What if consumers could use devices to sequence DNA?

Sequencing and analysis of the human genome – all the genetic information stored in our DNA – provide us with understanding about a person's ancestry, health and other traits. Thanks to DNA sequencing, medicine and the life sciences are able to predict and cure diseases. As DNA sequencing technologies continuously improve and become less costly, what if we all soon possessed our personal, smart DNA sequencers and apps to analyse our DNA?

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The genetic 'alphabet' contains four 'letters' or, in other words, four chemical codes for the information stored in DNA, representing a sequence that, if unwrapped, can stretch up to two metres and fill a book of one million pages. DNA contains complete hereditary information about a person, from their eye and hair colour to their sex, some personality traits and disease vulnerability. DNA sequencing is a method that determines the precise order of the 'letters' within DNA, allowing us to 'read' the information in a genome. While deciphering the human genome for the benefit of scientific research, medical applications and public health, DNA sequencing of other species is also important: to verify biodiversity, to gain knowledge of crops and farmed animals, and to learn about the origin of emerging pathogens. In the case of a public health crisis, such as the Covid-19 pandemic, genomic sequencing of SARS-CoV-2, the virus causing Covid-19, facilitates the design of treatments and vaccines. It also aids in mapping the spread of the virus as it evolves and helps monitor containment measure effectiveness.

The Human Genome Project, which aimed to decipher the complete human genome, was completed in 2003. With the aid of international research, this took about three years and cost approximately three billion euros. However, the cost of and time needed for genomic sequencing are falling. Currently, it takes only about an hour and less than a thousand euros to sequence a complete genome. This technological progress has paved the way for the commercialisation of genome sequencing. Dozens of personal genomics companies, such as 23andMe or Ancestry, offer direct-to-consumer DNA testing, that can be bought online for less than fifty euros, offering to provide a range of information. As advanced genomics becomes more accessible and increasingly affects our daily lives, ethical concerns such as identifiability, consent, data privacy and security rise. It is therefore desirable to have regulation in place that keeps up with developments in technology before we all own our personal DNA sequencers, which could provide more than ancestry and disease risk information.

Potential impacts and developments

The decreasing cost and increasing speed of DNA sequencing techniques make genomics more accessible. Increasingly important worldwide, genomics offers more effective healthcare solutions. For example, if genomics analysis indicates a person's genetic predisposition for type-II diabetes, modifications in lifestyle, such as exercise and a healthy diet, could delay or prevent the onset of the disease. Genomics facilitates a more precise characterisation of medical conditions – such as cancers, some rare diseases and infectious diseases – and, accordingly, the choice of medication to target the disease. It can provide better diagnostics for disease prevention and has the potential to move personalised medicine and therapies forward. Prenatal genomic sequencing for every embryo may become a standard protocol, serving as a life-long health reference. Besides the benefit of assisting individuals to make decisions about their health, gathering information about genetic

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traits and diseases within populations, the field of public health genomics, could offer strategies for improving public health. This not only helps to access and monitor health problems, but also to frame public policy options.

DNA sequencing technologies can be used to scan and detect pathogens in real time and provide a surveillance system to prevent epidemic outbreaks. Portable DNA sequencers, smaller than a smartphone, such as MinION, which is the first of its kind, were used during the Zika virus epidemic in 2016 and the 2014-2016 Ebola virus outbreak, to help identify and track the viruses. Farmers in Tanzania have benefited from portable DNA sequencing for the timely diagnosis of sick plants infected with viruses. IDseq, an open-source cloud-based platform defining itself as 'infectious disease detectives', launched a service where users can upload sequencing data for a free analysis. IDseq is also being used for molecular diagnostics to tackle Covid-19. Genomic sequencing of SARS-CoV-2 is key to understanding the evolution and trajectory of the virus, assists containment measures, and has provided evidence that the virus is not a bioweapon. Recent research has even claimed that an on-the-spot DNA test integrated into smartphones could make real-time monitoring of diseases and pathogens possible.

The technology for pocket-size, portable, smartphone-integrated DNA sequencing is already available. This, together with improved mobile genome analysis applications, which enable the exploration of desirable genetic data, are opening an era of great possibility and risk. It is only a matter of time before personal DNA sequencers are commercialised and used to detect pathogens, food contamination or information about ourselves and others. In future, sequencers may be found in public spaces, such as hospitals or bathrooms, cars, or integrated into a fridge in our kitchens, to monitor genomes in real time. However, the interpretation of all this genomic information is not as easy as it may appear.

While genomics holds invaluable information for scientists, doctors and others, it also provides much more information about who we are, how and even where we live, where we go and with whom we interact. In a human sample (e.g. saliva), genomics can also detect genomic sequences of the microbiome, microbes residing in the human body, which can reveal a lot about an individual. As data analysis advances, there will be much more to uncover in genomic data, and consumer genomics may possibly enable a person to be identified from anonymous DNA databases. We also know that investigation of the genomic information of a part of a population facilitates the inference of the DNA make-up of the rest of that population. All this information can be commercially relevant. The question of who owns the DNA sequencing data and who else has access to it after one has given consent over the data are therefore crucial. The consumer genomics business faces challenges regarding privacy, data ownership and sharing, as well as data security. Genomic databases have already been hacked. DNA testing services can be cracked, sometimes revealing email addresses alone, whereas, in 2020, hackers accessed an online DNA database used by police for criminal searches.

Anticipatory policy-making

Genomic sequencing unveils very rich data and raises considerable legal, security and private as well as societal and ethical issues, touching upon basic human rights. As genomic technologies are evolving rapidly, it is desirable that regulation match this pace and that adequate safeguards are put in place. It is also important to raise societal awareness about this technology, capable of revealing people's identities, and as such open to misuse. The declaration on genomics cooperation, signed by 13 European countries in 2018, on cooperation in sharing their genomic information, aims at sequencing '1+ million genomes' in the European Union (EU) by 2022, thereby linking genomic databases to create a secure health data infrastructure across Europe, while ensuring that ethical and legal aspects are covered and that Member States are aware of the opportunities and challenges of genomics. A 2018 JRC report provides a comparative overview of Member States' legislation on genomics.

Direct-to-consumer genetic testing is not currently harmonised at EU level. The legislative framework is fragmented and ranges from an absolute ban on these tests (France), to a complete lack of any legislative reference to this form of testing. At EU level, the revision of the In-Vitro Diagnostics Medical Devices Regulation that will come into force on 26 May 2022, will touch upon their clinical validity, meaning that EU Member States will continue to regulate medical supervision, genetic advice and monitoring and informed consent issues at the local and/or national level. In a future where personal DNA sequencers may be as common as smartphones, vigilance could be required on both legal and ethical issues. Genomic information is difficult to protect; we leave traces of our DNA, such as hair and fingerprints, wherever we go. Therefore, attention should be paid to data security, data ownership and sharing. Blockchain technologies may help to address some of these issues. Ethical issues such as individual autonomy and equality of access, as well as anti-discrimination laws and compensation for people affected by data abuse should also be considered.