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Afterword:

Personal Genomics

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Van Booy's story depicts a world in which 'disease' is defined in terms of expected death, as something that is prescribed and pre-determined far in advance of any immediately experienced — or real world — suffering or 'illness'. And as we have seen with the story's characters, such predictions are not without real effects on how one lives and feels about one's life and health.

So as to paint a picture of where biomedicine presently stands, I will briefly trace the developments that have unfolded over the last sixty years, locating the current capabilities of molecular-genetic medicine in its correct scientific, cultural and epistemic genealogy. As a potentiality then, genetic medicine may be said to have begun with the elucidation of the structure of DNA, achieved by Watson and Crick in Cambridge in 1953. They revealed DNA's double-helical structure and with that intimated a hypothetical function for it as the self-replicating substance of which genes are made. Following on from their discovery, the 1970s saw rapid advances in the ways that DNA could be precisely manipulated, and the broader field of molecular biology gave birth to recombinant DNA technology, revolutionising the basic biosciences by allowing DNA to be routinely mutated and artificially engineered — rendering possible the cloning and recombination of genes in ways that had not previously been possible. DNA had thus become a malleable and fungible material, and scientists had developed the tools to work it with dexterity. With further extension of these manipulative capacities, 'biotechnology' became one of the fastest growing industries in the 1980s; and in the 1990s — in the lead-up to the sequencing of the human genome — there was much speculation about the implications of uncovering the human genetic code and what it could make possible. So much so that researchers in several countries made plans to create genomic databases or ‘gene banks’, built up from citizens' genetic material — provided by volunteers, patients and the general public. And in several countries (including Britain, Sweden and Iceland), researchers promised that these data would reveal the genetic basis of many diseases, and that this knowledge would eventually lead to new diagnostic and therapeutic products. In 2001 then, with the first publication of the sequence of the human genome, there was great hope (and hype) around this promise of developing cures for many of the diseases afflicting mankind...

Running in parallel to these basic scientific developments in molecular genetic technology, since the 1950s there has also been a transformation in the way that human health is both perceived and detected in the clinic. Pre-WWII, disease was something that generally presented itself as a disruption in experienced daily wellness, where patients typically went to their doctors complaining about an experienced problem — they felt unwell or sick, and could identify the ailment by its effects on how they felt. With the advent of biochemical analysis and molecular diagnostics, disease recognition underwent a tectonic shift. From once being a mostly qualitative system of symptom presentation and self-directed description — reported by the patients' own experience of illness — there was born a more quantitative regime of risk detection, where patients became classed as being at risk, possibly long before they felt unwell. Nowadays in the 'developed world', patients regularly attend medical check-ups, and are indeed commonly determined to be at risk from diseases that have yet to show any sign of existence. Consider for example the crucial bio-indicators of high blood pressure, blood plasma cholesterol or blood glucose metabolism; these are all commonly employed as biomarkers of being at risk for the potential development of heart disease, diabetes and similar diseases of the so-called Western lifestyle. Screening for these biomarkers, often far in advance of any experienced
illness, now forms the basis of a standard medical exam. Being classed as having ‘hypercholesterolemia’ or ‘high-blood pressure’ — invisible and unfelt indications that may or may not lead to the development of a specific disease — one is thus classed as being ‘at risk’. While these preemptive strategies have greatly reduced the incidence of premature deaths from what are now treatable and gratefully avoidable life-curtiling diseases, they also bring with that enhanced longevity a preventative medical logic, with the potential for an extension of the experience of ‘illness’ into the domain of what was once a ‘healthy’, if at risk, life. Indeed, symptomless biomarkers can now be experienced as a disease in itself or as its looming onset; and while the benefits of some preemptive treatments are not in doubt, such movements also result in global pharmaceutical and therapeutics corporations aiming to progressively lower the scientifically published thresholds for being ‘at risk’ so that more people can be medically classed as at risk, thereby increasing the pool of candidates available for medication, and with that, boosting their long-term prescription sales. Patients may therefore now experience illness as a risk category — and be medicated for that risk — far in advance of any symptomatic experience; and precisely because of this transformation in ‘illness experience’, many people now take precautionary medications in order to prevent illness that would actually only occur in a small percentage of the total drug-taking cohort. These patients are medicated precisely because they all fall into an identifiable, quantitative risk category that is generally and statistically defined, but subjectively indeterminable.

This transformation in illness experience is critical to the way we approach Van Booy’s dystopia, which is the ultimate and grotesque extension of this type of preventative logic: a world where one’s medical fate becomes an inescapable burden that one is expected to live in accordance with. A prognosis for medical diagnostics that marks people with a certain probability of being alive at a particular juncture in the future — presumably based on how long other people considered to be in similar demographic and genetic groups have lived for — configures a world where people live their lives by relating their lived experience of the world to the most probable arrival time of their death, defining ‘life’ then as a morbid temporal state of being that dialectically affirms its very essence from ‘not dying’.

One problem militating against such a scenario is the fact that diseases are usually multifactorial and can rarely be identified as the consequence of a single gene in a perfectly predictable way. There are some exceptions to this, particularly with rare ‘Mendelian’ diseases such as Huntington’s, for example. Huntington’s is an inherited neurodegenerative disease that is already somewhat predictable (the child of a person with a copy of the mutated huntingtin gene has a 50% chance of developing the disease). Such diseases, with the possibility of their early detection and with their high probability of leading to the disease, do indeed raise ethical questions, particularly as to whether one should know early in life if one is likely to develop the disease. But these ‘Mendelian’ diseases are by far the exception, and most of the Western world’s most common diseases cannot be causally linked to a single specific gene.

So, given the current capabilities of DNA- and genomic technologies, could a person really have their personal genome analysed and correlated with statistical probabilities for a whole population so as to come up with a reliable index of healthiness, projected longevity, or even an estimated time of death? Well, steps have already been taken to attempt to make whole population genome analysis a powerful predictor of health and disease. Indeed, ‘personal genomics’ is a growing industry, offering people the opportunity to have their own genome sequenced and have the results analysed in order to identify individual differences in ‘normal’ and ‘disease-associated’ regions and gene sequences. Such services are now commercially accessible to ‘healthy’ individuals, and the US-based company Illumina, for example, offers a personal genome sequencing service at a price of $19,500 (as of October 2011), with discounts offered to groups of five or more. Individuals identified to have variations in certain genes could possibly be
associated with a higher 'risk' of developing certain diseases when compared to individuals without such variants, but these kinds of associations are unlikely to have any powerful or reliable predictive capacity on an individual basis alone. Indeed, since it is rarely a one-gene-per-disease relationship, personal genomics research aims in the future to map the interactions of many or possibly even all of the genes in the genome in order to map their complex, global interactions in the possible cause – or pre-disposition in the development – of diseases. Such efforts demand the coupling of genome analyses to long-term family medical records, making the task a long-term project. This strategy has yet to produce any conclusive evidence as to its effectiveness and reliability, but such a breakthrough would certainly mark an epistemic rupture and a tremendous overhaul of the capacities of genetic medicine. Having said that, it will likely remain that, more than genetics, individuals are far more susceptible to the varying and un-mappable influences of chance, changing environment, lifestyle, and possibly above all, individuals’ own agency in the ultimate outcome of their personal health and illness.

The story’s parodic future world, in which health and illness are categorically predetermined by one’s genetic code is therefore both amusing and shocking. While no statistical prediction of longevity could ever stake a claim to a pre-destined fate that is absolutely ‘true’ for an individual, the story still manages to shock because it offers a glimpse of a world where certain freedoms are taken away; namely the freedom to determine and control one’s own health; the perception of one’s health; and how one ‘feels’ about one’s own future. In this era of post-genomic mythology – despite the objective limits of genetic-medicine to predict real, lived experience – claims to genetic fate may well end up functioning as a mysterious but compelling imperative – an invisible threat one would only foolishly ignore.

Story Time

Sean O’Brien

June 1st

It is early summer. There is a metalled single-lane road that turns into a track and tilts downhill into the woods. Once I am in the woods the water becomes visible, a great reach of it with a scatter of islands. I suspect – I have no map – that the large forested area a quarter of a mile away may itself be an island, which in turn provokes the possibility that where I stand might be an island too. But this cannot be so, surely. I seem to remember that I arrived overland.

At the foot of the track is a clearing set back a little way from the water’s edge, and in the clearing stand a whitewashed farm cottage and one or two half-derelict looking sheds. A tabby cat slinks away at my approach and watches from behind a birch tree.

Although it is summer a curl of smoke rises from the cottage chimney. The door is open. I knock but no one answers. Hesitantly I put my head round the door. Beyond lies the dim kitchen – the heavy varnished table with its loaf and board and knife, a family Bible with a place marked deep in the Old Testament, the cavernous fireplace giving out slightly too much heat, the ugly dresser with its ugly carved flower-patterns. The whole apparatus of nameless provincial life, halfway between Balzac and Perrault. The clock, in pride of place on the dresser, ticks. It is just before noon. I look at the marked verse of scripture: yea, also the heart of the sons of men is full of evil, and madness is in their heart while they live, and after that they go to the dead.