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editorial

Genetics, Big Data and Globalisation – are we satisfied yet?

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Testing for the presence of genetic anomalies has seen huge technical advances in the last 40 years, particularly since the decoding of the human genome and has become a routine procedure in most countries worldwide, supported by medical and laboratory specialized professionals.[1] In each of these decades (1980-until the present date) hundreds of scientific papers have announced new ages of outstanding research that have allowed us to be very optimistic towards more and better prevention, testing and cure of genetic diseases. Concomitantly, bioethics has encompassed these big steps in science, medicine and technology, particularly after the Second World War, as shown by the production of "The Nuremberg Code, 1947", and "The Universal Declaration of the Human Rights, 1948".[2,3] While the prevention and diagnosis of human genetic diseases is undeniably a compassionate and civilised improvement in healthcare, the development and use of some new genome techniques, such as genome editing, is more unclear. It is possible to speculate on the goodness of the outcome of breakthroughs research, but the translation to a clinical setting may not be easy or even possible, at least for still many years to come. Therefore, the principle of precaution should always be applied because other ethical principles, *e.g.*, privacy, marginalisation and justice may be at stake and should not be overlooked.[4]

Completed in April 2003, the Human Genome Project (HGP) was a major advance in science because, for the first time, it became possible to read the sequence and map all of the genes - together known as the *genome* - that form a human being of our species, *Homo sapiens*.[5] It was so exciting – and yet, after about ten years and millions of sequenced genomes later, it was no longer enough. In 2015, the Precision Medicine Initiative (PMI) was announced in the U.S., having in mind a "unique and personalised medicine", with which true "customized treatment and prevention strategies to the unique characteristics of people" could be offered.[6] Three years on, in 2018, this Initiative has been transformed into another new project, "All of Us", with the aim of collecting genetic data and health data from one million volunteers by 2022.[7] This means there is a lot of investment, huge expectations and really big data scrutinizing. From "personalised", genetics has become "precision" medicine, although we might say now participatory – through this newest research, patients will now be able to participate more directly in the development of science, their own health and illness.[8] In Europe, until the 15th October 2018, 18 countries (including Portugal) have signed the declaration "Towards access to at least 1 million sequenced genomes in the EU by 2022", making a joint European effort to deliver cross-border access to genomic health data and thus hoping to "contribute to better prevention of diseases and more accurate personalised treatments, in

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particular for cancer and brain related diseases, as well as for rare diseases".[9] No doubt the combination of genomic research, population studies and the analysis of big data will allow better "precision medicine" - how the principles of equity and justice will be cared for and regulated during this research, we might wonder…

Finally, on 28 November 2018, a really revolutionary piece of scientific advancement was announced to the world by He Jiankui: Lulu and Nana were born normal and healthy. This researcher claimed to have edited the genome of a pair of twin girls, on his own and without any official permission, while also stating that he was proud of what he had done.[10] In April 2018, in relation to the technology of gene editing, Hofmann (2018) had already warned us about *the gene-editing of super-ego*, reminding us that "the aim is not to bar the development of modern biotechnology, but rather to ensure good developments and applications of highly potent technologies".[11] This news was not just about a piece of human DNA that had been removed from a patient in order to avoid a genetic defect in a somatic cell line, it was a direct intervention in an embryo and, consequently, the alterations that have been introduced will be transmitted to the next generation. And, at the present moment, the gene-editing technology is still on the investigational phase - therefore the international guidelines should be followed by responsible researchers, however tempting "playing God" might be. Meanwhile, on the 10th December 2018, the Universal Declaration of Human Rights will reach its 70th anniversary, and this date should be a good opportunity to highlight the important breakthroughs brought about by this UN document.[12] We may now wonder where do we stand and where do we go from here? From a purely scientific point of view, studying and interpreting the human genome analysis, by whatever is considered the best possible method for a "patient", applying this knowledge to the benefit of the "patient" and making the "patient" and family better, will always be the most rewarding scientific accomplishment of our time in a clinical setting, and it should be the success key for a reasonable, responsible and health professional behaviour.

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