

Implementation of genomic policies in Italy: the new National Plan for innovation of the Health System based on omics sciences

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For decades, genetics concerned the study of single genes and chromosomes, but especially after the sequencing of human genome it has become a driving force in medical research and practice [1]. Genome analysis, in fact, has acquired a specific role for progress of medicine and healthcare, resulting in rapid development of both genomics and molecular genetics. In this context, over the last few years, there has been an increasing and uncontrolled availability of genetic tests both for monogenic and complex diseases [2]. Therefore, progress in genomics had crucial implications for public health: in fact, this knowledge offers the opportunity to differentiate individuals and groups most likely to develop certain pathological conditions within populations, and this with new ways compared to those traditionally used by public health professionals. In the last two decades, the use of words ending in "-omic" has extended, from the initial "genomics", to a wide range of biomolecular disciplines addressed to the study of specific aspects considered as a whole. Therefore, the omics sciences study pools of biological molecules (eg ions, nucleic acids, proteins, enzymes) with various functions within living organisms [3] and have the primary objective to analyze as a whole

e.g. genes contained in DNA (*genomics*) and their multiple functions (*functional genomics*), DNA transcription product - RNA- (*transcriptomics*), proteins encoded by DNA through RNA (*proteomics*), molecules that interact within an organism or metabolites (*metabolomics*). Among the other goals of these sciences is also to study the connections and reciprocal interactions between the pool of biological molecules (*interactomics*) and between these molecules and microorganisms of the intestinal flora (*microbiomics*), foods and/or nutrients (*nutriomics*) [4].

The dissemination of omics disciplines has been possible mainly through the development of high-throughput technologies capable to generate a large amounts of data related on the different levels of biological complexity (DNA, mRNA, proteins, metabolites, etc.), contributing to revolutionize the approach to the study of living organisms [5].

Omics technologies such as Next Generation Sequencing (NGS) have a vast application potential ranging from increasing the understanding of different physiological and pathophysiological processes to their use in screening, diagnosis and evaluation of natural history, response to the therapy and/or prognosis of

various disorders [6].

The use of these new technologies for the characterization and analysis of different biological levels can better guide the knowledge of pathogenetic mechanisms at a molecular level, allowing, where possible, to identify subgroups of patients or biomarkers to improve diagnosis or planning personalized therapies.

In this context of research and innovation, the plethora of high-throughput technologies available to the omics sciences and their rapid evolution require the scientific community to adopt greater harmonization and standardization in data generation and analysis methods, and at the same time, the existence of extensive possibilities for the implementation of new data mining tools. Therefore, one of the current challenges is to overcome the gap between the production of omics data and the progress of high-throughput technologies and our ability to manage, integrate, analyze and interpret this huge amount of data. Consequently, in the next decade, genomics and other omics sciences will play a decisive role within of Big Data [7].

The complexity of *omics* sciences and related applications such as personalized medicine, certainly requires a governance from the healthcare systems. Implementing a personalized approach to health care will also require a change in the organization of health services. On a broader basis, public health leaders are responsible for helping to catalyze change in the organization of health services and public policies to ensure that genomics and other technologies are used to best effect [8].

An appropriate integration of genomics in healthcare requires that health system policy-makers, stakeholders and knowledge brokers are aware of the potentials and limits of the use of genomics in disease risk prediction, diagnosis and treatment, so that they can provide the necessary policy response. Therefore, it is necessary to engage current health system professionals, leaders and citizens in both grasping the opportunities and tackling the challenges of a personalized healthcare also. The way in which the public health system adapts to this evolving health ecosystem will be a significant determinant of whether personalization of healthcare can, in the end, lead to greater gains in overall population health [8].

Accordingly to WHO, "in light of existing health challenges, policy makers can capture the potential of genomics to meet public health goals through health policy. The exact role of genetics in the causation of human disease needs to be clarified which means investing in education and research. This understanding will pave the way to innovative new ways of applying the tools of genomics to address health challenges. There is a great need to evaluate the existing utility of genetic services and technologies in addressing existing disease burdens" [9].

While several European countries, such as Italy, have implemented a legal framework or even national plans for the integrations of genomics advances into healthcare

systems, according to the Italian Chief Medical Officers survey initiative [10], few countries have implemented a structured national policy [11].

In Italy the National Prevention plan foresees, since 2012, the macro-area of personalized medicine, along with primary/secondary/tertiary traditional prevention areas. In 2013, to implement the personalized prevention plan, the State-Region conference approved and published the national plan of public health genomics [12]. A further step has recently been made with the approval of a "National Plan for Innovation of the Health System based on omics sciences" [13].

This plan meets the challenge of a comprehensive approach to innovation, taking into account that such an innovation is deeply intertwined with the economic growth of the Country, in particular around the cross-cutting topic of the IT. According to OECD [14], the plan also aims to support the careful implementation and the intelligent use of big data in the health care sector and to foster the eventual achievement [15] of significant benefits in terms of both population health and economic system.

The Plan is also committed to implement the recently issued European Union Council conclusions on personalized medicine for patients [16].

The Plan outlines the ways in which innovation in the -omic field should re-shape the National Health System (NHS) in the areas of prevention, diagnosis and care, taking into account effectiveness (evidence-based) and sustainability (cost-effectiveness) of the NHS to improve the health of the individual and the population.

The Plan aims to support the NHS in order:

1. to increase the awareness of all stakeholders on the innovation of *omics* sciences and its effects on the health of individuals and populations enhancing the capacity of the society to cope with the cultural, ethical, psychological aspect of the 'genomic revolution';
2. to put in place a strategy of "government of innovation" of genomics and related fields;
3. to evaluate and implement the opportunities currently offered by genomics and by the other *omics* sciences for the health of the population.

The main objectives of the national plan for innovation are:

1. to transfer genomic knowledge into the practice of health services, in a patient-centric approach;
2. to increase the effectiveness of prevention, diagnosis and treatment of diseases at a higher burden, taking into account individual differences in genetic heritage, lifestyles and the environment, and providing professionals with the resources needed to customize interventions;
3. to promote the cultural, scientific and technological innovation of the healthcare system.

The Plan focuses on several topics including: genomics in the diagnosis of mendelian and complex diseases (eg

tumors), personal prevention (pre-conception tests, pre and postnatal tests, neonatal screening), genomics in therapy (pharmacogenomics and personalized tumor therapy). In addition, the plan highlights what are the functions of the central government and actions to support the implementation of the Plan. It also provides the main indications to foster research and innovation in the following lines:

- big data and computational medicine,
- literacy technologies,
- opportunities for the national health system to remain sustainable by replacing drugs,
- making pharmacogenomics research more efficient,
- opportunities for the national health system to remain sustainable through pre-primary prevention aimed at reducing the burden of disease,
- opportunities for the national health system to remain sustainable through secondary prevention aimed at reducing the burden of breast cancer,
- opportunities for the national health system to remain sustainable through early diagnosis aimed at reducing the burden of cancer disease,
- undiagnosed patients.

Italy has already started developing a governance and a policy on genomics and the related innovation process. However, it is clear that such a policy cannot be implemented nor be conceived without a strict and advantageous cooperation with professionals and citizens through their scientific and civil society organizations.

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