



## Social implications of genomic medicine: Is medicine ready?

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### EDITORIAL

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In last month's issue of the *AMJ*, Jon Cornwall and his colleagues asked the salient question: is society ready for advanced genomic medicine (AGM)?<sup>1</sup> This sociologist of diagnosis follows up with a question of her own: can medicine accommodate the social aspects of genetic diagnosis? The advent of genetic testing brings to the fore the important social role of diagnosis in conceptions of health and illness, a point on which I will dwell here.

Diagnosis is present in places we do not expect. While the World Health Organization (WHO) clearly rejects the idea that diagnosis (or rather its absence) defines health,<sup>2</sup> Western medicine stubbornly resists considering health outside of diagnosis. Genomic medicine reinforces this view. The idea that ostensibly healthy individuals can gain insights into their inner workings to look for disease potential even in the absence of symptoms anchors this thinking. This approach predates AGM by more than a century; the periodic health examination in the asymptomatic individual was first touted in the 1860s.<sup>3</sup>

If we can consider disease a silent, unobtrusive potentiality ready to rear its ugly head in the seemingly healthy when least expected, this changes how we think of health. It transforms the healthy into the always-potentially-ill, and puts in place a rationale for surveillance medicine and, of course, for AGM.

As David Armstrong writes, surveillance medicine extends diagnosis to a space *before* disease, with risk factors pointing to "a potential, yet unformed, eventuality".<sup>4</sup> This elevates the screening event to a place previously held by diagnosis alone. The genetic report provides the rationale for consultation, treatment, or lifestyle modification in one who would have thought him/herself to be healthy. Western medicine tends to talk about this as a problem of the "worried well", the layperson who should stop fretting, when we could just as easily point the finger at an omniscient medicine which sees knowledge as the first step in the control of human destiny.

This is not to say that diagnosis is unimportant: diagnosis serves an important social role.<sup>5</sup> It separates lay from professional: after all, it is in the pursuit of a diagnosis (i.e., what's wrong with me?) that a person makes an appointment to see the doctor. The diagnosis "organises" symptoms, as it provides an explanation for findings, outlines a prognosis, determines options for treatment, and in some cases, gives patients an identity. Diagnosis, in general, is key to the allocation of resources, whether it be the prescription, the sick leave certificate, the insurance reimbursement, or the disability status. Genomic medicine similarly offers explanation and treatment. One needn't look further than BRCA1 (breast cancer early onset) and its associated pre-emptive treatments: we've all read about Angelina Jolie, who had a preventive double mastectomy. But there is the less publicised HLA-B27 blood test to explain the spondylitis, the iritis, and, of course, many more disorders.

However, diagnosis can result in poor social outcomes as well. Some diagnoses stigmatise, others terrify. HIV, syphilis, psychosis, gout, or even depression can produce undesirable social outcomes, including prejudice, marginalisation, and disadvantage. Not only this, even the simple utterance of some diagnoses can shift one's sense of self, dividing life forever more into an indelible "before" and "after" where even though nothing has changed in the biological process giving it a name has changed everything.<sup>6</sup> How would you feel if you learned



you had “the C word”? The impact of its naming so great, we can’t find a way to pronounce all of its letters.

Genomic medicine tests us, and disturbs the social role of diagnosis, blurring lines which were previously (possibly) cleaner. Patients come to the doctor, diagnosis in hand, primed by the industry that delivered the **23andme** or the **Gentle**.<sup>7</sup> The intervention of the genetic testing industry shifts the starting point in the medical consultation. Plagued by the changes wrought by AGM on her relationship to her patient, the clinician could reflect on the social role of diagnosis to find both explanation and solution.

Clinicians can no longer see lay people as “patients” who endure our treatments and follow our instructions. They come to the consultation armed with information that would have, only a few decades ago, been inaccessible. This information gives the clinician a valuable opportunity for figuring out the person, rather than focussing on disease. What does this information tell you about the person in your rooms, and how can this help you to identify the best way forward?

If society needs to be prepared for AGM, so, too, do clinicians. In the recently published textbook “Social Issues in Diagnosis,” we have proposed the CLASSIFY mnemonic to assist the clinician to consider the social factors which are involved in diagnosis, and which can certainly help the clinician navigate the shifting social role of diagnosis in the era of AGM.<sup>8</sup>

## Conclusion

Diagnosis is much more than just a pathophysiological process. It is a socially negotiated classification process for assigning labels that are negotiated and agreed upon (i.e., socially framed). It results in social consequences that can have an even greater impact on the patient than the disease itself. Clinicians: for your words to be heard, your instructions followed, you must be aware of, and heed the social role of diagnosis.

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## PEER REVIEW

Externally peer reviewed.

## CONFLICTS OF INTEREST

The author declares no conflict of interest.