

## Diagnosis: a critical social reflection in the genomic era

### Diagnóstico: uma reflexão social crítica na era genômica

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**Abstract** *Diagnosis is a pivotal tool for the work of medicine as they categorise and classify individual ailments via a generalised schema. However diagnosis is also a profoundly social act, which reflects society, its values and how it makes sense of illness and disease. Considering diagnosis critically, as well as practically, is an important job of the sociologist. This paper reviews how a social model can provide a critical tool for viewing diagnosis in the genomic era. It explores how the formulation of diagnosis, be it via genetic explanations or microbiological ones, are the product of social discovery, negotiation, and consensus.*

**Key words** *Sociology of diagnosis, Power, Conflict of interest, Stigma, Classification*

**Resumo** *O diagnóstico é uma ferramenta essencial para o trabalho da medicina, uma vez que categoriza e classifica o padecimento do indivíduo por meio de um esquema genérico. No entanto, o diagnóstico também é um profundo ato social, o qual reflete a sociedade, seus valores e como dá sentido para o sofrimento e a doença. Considerar o diagnóstico de maneira crítica, assim como prática, é um trabalho importante dos sociólogos. Este artigo analisa como um modelo social pode fornecer uma ferramenta crítica para vermos o diagnóstico na era genômica. Explora como a formulação do diagnóstico, seja através de explicações genéticas ou microbiológicas, é o produto da descoberta social, negociação e consenso.*

**Palavras-chave** *Sociologia do diagnóstico, Poder, Conflito de interesses, Estigma, Classificação*

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## Introduction

The field has often made calls for sociologies. From the sociology of stuttering to the sociology of mobile phones, “sociologies” are language of appeal: this particular topic deserves sociological attention. Let’s make it the object of study! The call for a sociology of diagnosis had a rather different intent. From its initial mooted in 1978, by Mildred Blaxter<sup>1</sup>, to its status today as a *bone fide* sub-discipline, sociologists of diagnosis have focussed on how this particular sociology shines a light, not on itself as an object of study, but other aspects of social life: power, distribution of resources and interests. Elsewhere I have written that diagnosis is “a kind of focal point where numerous interests, anxieties, values, knowledges, practices and other factors merge and converge”<sup>2</sup>. It is implicated in global politics, commercial agendas, health care relationships, boundary work, and so on.

Emerging as a discipline, the sociology of diagnosis was first an idea, proposed by a number of scholars, discussed and debated amongst them; then a collection/special issue; a monograph, and a field. It now fills the role that Sarah Nettleton and I argued it should in the first special issue: that diagnosis “provides a not only a category and process but a neat analytic tool that serves as a prism that reflects and casts light on a multiplicity of issues in health, illness and medicine”<sup>2</sup>.

Diagnoses have increasingly been at the fore as a result of evidence-based practice movement, which has argued, with overwhelming success, for clinical medicine to use principles of clinical epidemiology. This in turn is only possible in the presence of countable categories of disease, that is to say, diagnoses. Diagnoses are the means by which cases are assembled to enable generalisation. Recognising the similarity between cases, and thus diagnosing them, achieves a number of classificatory aims, including organizing knowledge<sup>3</sup>; recognising clusters<sup>4</sup>; and perhaps, above all, as Richardson wrote at the beginning of the last century, reducing a disorderly mass to an orderly whole<sup>5</sup>.

Categorization of course, is a social activity, as deciding how the continuum of human function will be broken into manageable and explainable parts is the result of deliberation, power and consensus<sup>6</sup>. How conditions come to be given official status within medical diagnostic systems like the *International Classification of Diseases* or the *Diagnostic and Statistical Manual of Mental*

*Disorders* is a matter of interest to the sociologist of diagnosis.

The way in which diseases are categorized says a great deal about what a society values, how it makes sense of nature, as much as it does about pathophysiology. It takes more than the technical capacity to recognise an ailment for it to be given diagnostic status. For example, that microscopy enables one to view the spirochete responsible for Lyme disease is of no interest if no one is attempting to look. In the case of Lyme, the fortuitous disease cluster amongst children in Lyme Connecticut enabled a recognition of similarity which could then lead to its technological discovery<sup>7</sup>.

But at the same time, there has to be a will to see a particular disorder as disease. Contested diagnoses like electromagnetic or multiple chemical sensitivities are often overlooked, and become point of extreme contest between sufferers and the medical institution<sup>8</sup>.

Diagnoses are the categories we use to create order; sort through particular symptoms and presentations; place them together or apart, and do the work of medicine. The categories used in medicine reify, serve as heuristic and didactic structures, determine the treatment protocol, predict the outcome, and provide a sense of identity for lay and professional alike<sup>9</sup>. They also serve to explain deviance. Conrad & Schneider<sup>10</sup> and Rosenberg<sup>11</sup> all have demonstrated, in different ways, how diagnosis guards the boundary between deviance and normality. What may not fit social norms, can be variably viewed as bad, or as sick.

Diagnosis, is, as Blaxter famously described, both a category and a process<sup>1</sup>. The process is the means by which the diagnostic category does its social work. Already it designates social role, as lay person approaches clinician in the pursuit of diagnostic explanations. Each has her own role as the diagnostic process is undertaken. But at the same time, many other social functions are triggered and resources allocated. Treatment and prognosis are determined, medical speciality is defined, and identity rejigged.

The diagnosis can legitimise as well as stigmatise; not all diagnoses are created equal. Diseases with sexual or psychiatric connections may reduce the social status of the diagnose. To have AIDS or Syphilis has a very different impact than arthritis or influenza. Dag Album has referred to “disease prestige” in a model which shows medical predilection for particular diagnoses ranked in order of heroic potential<sup>12</sup>. But diseases create other types of identities as well. Maren Klawiter<sup>13</sup>

demonstrated how breast cancer at different eras generated different identities: fighter, survivor and activist. Different disease regimes offered different ways of experiencing the cancer diagnosis.

In addition to identity, the diagnosis can be a source of commercial exploitation. While many countries have legal restrictions in place in relation to the advertising of prescription pharmaceuticals, few prevent the pharmaceutical industry from promoting disease awareness. Industry's involvement in the promotion, identification and cure disease is disease branding: a way of marketing, not the therapy, rather the awareness of the condition that the therapy is supposed to cure. An effective disease-branding strategy results in sufficient public awareness such that intervention is no longer required: the patient and doctor are vigilant monitors of diagnostic potential<sup>14</sup>.

The social model of diagnosis provides a heuristic for considering diagnoses in their social and cultural context. It juxtaposes on the one side, the way in which diagnostic categories are socially framed, and on the other side, the social consequences of their attribution. Further, it places the process of diagnosis squarely in the centre, with the doctor-patient interaction, troubled as it is by the democratization of diagnostic information, the advent of self-diagnostic apps and tools, the encroachment of other professional diagnosticians and so forth. There is a circular relationship between the two sides of the model with diagnostic categories shaped by the consequences they entail, and the consequences shaped by the categories imposed<sup>2</sup>.

#### **Genetic diagnoses and the social model**

Using the social model of diagnosis as a starting point, in the pages which follow, I will point out some of what critical scholars of genetic medicine should consider as the field opens before them. As genetic explanations for ailments multiply and the science around genetic disease moves ineluctably forward, there is concomitant rise in social issues related to this new diagnostic paradigm. I will propose some thoughts for social scholars which focus on not only the way that genetics shapes diagnostic process and classification, but also on the role of diagnosis itself as a sense-making tool in health, illness and disease. This paper will propose how the social model of diagnosis may apply in the context of genetic diagnosis including identity, screening, disease definitions, and the (un)certainly of genetic determination (Figure 1).

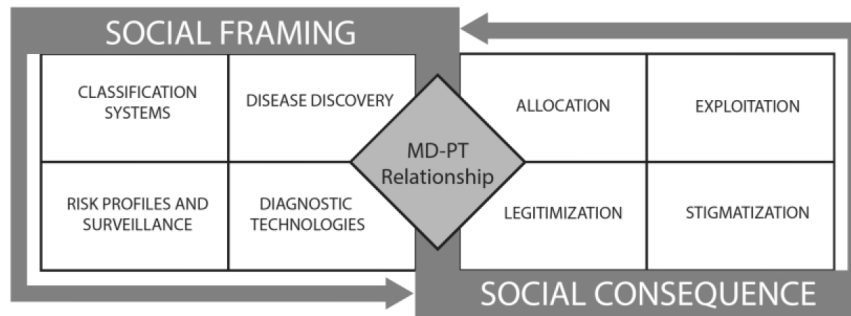
#### **Social Framing**

Social framing is a term which Robert Aronowitz highlighted in his 2008 paper in *Social Science and Medicine*<sup>15</sup>. He used this term, he explained, as a way of referring to how social forces shape what we consider as disease without falling into the trap of anti-social constructionism, understood by some to mean that there is no reality. Social framing acknowledges the material reality of illness or of disease but "...avoid[s] a few unwanted connotations sometimes associated with constructionist arguments -- a style of dated cultural relativism, a lack of common sense, and a reflexive opposition to biomedicine". The critical scholar of genetic medicine will need to consider what social frames the genetic turn brings to the understanding of diagnosis, health and illness.

#### **Believing is seeing**

An important social frame that the critical diagnosis scholar should consider as she approaches the subject of genetic diagnosis is the degree to which science replicates belief patterns, rather than disrupts them. Laqueur eloquently used historical anatomical drawings to demonstrate how social beliefs about, in his case, sexual roles, shaped the way in which scientists of an era were able to see the differences between the female and male bodies<sup>16</sup>. Depicted as an inside-out penis, the female genital organs embodied the relationship between men and women. "Ideology, not accuracy of observation, determined how they were seen and which differences would matter". While it would be facile to retort that contemporary science has "moved on," that would be a mistake. Every era imposes its own normative values on the human body, and contemporary Western medicine takes biology as the cause, and behaviours as the emerging effect<sup>17</sup>, much like Laqueur's images which propose a particular relationship between men and women from this anatomical starting point of inside-outness of the 16th century.

The critical scholar of the genetic turn should ask a number of questions in relation to this conundrum: now that we CAN see human biology in different ways, what should/are we looking for and how is this shaped by extant belief patterns and norms? Just as the phrenologist asked questions about the relationship between personalities, or behaviours and skull shapes, how can/should we justify the questions we ask about genetic profiles?



**Figure 1.** The social model of diagnosis (adapted from Jutel<sup>9</sup>).

For example, the pursuit of an obesigenic genetic profile (a genetic explanation of why some people get fat and others don't) is linked to an assumption that fatness is an indicator of poor health, which in turn is based on a long-held belief that the appearance of the individual provides a portal to the inner self, a glimpse of the hidden workings of the body, a kind of aesthetic of health<sup>18</sup>. This belief, inadequately examined, leads to a focus on obesity rather than on its causes, and is not upheld by many epidemiological studies<sup>19,20</sup>. It has been referred to as a moral panic by Campos et al.<sup>21</sup>.

### Potential patients

Further troubling the framing of diagnoses is the problematic nature of screening in which genetic diagnosis today has the potential to play an important role. I turn to the work of David Armstrong to anchor this discussion<sup>22</sup>. His seminal "The Rise of Surveillance Medicine" underlines the degree to which surveillance and screening problematises normality and transforms the individual into a compliant, always-potentially-ill subject. Armstrong describes how health has come to focus on the seemingly well individual, constantly and continuously checking for his or her disease potential. He calls this "surveillance medicine" whose "boundaries are the permeable lines that separate a precarious normality from a threat of illness".

Armstrong's sophisticated historico-sociological analysis are also captured-albeit far less critically - by elements of the medical community, concerned with over-diagnosis, "incidentilomas," and the insurgence of the medical technology and

pharmaceutical industries into the realm of diagnosis, creating an ever-watchful population<sup>23-25</sup>.

Genetic diagnostic technology offers an additional leaf to surveillance medicine. Particularly in this moment of emergence, where the generalisability of genetic information and its correlation to disease is still being confirmed, the presence of such-or-such genetic mutation or profile may be used to provide a set of probabilities about the future of an individual, even in the absence of disease. For some individuals, this provides a window of opportunity where therapeutic actions can be taken, as with hereditary diffuse gastric cancer<sup>26</sup>. In others, it does little more than describe a genetic profile without therapeutic avenue, or without even necessary presaging the outcome.

### Roads not taken

The development of a genetic explanation for diagnosis can for illness may be salutary. If it contains causal or therapeutic information with associated remedies, it may, as with the case of hereditary diffuse gastric cancer, mentioned above, save entire families. On the other hand, with each new genetic explanation, other avenues of explanation are closed down, and a foundation for the recognition and study of illness is cemented in a way which may resist later restructure.

One poignant example of how this can be problematic is in the case of fibromyalgia. This contested and oft-debilitating disease has been dismissed by many sectors, including, frequently, main-stream medicine, for its fluid diagnostic nature. A disease "for which there is no blood test" does not achieve the same legitimacy as

those with measurable features. The current diagnostic case definition is symptom based. In ICD 11, fibromyalgia is subsumed in the category of “Chronic widespread pain” and is described as “diffuse pain in at least 4 of 5 body regions and is associated with significant emotional distress (anxiety, anger/frustration or depressed mood) or functional disability (interference in daily life activities and reduced participation in social roles)”<sup>27</sup>. A genetic marker would be on the one hand helpful, and on the other, problematic for those with the symptom, but without the marker. By changing diagnostic criteria, there is a reconstruction of explanation, of population and of impact which should not be dismissed. The social impact is significant.

### Diagnostic processes

The previous pages have focused on diagnostic classification. However, as Blaxter pointed out in her seminal piece, diagnosis is at the same time classification and process. The entry into the world of diagnosis is triggered by the process: the person suffering for an ailment for which he or she would like a diagnosis. The pursuit of diagnosis will lead the patient to the doctor and at the same time will define their respective roles: doctor as interpreter and allocator of diagnosis (and its related resources); patient as compliant, patient recipient.

There are many models for the diagnostic encounter; my own leaning is towards Leder’s “diagnostic hermeneutics” which describes assembling four texts to arrive at diagnostic interpretation. These include the experiential, narrative, physical and instrumental, each of which are troubled by the genetic turn<sup>28</sup>.

The experiential text is the perception, kinesiological or other, of troubles, by the patient, brought to the clinician for interpretation. The narrative is the story that the patient tells about the troubles, augmented, altered or otherwise transformed by the clinical interview in which the doctor asks for further information. The physical examination and the “instrumental” or texts produced by diagnostic technology such as x-ray and laboratory findings, complete the picture that the doctor will then interpret.

The genetic patient on the other hand disturbs this long-held pattern of clinical hermeneutics, the patient doctor relationship, and the order of texts. The scientist and the genetic counsellor take a dominant role in the interpretive endeavor; the patient may be symptomatic

or not. The presentation may be via other family members, rather than via the individual herself.

The social scholar should be interested in how this new arrangement disturbs power relations, distribution of resources, and diagnostic impact. Like the 20<sup>th</sup> century move to “serum diagnosis”<sup>29</sup> which was robustly resisted by doctors of that era, there is a shift in how disease is understood and diagnosis delivered. Friedson underlined the importance of diagnosis in power arrangements when he wrote “Where illness is the ubiquitous label for deviance in an age, the profession that is custodian of the label is ascendant”<sup>30</sup>.

Bourret et al.<sup>31</sup> describe how post-genomic platforms contribute to tensions among health practitioners over clinical jurisdictions, but also how they conflate the issues of diagnosis, prognosis and therapy. There is a more-than-diagnosis at play here, as at the same time, the diagnosis, linked of course to prognosis, is now also predictive, suggesting the likelihood of therapeutic success.

### Social consequences

Following the social model of diagnosis, social framing of diagnostic categories and the process of diagnosing lead to social consequences, which, in turn feedback in to the way that diagnosis is framed, understood and delivered to patient by clinician. Diagnostic consequences can be salubrious, problematic or both. What is invariable is that the consequences, regardless of their type, will have an impact on how the condition is understood, explained, represented and ultimately diagnosed.

### Legitimization

That diagnosis legitimizes patient complaints is a well-known concept. The sick role, as described by Talcott Parsons, is linked to diagnosis. For the individual to have access to the sick role, they must have a recognized disorder, and comply with a prescribed treatment regimen<sup>32</sup>. The absence of diagnosis in the presence of illness is a heavy burden for the individual who does not receive the official sanction of the diagnosis. Joseph Dumit has described this in terms of “illnesses one has to fight to get.”<sup>33</sup> These are conditions which have “fuzzy boundaries,” are frequently mistaken for others, and are frequently either dismissed as being psychological in nature, or result in psychological distress, given the absence of diagnosis.

Genetic medicine may be very beneficial for those who have to fight to be ill. The genetic explanation may, in an instant, legitimize suffering which was otherwise unsanctioned.

With the power instilled in diagnosis for making sense of disorders, the possibility of genetic explanations in the absence of others fulfills an important legitimizing function. Organizations like the *Undiagnosed Diseases Network* put important weight on the role of genetics in explaining, understanding, and researching rare diagnoses. Not only do the genetics explain, they offer a sense of identity, regrouping, via genetic identity, people suffering from otherwise isolating conditions which cannot be generalized.

### Exploitation

New anxieties around the potential of genetic diagnosis makes the lay person ripe for exploitation by the marketing of genetic industry, at the same time as it places the e-scaped individual in a position to navigate medical information in ways previously unavailable. It's easy to send a scraping off to *23 and me*, or *ancestry.com* without the medical gate keeper.

Direct-to-consumer advertising about genetic risk, on the surface, increases disease awareness at the same time as it generates customers, anxious to find out the genetic truth about their future disease potential. Rather than create a calm, and measured approach to targeted conditions and populations whose outcomes may be improved by testing, it promotes referrals, demands and interpretations which may or may not be appropriate for the individual, the diagnosis or the situation. Those who are tested, and for whom variants are located, often participate in the creation of health social networks revolving around these genomic variants. They further elicit participation in the testing programmes, reinforcing and solidifying the networks and the commercial testing agencies at the same time as they create diagnostic awareness<sup>33</sup>.

However, it would appear that this relocating of knowledge has resulted in new ways of consumption, blurring as Michael Arribas-Allyon has explained, the "boundaries between consumer, producer and expertise"<sup>33</sup>. The on-line testing kit, as one point of access for the consumer, and one commercial opportunity for the biotechnology industry, creates as byproduct (or as intended outcome?) an enormous database of genetic information and genetic customers which shape another powerful commercial incentive to re-

cruit a worried subject. The biotechnology industry can then remarket its data to the scientific community<sup>31</sup>.

The genetic data ensuing from individual tests escapes ownership of the person who created it, and in a style reminiscent of what happened to Henrietta Lacks' cervical biopsy. Lacks, who died of an aggressive form of cervical cancer in 1951 became an unwitting contributor to the enrichment of medical researchers who cultivated her cells, and finally patented them, generating millions of dollars in profit as they sent them to laboratories around the world. Not knowing how genetic data will be used (or misused) creates an important ethical challenge. Genomic data has ended up in paternity suits, bone marrow transplant registers, and courts of law, with and without consent of the individuals<sup>32</sup>.

### Stigma and Blame (and eugenics)

The social model of diagnosis includes stigma as one potential consequence. Diagnosis can threaten the identity and self-esteem of the individual, as well as her potential status in social groups, or her worthiness for social roles. Being identified as ill, or potentially ill, can lead an individual to retreat, pursuing secrecy and concealment<sup>33</sup>. It can lead to discrimination on the basis of diagnosis and diagnosis potential, by employers, health insurance and even potential life partners.

With genetic diagnosis, this stigma may extend well beyond the expression of the individual case. In communities with particular genetic risk, the association with a particular genetic profile casts wider aspersions, inferring moral and problematic behaviours on the wider group<sup>34</sup>. One salient example are the Ashkenazi Jews who have a high risk for a number of lethal and debilitating genetic diseases which have been managed, in recent times, by genetic testing programmes such as the Dor Yeshorim programme<sup>35</sup>.

This community testing programme, which was designed as a means to reduce, if not eliminate the genetic disorders common in Jewish families, raises important questions about genetic responsibility, genetic couplehood, and indeed, stigma. While the intent of the programme is to reduce stigma; in one ultraorthodox group, being identified as a carrier added to, rather than decreased stigma in those so labelled<sup>36</sup>. Diagnostic stigma therefore may extend beyond the expression of the disorder to the potential to carry it. We cannot overlook the specter of eugenics, raised

by the association of biological exploration with race-related diagnostics. As with all the other social features of a classification system, what can be driven as minority inclusion and social justice may at the same time be a way of serving dominant values and stereotypes<sup>37</sup>.

Along with the genetic responsibility are complex problems of blame and self-blame, autonomy, and stigma. Identifying to whom, in a couple, a particular genetic disease cause may be assigned can result in, as fictionalized in the film, *Still Alice*, a deep sense of self-blame for what is to become a family illness<sup>38</sup>. As in *Still Alice*, the heritability of genetic diseases can, in the words of Arribas-Ayllon and his team “can potentially alter and expose the (mis)-alignment of family relations”<sup>39</sup>.

Family relations also provide opportunities for blame. The refusal to be tested for a genetic disorder may be seen as dereliction of duty, involve complex negotiation around disclosure and management of genetic knowledge. Blame features prominently, and “...is the distinguishing feature of how families manage and (not) [to] disclose their genetic status and the attendant genetic risk for ‘significant’ others”<sup>40</sup>.

## Conclusion

There is much for the critical genetic diagnosis scholar to consider in a new era which contains many new fish hooks, but also much of the same. Diagnosis continues to provide a method of generalization about individual cases that is based on consensus, linked to power, reflective of social angst and beliefs about what it is to be healthy. It promotes particular configurations of illness at the expense of others.

Post-genomic diagnosis also has the potential to open many doors, and provide explanations for what is currently unexplainable, diagnoses for what is currently undiagnosable. In so doing, it will give access to resources, identity, explanation and hopefully, therapy. Returning to Richardson, whom I cited in the introduction, being able to provide these diagnoses, to sort out the unexplained illness creates order from disarray<sup>6</sup>.

But we must be careful about what kind of order we make here, so as not to simply heap one type of confusion upon another. Despite its promise, post-genomic diagnosis is unlikely to be able to heal all ills, or explain all disease. While this short commentary just scratches at the surface of what post-genomic diagnosis begs the social scholar to consider, it does provide a starting point for critical questions. Each advantage brought to the fore reveals at the same time, a potential disadvantage. Behind each putative empowerment resides an exercise of power.

I have not offered theoretical perspectives from which to consider these topics. This has been done by others in the context of geneticization, biosocialization, bricolage and more... The critical social scholar will also need to think about what theoretical frames illuminate and conceptualize the array of factors that genetic diagnosis and its related biotechnologies raise.

A critical scholar cannot look at any of the products of genomic medicine without considering the full picture. If we are to capture the promise of genetics, we must at the same time recognize its downsides. However diagnosis is formulated, be it via genetic explanations or microbiological ones, they are the product of social discovery, negotiation, and consensus. They are dispensed by social agents, vested with the power to label health and illness, and they have social consequences.

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