All that glitters ...

Victor Grech

Introduction

The full human genome sequence (via the Human Genome Project) was mapped and published 15 years ago, a 10 year project that cost circa \$30bn.\(^1\) Since then technological advances and cheap computing power have markedly accelerated genomic sequencing at a reduced cost, to under \$500. For example, the company Oxford Nanopore has taken sequencing to point-of-care status by developing a hand-held reader that can sequence genetic material in minutes.\(^2\) The applications will not be just for healthcare but also, for example, to establish the provenance of foodstuffs, the presence of dangerous microbes and crime scene DNA analysis.\(^2\)

This is possible as the Nanopore reader squeezes individual DNA strands though a nanopore (an individual hole) and then proceeds to read the DNA sequence electrically. It has been speculated that within the next 10 years, everyone will be genetically sequenced at birth with the corollary that preventative steps can be taken early vis-à-vis genetic predispositions to diseases.²

The combination of this type of data analysis, along with other variables, may further improve the prevention, diagnosis and treatment of diseases. For example, researchers at Brigham and Women's Hospital in Boston, Massachusetts used such data combinations to highlight an increased risk of developing type 2 diabetes among shift workers.³ A concise and elegant summary by Janssens and van Duijn states:

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Genomics research will substantially increase our understanding of disease pathogenesis, particularly through the identification of novel disease pathways and new biomarkers ... one of the major promises is that these advances will lead to personalized medicine, in which preventive and therapeutic interventions for complex diseases are tailored to individuals based on their genetic profiles ... Yet, the etiology of complex diseases is essentially different from that of monogenic diseases. and hence translating the new emerging genomic knowledge into public health and medical care is one of the major challenges for the next decades.⁴

What if?

Science fiction has explored the "what if" scenario of universal genetic profiling, and the dystopian film GATTACA does precisely this, with the added twist of the additional availability of embryo genetic selection so as to ensure that offspring possess the best hereditary traits of their parents.⁵

The film commences with a famous quote: "As night-fall does not come at once, neither does oppression...It is in such twilight that we all must be aware of change in the air - however slight - lest we become victims of the darkness" by Justice William O. Douglas (1898 –1980), the longest-serving justice in the history of the Supreme Court.⁶

In this fictional, not very distant future, universal genetic profiling at birth had led to the creation of a genetic database that is used to classify individuals. Individuals may be antenatally enhanced as explained by a geneticist to a prospective couple:

You've already specified blue eyes, dark hair and fair skin. I have taken the liberty of eradicating any potentially prejudicial conditions - premature baldness, myopia, alcoholism and

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addictive susceptibility, propensity for violence and obesity ... You want to give your child the best possible start. have Believe me, we enough imperfection built-in already. child doesn't need any additional burdens. And keep in mind, this child is still you, simply the best of you. You could conceive naturally a thousand times and never get such a result...Is there any reason you'd want a lefthanded child? ... Some believe it is associated with creativity, although there's no evidence. Also for sports like baseball it be can an advantage...he's going to be at least a head taller than you.

The parents want even more, the imposition of what is to them, a normative heterosexual inclination: "we were hoping he would get married and have children. We'd like grandchildren." The geneticist reassures them "I understand. That's already been taken care of." He also however adds:

Now you appreciate I can only work with the raw material I have at my disposal but for a little extra...I could also attempt to insert sequences associated with enhanced mathematical or musical ability...I have to caution you it's not fool-proof. With multi-gene traits there can be no guarantees.

In this future, genetic discrimination is illegal, but genotype profiling is still used to identify the genetically enhanced for professional employment, while normal conceptions are relegated to menial jobs, and this is precisely what the storyline revolves around, along with the possibility that the individual may actually exceed his/her expected potential. This is evidenced in GATTACA's poster with the statement that "there is no gene for the human spirit" (figure 1)

Inevitably

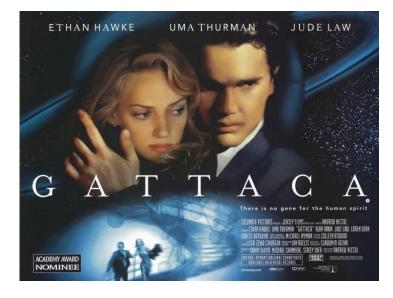
One can easily take this further and imagine the repercussions, say, of individual insurance prospects, both life and health, were disease predisposition made available to insurance companies. Discrimination would be almost

inevitable, with highly negative personal economic consequences, not only in insurance, but also in occupation and so on. Truly, "before implementation in health care, all applications of genetic profiling need appropriate evaluation to assess whether the predictive value is sufficient e.g. to improve population health or to improve the efficiency or quality of health care".

Furthermore, the active implementation of genetic antenatal embryo changes no longer lies solely within the demesne of science fiction. CRISPR (Clustered Regularly Interspaced Short Palindromic Repeats) technology forms the basis of a technique that can specifically change genes within organisms.⁷⁻⁸ The dystopia that is GATTACA thus lies within our grasp.

Therefore in this field too, all that glitters is not gold, and once this particular genie emerges from its bottle, it will be well-nigh impossible to avoid these consequences.

Figure 1



References

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Cover Picture:

"Valletta Nocturne'
Oil on canvas with palette knife
By Victor Grech

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