectedness and of never separating science from morality.

\textbf{Acknowledgements}

A.S. was supported in part by a grant from The Chemotherapy Foundation.

\textbf{References}