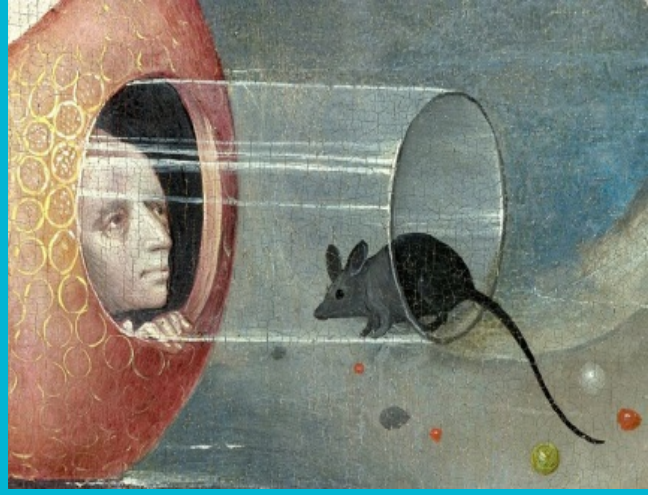
Erasmus
School of
Philosophy

Personalised Medicine



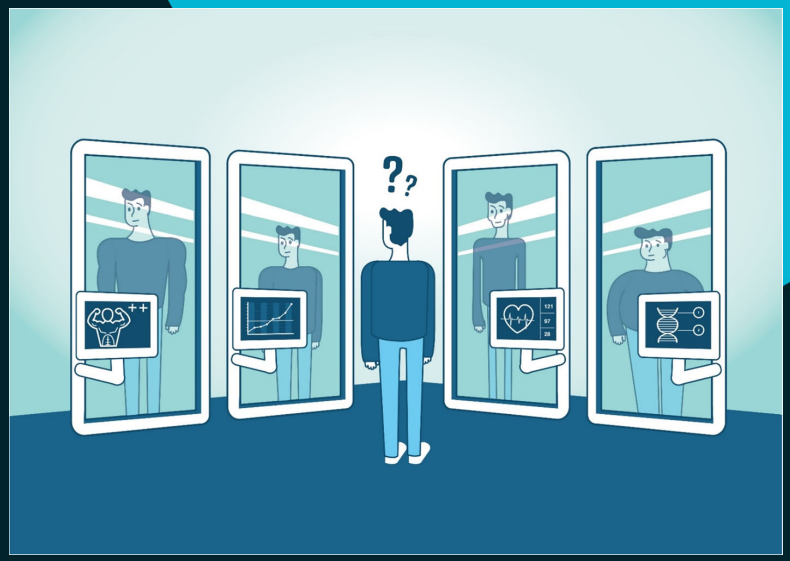


Fig. 1 Data mirrors provided through personalised healthcare. The funhouse mirror. The I in personalised healthcare. Illustration by Lisa Renee https://www.lizarenee.nl/

ARRIBA Symposium
Personalised medicine 22 3 2021
Prof. Dr. Hub Zwart

Erasmus University Rotterdam

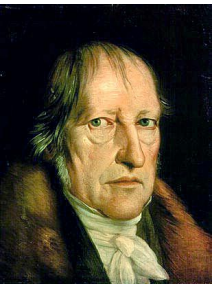
Erasmus

Personalised medicine

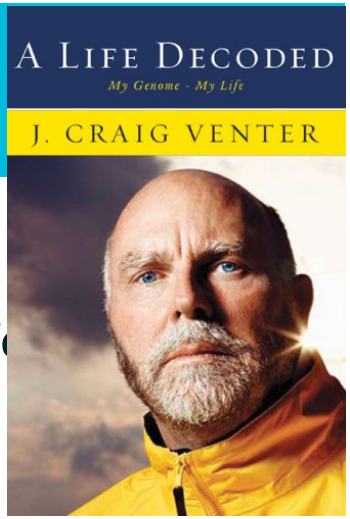
- Timely and topical (Zeitgeist)
- Health (biomedical)
- How we see ourselves (Know thyself; oracle)
- Self-management, empowerment, power to the patient
- Citizen science
- SWOT analysis

Philosophy of Science

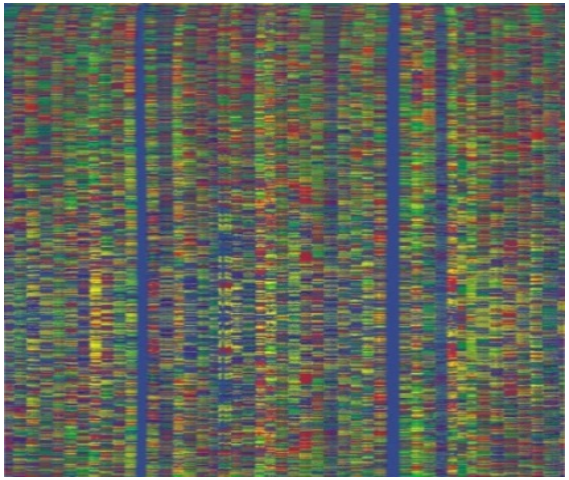
- Addressing questions raised by science in dialogue with science
- *Diagnostics of the present*



June 26, 2000: the *We*-genome



- Human Genome Project (HGP):
- *Human Reference Genome* (2004) – Composite genome (W genome)
- Personal genomes of genomics celebrities



15 February 2001

nature

articles

Initial sequencing and analysis of the human genome

International Human Genome Sequencing Consortium*

* *A partial list of authors appears on the opposite page. Affiliations are listed at the end of the paper.*

The human genome holds an extraordinary trove of information about human development, physiology, medicine and evolution. Here we report the results of an international collaboration to produce and make freely available a draft sequence of the human genome. We also present an initial analysis of the data, describing some of the insights that can be gleaned from the sequence.

The rediscovery of Mendel's laws of heredity in the opening weeks of the 20th century[1–3] sparked a scientific quest to understand the nature and content of genetic information that has propelled biology for the last hundred years. The scientific progress made falls naturally into four main phases, corresponding roughly to the four quarters of the century. The first established the cellular basis of heredity: the chromosomes. The second defined the molecular basis of heredity: the DNA double helix. The third unlocked the informational basis of heredity, with the discovery of the biological mechanism by which cells read the information contained in genes and with the invention of the recombinant DNA technologies of cloning and sequencing by which scientists can do the same.

The last quarter of a century has been marked by a relentless drive to decipher first genes and then entire genomes, spawning the field

coordinate regulation of the genes in the clusters.

- There appear to be about 30,000–40,000 protein-coding genes in the human genome—only about twice as many as in worm or fly. However, the genes are more complex, with more alternative splicing generating a larger number of protein products.
- The full set of proteins (the 'proteome') encoded by the human genome is more complex than those of invertebrates. This is due in part to the presence of vertebrate-specific protein domains and motifs (an estimated 7% of the total), but more to the fact that vertebrates appear to have arranged pre-existing components into a richer collection of domain architectures.
- Hundreds of human genes appear likely to have resulted from horizontal transfer from bacteria at some point in the vertebrate lineage. Dozens of genes appear to have been derived from trans-

November 9 1989

nature

NEWS

HUMAN GENOME PROJECT

Dealing with the data

Washington

REPRESENTATIVES from the National Institutes of Health (NIH), the Department of Energy (DOE) and Genbank, the national sequence database at Los Alamos National Laboratory, are meeting here this week to draw up recommendations for a new interagency informatics task-force to oversee national policy on handling the data created by the mapping and sequencing of the human genome. The task-force will put proposals on both policy and technical issues before the NIH genome advisory meeting due to be held in December.

The deputy director of the NIH human genome office, Elke Jordan, warned the audience at an international conference on sequencing at Wolf Trap, Virginia, last month that unless coherent policies plans are developed now, "we are going to run into a big problem later on".

Central to the debate is the role of journals in publishing sequence data. At present Genbank shares with the European Molecular Biology Laboratory (EMBL) in Heidelberg, West Germany, responsibility for searching journals and storing genetic sequences. But Paul Gilna words, try to find "new ways to give brownie points" for research, since the human genome project is likely to create lengthy sequences that will not be publishable in the more traditional journals. Mark Pearson, director of molecular biology at Dupont and head of the NIH informatics committee, says that researchers will increasingly include in their curriculum vitae accession numbers given to them when they deposit data in the databanks.

But with this, he warns, will come come "all the perils and problems" of counting accession numbers and assigning quality weightings to them. Some researchers at the meeting balked at the suggestion that the data from their research might not merit publication. When Graham Cameron of the EMBL databank suggested that sequencing would become a routine task and that only interpretations of the data would merit publication, Sidney Brenner of the Medical Research Council in Cambridge, where researchers are sequencing the genome of the nematode, quickly retorted that if that was the case then perhaps those at the databanks would care to do the sequencing.

Christine McGourty

■ Databases have prompted correspondence this week, page 114.

Ethical matters

Paris

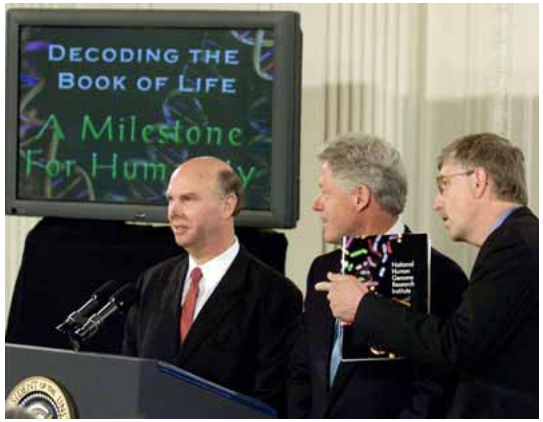
AT a four-day European meeting on Genetic Heritage and Human Rights held in Paris at the end of last month, a series of workshops were devoted to discussions of research on the human genome and its implications, predictive medicine, *in vitro* fertilization and biotechnology, from both a scientific and an ethical perspective. Participants formulated recommendations dealing with, among other subjects, the freezing of embryos, the psychological dangers of screening for as-yet incurable monogenetic diseases or diseases that appear only later in life, genetic 'fingerprinting', the need to conserve genetic diversity through gene banks and the existing dangers of eugenic choices. A summary of each of the 13 workshops and their recommendations will be published at the end of the year, followed at a later date by more extensive proceedings. Peter Coles

INFECTIOUS PERESTROIKA

November 9 1989

HGP

- “the working *blueprint* of the human race”

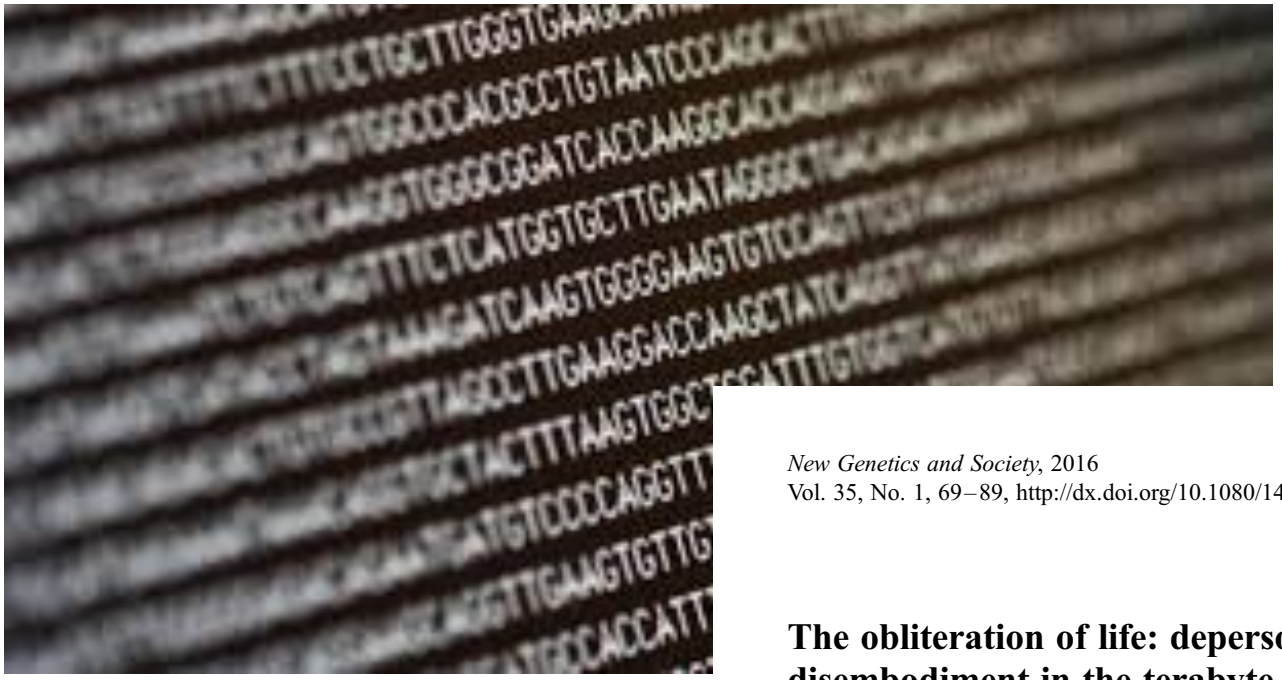


Promises

- “Our children’s children will know the term cancer only as a constellation of stars”

Erasmus

Obliteration



New Genetics and Society, 2016
Vol. 35, No. 1, 69–89, http://dx.doi.org/10.1080/14636778.2016.1143770

Routledge
Taylor & Francis Group

The obliteration of life: depersonalization and disembodiment in the terabyte era

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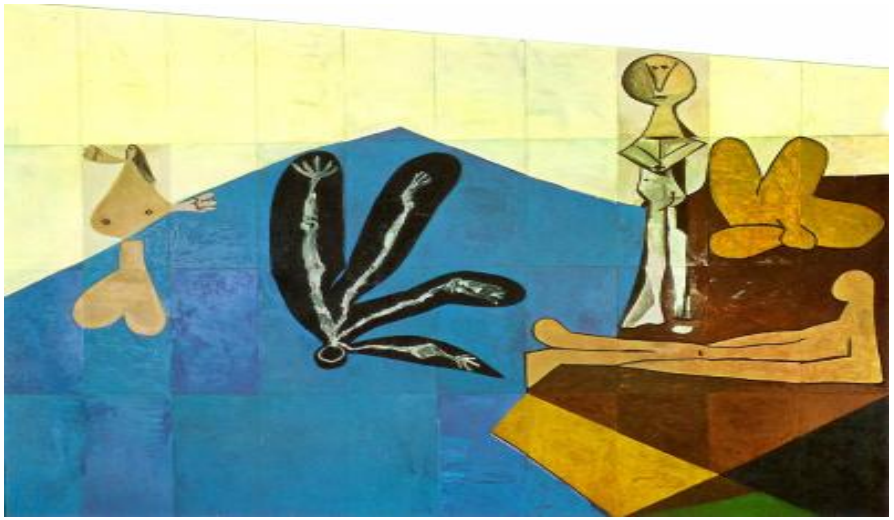
(*Received 5 April 2015; final version received 14 January 2016*)

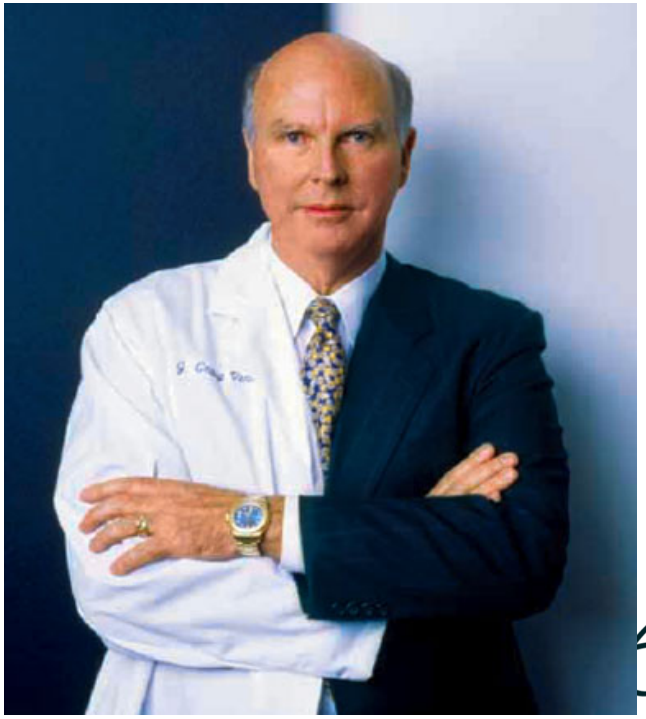
Post-genomics allegedly allow us to become the "managers" of our own health. And yet, human individuality seems to dissolve into massive data streams. What is the fate of the human subject in the terabyte age? The Human Genome Project already resulted in personalizing and depersonalizing trends, exemplified by two types of genomes: the anonymized Human Reference Genome versus the personal genomes of genomics celebrities. This ambiguity is radicalized by

Picasso: Fall of Icarus



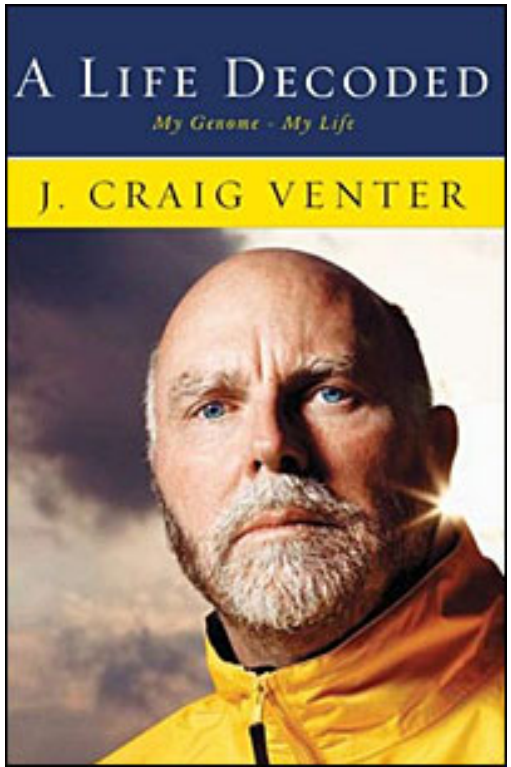
Craig Venter

- “The order of the letters in my DNA provides a recipe to make Craig Venter..”

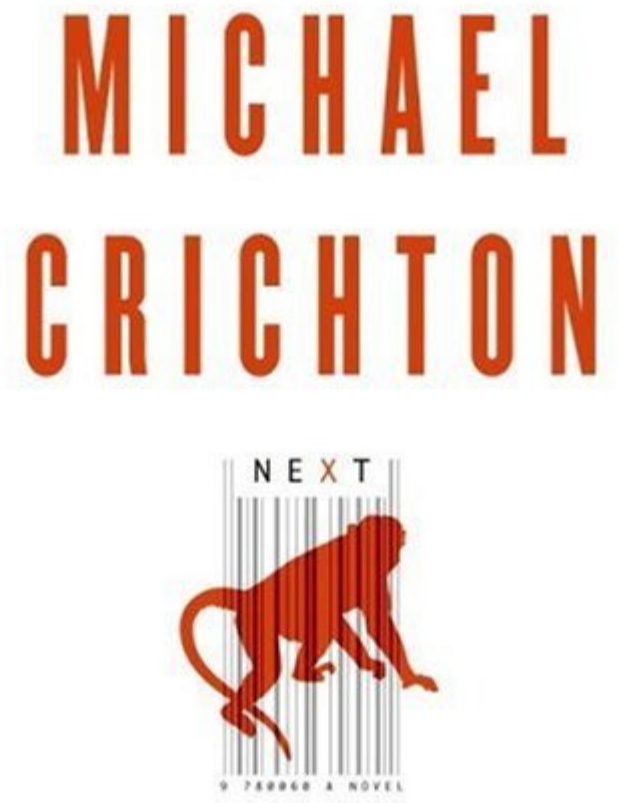


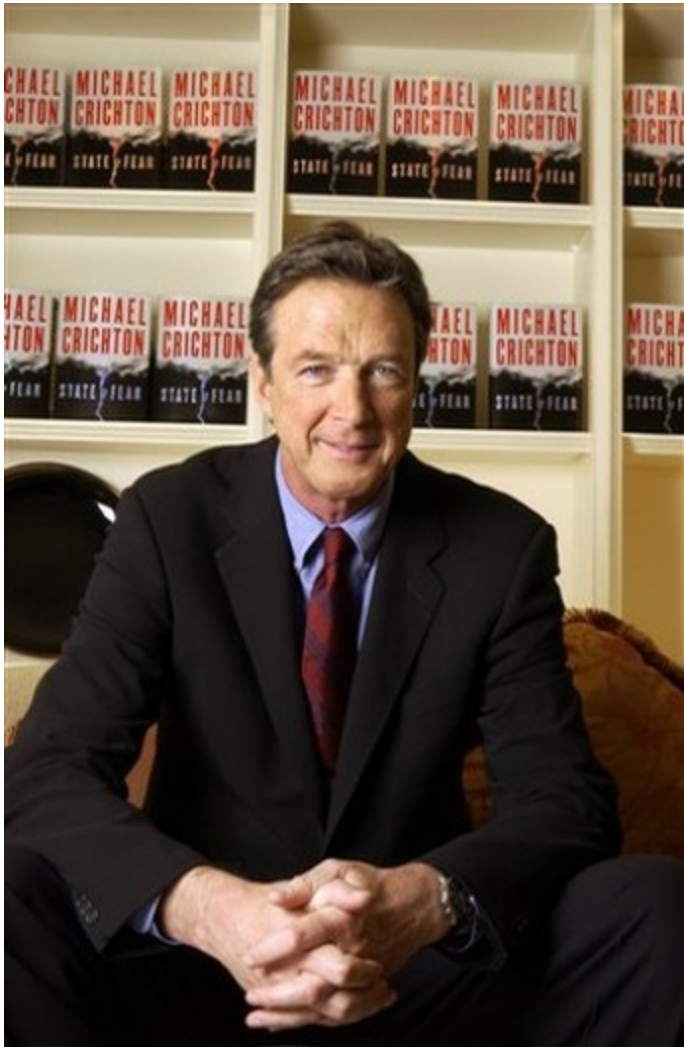
A life decoded (2007)

In separate boxes dedicated to his sequenced genome, he especially focuses on genes that are associated with behavioural characteristics such **as thrill-seeking behaviour, ADHD and the ability to cope with stress**.



Michael Crichton (1942 – 2008)



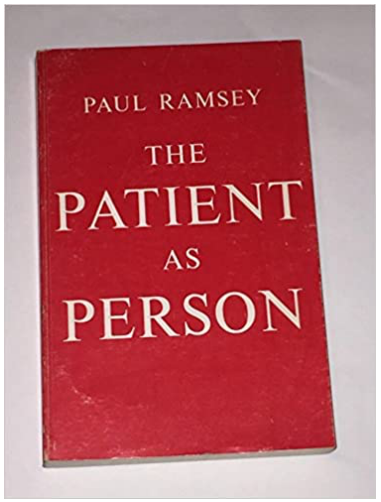


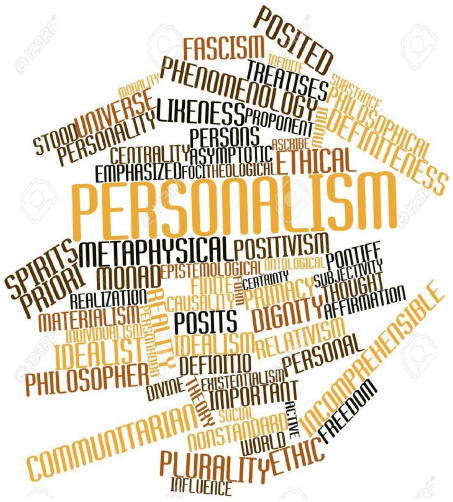
Next (literary laboratory)

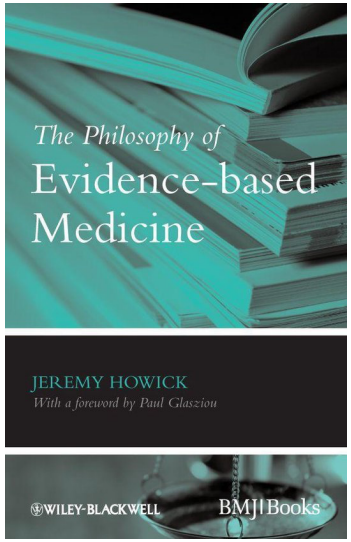
- Maturity gene
- Novelty- (or thrill-seeking) gene
- Sociability gene
- Infidelity gene

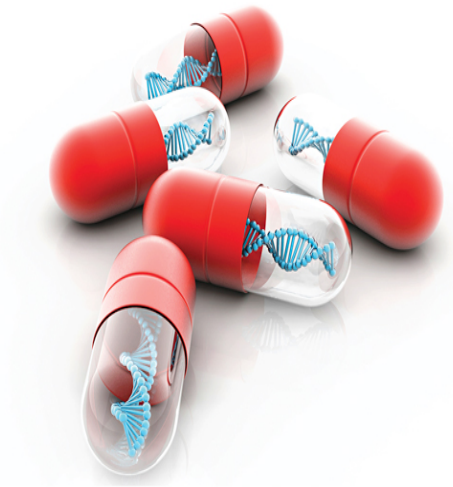
Three stages

- 1970s: The patient as a person – personalism (Dorothy Day, Karol Wojtyła) – Back to the beginning: Hippocratic medicine (N=1, clinical gaze, personal encounter)
- Evidence-based medicine (N=many) – de-personalisation (negation)
- Present: Personalised medicine (N=me) – re-personalisation







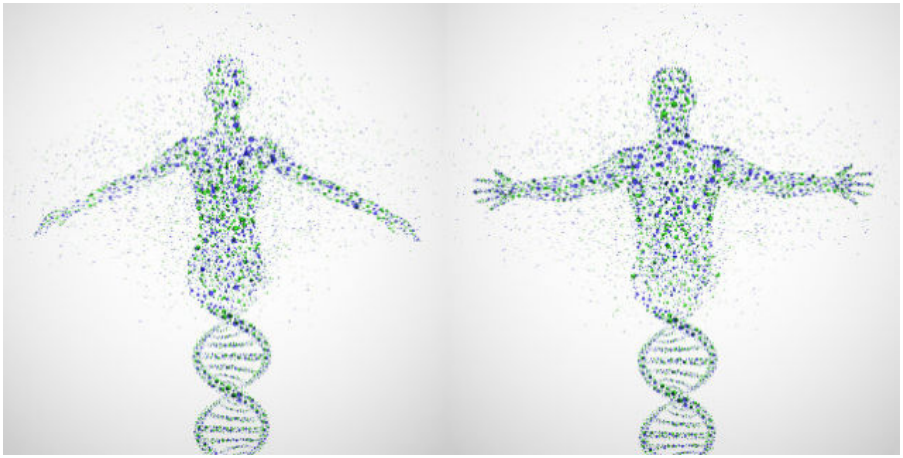


Personalised Medicine

- Personalised medicine, precision medicine, pharmacogenomics, prediction, participation

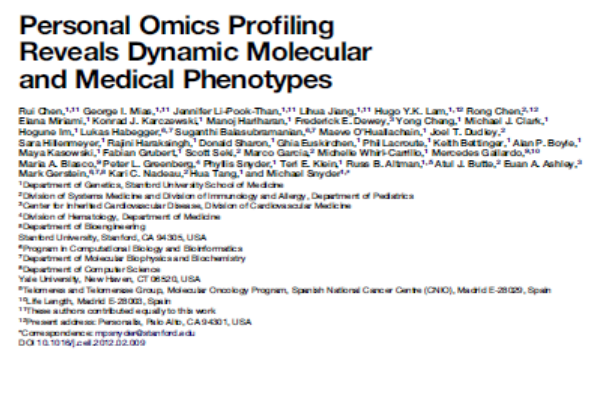
President Obama

- “You can match a blood transfusion to a blood type. That was an important discovery,” Obama said Friday during a speech at the White House about the initiative. “What if matching a cancer cure to our genetic code was just as easy, just as standard? What if figuring out the right dose of medicine were as simple as taking our temperature? That’s the promise of precision medicine.”



IPOP: the Snyderome

- Integrated Personal Omics Profiling of a single individual, 54-year male closely monitored during 14 months; combining deep sequencing with > 3 billion measurements of molecules
- High resolution molecular self-portrait
- The Snyderome



Personal Omics Profiling
Reveals Dynamic Molecular
and Medical Phenotypes

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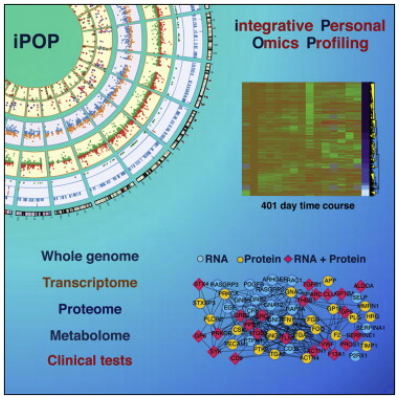
DOI 10.1016/j.cell.2012.02.009

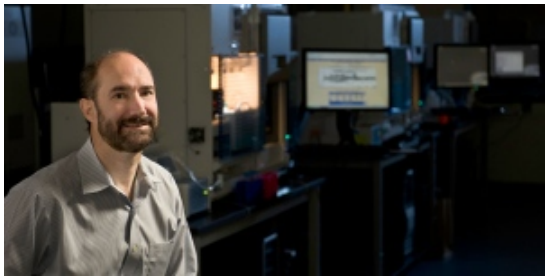


Measure everything

- Freud: “We instruct the patient to report everything, however *disagreeable*, *indiscreet*, *unimportant* or *irrelevant it may seem*”.
- iPOP: measure and report everything, data of any kind must be included. Especially waste products (urine, faeces, bodily ‘litter’) may contain highly valuable information about what is going on under the surface

Snyderome Objective

- Via high resolution continuous self-monitoring, human individuals will become the proactive managers of their own health condition. Longitudinal multi-omics analysis will allow ‘us’ to take medicine into our own hands
- Measurements of thousands of factors can be integrated through devices such as iPhones and compared with big data references, available 24/7 at open-source repositories (vast science clouds), after which they can be translated into every-day options (diet, exercise, etc.).



Paradox

- Practices of the Self, self-management, allowing individuals to shape their own lives?
- Big Data repositories provide reference data (standards for normality)
- Molecularised super-ego, ‘voice of conscience’ of the terabyte age, the Big (digital) Other. On a daily basis, computer ‘monitors’ will be telling individuals to change their lives to optimise somatic functioning and live up to normalcy standards, so as to mitigate the impacts of unhealthy life-styles and ageing.

Data hermeneutics: skills required to interpret personalised data

Vegter *et al. Life Sciences, Society and Policy* (2021) 17:1
https://doi.org/10.1186/s40504-020-00108-0

Life Sciences, Society and Policy

RESEARCH Open Access

The funhouse mirror: the I in personalised healthcare

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Abstract

Precision Medicine is driven by the idea that the rapidly increasing range of relatively cheap and efficient self-tracking devices make it feasible to collect multiple kinds of phenotypic data. Advocates of $N = 1$ research emphasize the countless opportunities personal data provide for optimizing individual health. At the same time, using biomarker data for lifestyle interventions has shown to entail complex challenges. In this paper, we argue that researchers in the field of precision medicine need to address the performative dimension of collecting data. We propose the fun-house mirror as a metaphor for the use of personal health data; each health data source yields a particular type of image that can be regarded as a 'data mirror' that is by definition specific and skewed. This requires competence on the part of individuals to adequately interpret the images thus provided.

Keywords: Precision medicine, Digital health, Self-tracking, Wearables, Data double, Eccentricity, iPOP, Ethics, Embodiment, Self, Personalised healthcare

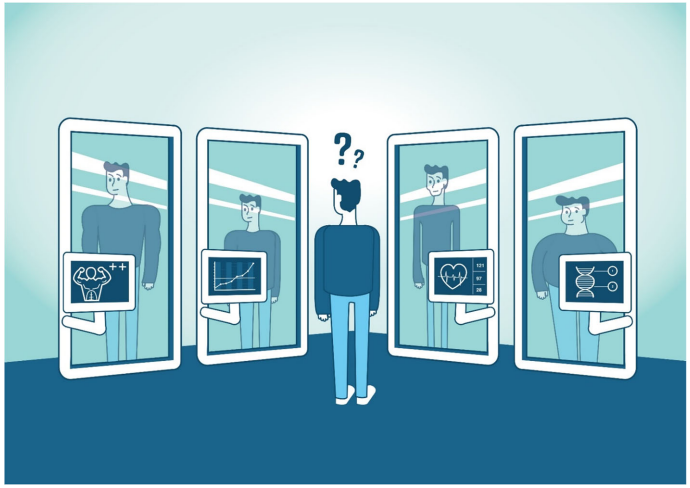

Fig. 1 Data mirrors provided through personalised healthcare. The funhouse mirror: The I in personalised healthcare. Illustration by LizaRenee https://www.lizarenee.nl/

Living labs

- Rapidly expanding -omics fields promote molecular profiling;
- The increasing amount of relatively cheap and efficient devices to collect data, such as the Apple WatchTM;
- Genomics labs + living labs, life world
- wellness study : for each measurement in an individual that is outside the clinical reference range recommended by the clinical laboratory, the lifestyle coach may recommend lifestyle changes that have been previously demonstrated to produce improvements in that marker
- Behavioural coaching to improve clinical biomarkers.

Ethics of self-tracking

- Data double, our digital avatar or twin
- Digitalization of healthcare in terms of big data and wearables
- Retrieve data from devices, framed in a certain way. Rather than direct measurements of skin temperature or heart activity, the user first has to upload the information for analyses, and it is often preformatted by the makers of the app
- Standards of normalcy: personalisation requires an “other”, a standard
- Personalisation as a product of the We (human reference genome), the self (self-tracking) and the other (standards of normalcy)
- Participation: data sharing, gift, data harvesting, biological citizenship, solidarity or appropriation?

Conclusions

- Promise management (forgo overpromising, credibility of science)
- Me-medicine: the other (We) as reference
- Person-centred or technology-centred?
- Genome and context (ecosystem, environment)
- We are not our genome, we are the outcome of a dialectical interaction between nature and nurture, genome and environment, biology and culture, etc.