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Abstract

Bioethical reflections on precision medicine and diagnostic-therapeutic developments

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The opinion was drawn up in the context of a mixed working group, between the National Committee for Bioethics and the National Committee for Biosafety, Biotechnology and Life Sciences.

The document initially describes the key aspects, from a technological and epistemological point of view, of the "genetic revolution" over the last two decades, which has allowed the development of precision medicine (PM): under the first profile, the reference is to next-generation sequencing technologies (NGS) and to "gene editing" techniques, under the second profile, to the transformation of the paradigms of human genetics that marks the transition to a "post-genomic" and "holistic" concept of the human body.

After distinguishing precision medicine, centred on the stratification of patients based on the molecular profile, from "personalized medicine", tailored to the specific characteristics of the individual, the opinion illustrates, giving some examples, of the state of the art of PM (pharmacogenomics and diagnostic-therapeutic advances). In addition to the difficulties encountered in implementing this promising approach to post-genomic medicine, it then focuses on the bioethical and bio-legal issues it raises. The issues related to the different way of understanding the pathogenesis of diseases and, consequently, their traditional classification, are reported, as well as those of an organizational-formative nature and highlights the challenges related to the balance between the resources to invest in the most innovative research on which the hopes of many patients converge, and those necessary to continue research in more traditional sectors, from which an improvement in the health conditions of the population in general is expected. From a bioethical and bio-juridical point of view, specific questions arise regarding protection of the privacy of the subjects receiving medical care, participating in clinical trials and/or donating their biological samples.

The opinion recommends that the introduction of PM should respect the bioethical principle of equity in health care, avoiding abuses in the prescription and performing of genetic tests, making more investments in innovative research (pharmacogenomics would already allow an economic return capable of balancing costs by avoiding wasteful spending on ineffective and/or harmful drugs) and that it should adopt legal instruments to protect the personal identity of patients, without prejudice to the sharing of data and collaboration, at national and international level, between research centres.

In addition, it calls for a reorganization of the health care system needed to support the expansion of PM and recommends implementation of planning aimed at innovation in the provision of services (increase in the number of centers equipped with the skills required for PM, control of quality standards of laboratories, promotion of disciplinary interaction, adaptation of informed consent) and planning of public information and awareness-raising strategies on the importance of the new findings of human genetics, with reference to the "best practices" that already have produced remarkable results in this direction.

The document is accompanied by a glossary.