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Joëlle Vailly

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Genetics

Joëlle Vailly

Inserm, Paris, France

Abstract

The current focus on genetic conditions is part of a fundamental trend that has placed chronic diseases at the forefront of concerns in the “Western” world. This interest is emblematic of the extent to which technoscientific approaches and clinical practice have become intertwined. To a greater extent than other diseases that also attract biomedical attention, genetic conditions call upon a rapidly developing field of knowledge and a fertile imaginary, both of which have altered drastically over the past 30 years. Moreover, their hereditary nature sets them apart from ordinary chronic diseases.

Keywords: DNA; genetics; health; nature–nurture debate

The current focus on genetic conditions is part of a fundamental trend that has placed chronic diseases at the forefront of concerns in the “Western” world. This interest is emblematic of the extent to which technoscientific approaches and clinical practice have become intertwined (Clarke et al. 2010). To a greater extent than other diseases that also attract biomedical attention, genetic conditions call upon a rapidly developing field of knowledge and a fertile imaginary, both of which have altered drastically over the past 30 years. Moreover, their hereditary nature sets them apart from ordinary chronic diseases.

Epidemiology of Genetic Diseases

Epidemiologists estimate that approximately 4 percent of newborn babies are affected by so-called Mendelian diseases (primarily attributed to a gene), chromosomal anomalies (in number or structure), or major malformations (the potential genetic origin of which is not always easy to determine). Surveys reveal

that 15 to 20 percent of adults identified as being at risk of passing on an illness to their descendants wish to carry out a genetic test on the fetus, although this varies according to country and disease (Lock and Nguyen 2010, 311). As for pre-implantation diagnoses, these concern a few thousand children born alive in the world. To these data, it is necessary to add the extreme diversity of the conditions in question: over 4000 Mendelian diseases have been identified. In other words, while their cumulative impact is by no means insignificant, their individual quantitative impact is sometimes quite limited. For this reason, most of them are classified as “rare diseases” which, by definition, affect less than 1 person in 2000 of a given population. Some of these diseases show a different distribution depending on the population in question, which has encouraged the implementation of targeted screening programs with a view to greatly limiting the birth rate of affected children. Examples include Tay–Sachs disease amongst the Ashkenazi Jews in the United States, and thalassemia in Sardinia and Cyprus in particular.

The situation is different in the case of so-called multifactorial conditions such as diabetes, high blood pressure, and obesity, which involve several genetic factors and which are affected by the environment (through diet, being exposed to risks, etc.). The genetic element involved is more or less clearly established. In the case of breast cancer, in particular, 5 percent of cases are associated with a hereditary genetic “predisposition” linked to the BRCA1 and BRCA2 genes, which cause the risk to increase between two and five times. In the case of type 2 diabetes, susceptibility genes have been identified – the softer nature of the term compared to that of “predisposition” should be noted – but these genes entail a very low increase in the risk of being affected by the disease. In sum, whereas Mendelian diseases are rare and clearly of genetic origin, multifactorial diseases are far more common and their genetic element is subject to debate. The rise of genetic diseases to the forefront of current concerns in wealthy countries can therefore not be understood simply in statistical terms.

Knowledge and Society

During the second half of the twentieth century, biologists considered the genome – in other words the total genetic material of an individual – as a program that controlled the characteristics of the living being. Genetics was supposed to

influence how we considered abilities, handicaps, social problems, family relationships, and quality of life (Conrad and Gabe 1999). This is why, in the 1990s, researchers in the social sciences put forward the notion of “geneticization,” referring to an approach to medical or societal questions that was overly influenced by genetics (Lippmann 1992). However, the sequencing of the human genome that was established at the turn of the millennium did not provide the keys to the secrets of life that many biologists had hoped for (Fox Keller 2000). They therefore had to formulate more complex and less deterministic models resting upon the notion of reciprocal effects between genetic and non-genetic factors. This shift towards greater complexity has given rise today to projects involving large banks of samples in order to understand the respective and combined roles of DNA and the environment in the etiology of multifactorial diseases. Research on “epigenetics” is now developing, focusing on the effects of the environment on how DNA is used by cells and the inheritability of these changes in the absence of modified genetic sequences. Moreover, the range of psychiatric and psychological conditions considered to have a genetic component is widening. The molecularization of the living being is thus pursuing its path, integrating greater levels of complexity along the way, and the attention paid to DNA continues.

Over the last few decades, this genetic knowledge has been disseminated in society and has become widely shared, albeit to a limited extent of course. Patient associations and the media have contributed, and continue to contribute, to this propagation of knowledge and to the hopes linked to genetics. In the United States, members of associations have brought about a sort of biological or genetic citizenship by demanding in particular that their experience and wishes in terms of research and health policies be taken into account. In the same vein, a certain “bio-sociality” seems to be developing – a form of sociability bringing individuals together according to their genetic features (Rabinow 1996). However, further research is necessary in order to establish whether the emergence of citizens or subjects on the basis of genetic characteristics is in fact radically different to previous forms of citizenship and subjectification.

This joint construction of a genetic culture within societies also concerns many other actors, such as political leaders. Private sector firms have not been left by the wayside either in this context of the international circulation of biological

knowledge, methods, and materials. The notions of “bio-capital” and “bio-value” testify to forms of organization and exchange that have the specificity of seeing biological material and knowledge entering into transactional chains (Rajan 2006). In sum, the genetic knowledge that is disseminated in society, the alliances that it calls upon, and the media interest it generates, as well as the attention garnered by patient associations, come together to give a certain precedence to genetic diseases and thus feed into policies regarding “life itself” (Rose 2007). What emerges from all of this is a relatively complex way of approaching genetics within society, ranging from the hope of finding cures to the fear of scientific manipulation.

The Properties of Genetic Information

The information provided by medical genetics shares certain properties with that provided by other approaches. However, medical genetics not only reconfigures these properties but also reveals its own specific features. First, if uncertainty is inherent in clinical activity, in this field it is furnished with many new sources. Whereas on the one hand genetic mutations can contribute to establishing a diagnosis, on the other they often cannot predict how serious the disease will be, or how it will progress. In particular, the “penetrance” of genes – that is, the proportion of people carrying the mutation who develop the disease – is variable. Here, the specificity lies within the fact that this uncertainty seems in direct contradiction with the more or less persistent notion in society of genetic essentialism, according to which the essence of who we are (or what a disease is) lies in our genes. This is why the idea of “predictive medicine,” according to which genetic examination will eventually be able to produce information regarding the future health of an individual, seems to want to push genetics beyond its actual remit. With the exception of Mendelian diseases with high penetrance, predictive medicine remains to a large extent virtual, for now at least. Nonetheless, it is indicative of a very real shift linked to the times. Grounded in the medical obsession of early diagnosis, which is now a structural element of medicine, genetic research seeks not simply to anticipate but to do so from as early a stage as possible. This implies a chronological reversal not only between diagnosis and the appearance of clinical signs, but also often between the birth of an individual and the appearance of the disease.

Another property of genetic information is linked to the fact that revealing mutations can contribute to altering the individual or collective identity of the people affected. This is particularly the case when they discover that they carry a mutation that may have a delayed onset (Huntington's disease) or that involves a certain proportion of risk (family breast cancer). Of course, the notion of the "risk" of a disease reaches far beyond the field of genetic conditions, as different kinds of screening or bioassays can also reveal risk. One of the specificities here is that the person carrying the mutation will share this trait with other members of his/her biological family. This raises the question of how family links change once the emphasis is placed upon biology, and it calls upon a conception of family that pre-dates current forms, which are often blended (Finkler, Skrzynia, and Evans 2003). This characteristic is specific to genetics, which is unsurprising when one recalls that genetics is defined in dictionaries as the "science of heredity." Also linked to heredity, another issue arises: that of passing down health, a highly precious commodity in contemporary "Western" society, to one's descendants. Genetic information is therefore twofold: it is both individual by nature and also shared within family groups, or even, as we have seen, within geographical communities. In the latter case, this can reinforce collective identities that are grounded in biology and in ethnic or racial categories, while also being intertwined with older notions of collective history (Wailoo and Pemberton 2006). More generally, genetic knowledge seems to coexist with older modes of clinical perception (Atkinson, Glasner, and Lock 2009, 11); and, for the people concerned, rather than replacing older notions of kinship, heredity and the reasons for ill health combine with them (Lock and Nguyen 2010, 313).

Use of Genetic Tests

Tests for genetic conditions vary in terms of the target (individual, family, population), the aim (research, diagnosis, risk evaluation and prevention, decision regarding reproduction), the developmental stage concerned (embryo, fetus, newborn, child, adult), and the methods used (direct DNA analysis, study of genetic markers, bioassays, examination of chromosomes). These tests raise numerous questions according to their different uses, and only a few key examples, aside from those already mentioned, will be outlined here.

The first question, which concerns the autonomy of people receiving genetic counseling, is particularly relevant here given the extent to which the history of eugenics has made it a sensitive issue. Some research highlights the dilemmas with which professionals are confronted due to the neutral position they must show when faced with parents' reproductive choices, and with which parents are sometimes confronted when faced with a decision to abort for medical reasons (Conrad and Gabe 1999). Other studies show how cultural and social constraints, the influence of professionals, and the lack of social benefits for handicapped people can weigh upon parents' choices. One of these examines the social impact of amniocentesis in the United States, showing how, in a context of cultural and socioeconomic inequality, women become "moral pioneers" who have to take decisions on the cusp of new techniques (Rapp 1999).

Another issue is linked to genetic tests becoming part of health policies, such as population screening. In this respect, neonatal screening is globally the most common form of test for genetic conditions. These tests are increasing rapidly, particularly in the United States where on average more than 40 diseases are screened for at birth. A slower but nonetheless regular increase can also be seen in Europe. When screening is combined with effective treatment, it tends to meet with unanimous approval. However, when no such treatment is available, which is the most frequent situation in the United States, it can be subject to debate. It thus illustrates the evolution of norms concerning screening for genetic conditions, while also shedding new light upon more general societal changes (Vailly 2011).

Another rapidly evolving practice concerns the sale of genetic tests on the Internet. Today, for a few hundred dollars, companies will provide – or rather claim to provide – their clients with information about their predisposition to develop several dozen diseases or traits (breast cancer, heart attack, obesity, tendency towards manic depression, risk of alcohol addiction, etc.). In parallel, private laboratories offer screening to couples for Mendelian diseases before birth, but in doing so neglect the question of the diversity of possible conditions. These different tests raise opposition by geneticists who contest their scientific validity and are concerned about some of their psychological and social effects. Furthermore, observers regularly point out the risk of discrimination by insurance companies or employers. Given the acceleration in sequencing techniques and the

increasing use of DNA chips (small strips upon which a huge number of DNA samples is analyzed), the entire regulation of genetic tests is at stake here. While bioethics laws are sometimes very strict in their regulation of the use of these tests in the hospital context, the self-testing market on the Internet is a legally gray area. This all goes to illustrate the social dynamics and new issues at stake surrounding the living being.

Globally, the development of genetics has had a substantial impact in terms of molecular knowledge and how we view the living being and indeed ourselves. It also has useful applications as a diagnostic aid. However, for the time being at least, it has few direct effects in terms of new treatment other than for the analysis of DNA bacteria or viruses, allowing faster diagnoses and treatment. Gene therapy (correcting a genetic defect using the tools of molecular biology) remains limited to a few success stories, and pharmacogenomics (adapting medication to specific DNA) remains more of a project than a reality. This highlights the need to distinguish between the ambitions outlined by the actors involved, the phenomena that can actually be observed, and the idea that a given society harbors of both.

SEE ALSO: Biosociety and Genetics; Epigenetics; Eugenics; Gene; Genetics and Genomics in Public Health; Medical Sociology and Genetics; Prenatal Diagnosis and Screening

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