

Precision medicine: a revolutionary trans-disciplinary field

Olga Amaral

olga.amaral@insa.min-saude.pt / <https://orcid.org/0000-0002-3478-2122>

Instituto Nacional de Saúde Ricardo Jorge - Human Genetics Department –Unit of Research & Development - Porto, Portugal

Author disclosures: nothing to disclose

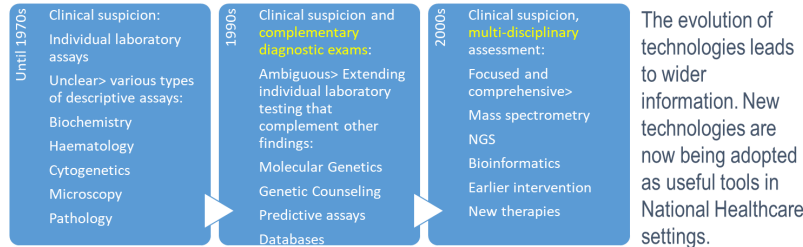
Abstract

The advances in molecular biology and biotechnology, applied to human diseases, have allowed the rise of precision medicine. New approaches to medical diagnostics and care are now becoming a reality with precision healthcare.

For years, diagnostics were based on clinical findings followed by strenuous work in the laboratory. The detective work of health professionals could take many years until a diagnostic was achieved and in the meantime the patients were degenerating with devastating disease.

Advances in molecular biology and biotechnology

Evolution from isolated testing to organized diagnostic laboratory workflow. The advances in molecular biology and biotechnology, allowed faster and more focused approaches to diagnosis



Time lag between clinical suspicion and diagnosis



The detective work of health professionals can take many years until a diagnosis is reached

Automation of testing suffered many generations of change, from rudimentary adaptations of laboratory instruments to the development of high-tech high-throughput apparatus



These new tech instruments are transversal to most laboratories and can be adapted to various fields and methods

- Molecular biology is now highly developed and covers varied areas.

Time between clinical suspicion and diagnosis

The detective work of health professionals can take many years until a diagnosis is reached.

Automation of testing suffered many generations of change, from rudimentary adaptations of laboratory instruments to the development of high-tech high-throughput apparatus. These new tech instruments are transversal to most laboratories and can be adapted to various fields and methods.

Molecular biology is now highly developed and covers varied disciplines.

Bridging the time gap

In 2000 a first draft of the human genome was announced, in 2001 about half of the work was already available in public databases and in 2003 the final version was concluded.

A diagnosis of a rare disease, that could take decades, can now be much faster, although it still requires a few years. From the first medical observation, specialists considering the various hypotheses and geneticists being called to carry out a deeper analysis and identifying the relevant variants, several years may go by. In the meantime, health deteriorates depending on the type of disease. Nevertheless, hundreds of rare diseases are identified on a daily basis. Although it is important to identify the causes, providing early intervention is fundamental.

Wide-reaching cooperation

Timely intervention is desirable prior to irreversible damage. Ethical issues may prevent predictive diagnosis and sharing of sensitive results. Sharing data may contribute to early intervention, it may reduce the burden of the unknown for those affected, and the cost of long-term approaches with limited results. But platforms for safe keeping of big data, faster capacity for the analysis of the high volume of data and the ethical issues related to equitable access to advanced healthcare means need to be globally addressed.

In conclusion, cooperation and sharing of information is pivotal to achieve diagnosis, to improve testing, to understand variants and to provide answers in the benefit of patients. Even with infectious diseases we are witnessing the importance of global sharing of data and molecular biology advances.

Precision medicine

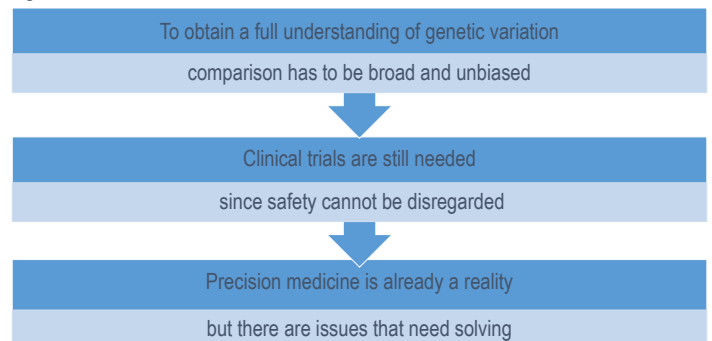
The concept of precision medicine implies that the approaches followed are on the basis of the genetic characteristics of the individuals. The multi-disciplinary advances in molecular biology and biotechnology, applied to human diseases, have allowed the growing availability of healthcare data and the possibility to offer precision medicine based on the patient's individual characteristics.

New approaches to medical diagnostics and care are now becoming a reality with precision healthcare.

To overcome the challenges of precision medicine sharing of information among medical professionals, as well as between them and the public has to be wide spread. Information cannot be polarised or manipulated, trust and transparency are key factors for the success of precision medicine.

Precision medicine challenges

To ensure equitable access to precision medicine there are obstacles that require fine tuning



References

Ginsburg GS, Phillips KA. Precision Medicine: From Science To Value. *Health Aff (Millwood)*. 2018 May;37(5):694-701.
 .Woodbury RB, et al.. Community Perspectives on Communicating About Precision Medicine in an Alaska Native Tribal Health Care System. *Front Commun (Lausanne)*. 2020 Sep;5:70.