Science, Technology and Innovations in Oncology

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Ciência, Tecnologia e Inovações em Oncologia

Ciencia, Tecnología e Innovaciones en Oncología

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The first major groundbreaking of genomic and precision medicine happened in the beginning of 2000. The Human Genome Project (HGP) initiated in 1990 and completed in 2003, has not only significantly impacted our understanding about the architecture of the human genome and its correlation with different diseases but pushed forward a multidisciplinary[1] technological revolution. Twenty years later, they are still innovative worldwide as its implementation by health systems is not simple requiring an array of complex[2] actions and initiatives. The rapid progress of molecular diagnosis, advanced therapies and precision medicine highlight the necessity of translating the knowledge to optimize the applicability by health services for the population[3].

As the second main cause of deaths in developed and in development countries, their initiatives in genomic and precision medicine have prioritized oncological[4-6] diseases. According to the Global Cancer Statistics[6] in 185 countries, it is estimated a global increase of nearly 50% of new cases between 2020 and 2040[6]. This reflects the populational ageing and growth associated with prevalence of risk factors.

An oncologic patient is diagnosed through lab and histopathological tests and biopsy and referred for a standard medical consultation for surgical, chemotherapy and/or radiotherapy treatment in the traditional medicine. Precision or personalized medicine is an emerging approach to the genetic profile of the individual with data aggregation already utilized for diagnosis and treatment (signs, symptoms, personal and family history and complementary exams). Through this approach, it is achieved boosted accuracy of the diagnosis, enhanced prediction foreseeing the appearance of genetic and hereditary infirmities and patient-centered treatment[7].

The identification of genes associated with predisposition to chronic non-communicable diseases (NCDs) was boosted by the development of next-generation sequencing methods (NGS). According to the National Health Institute (NIH)[8], the cost of human genome sequencing is below US$1,000 per person in contrast to the cost estimated in 2001 of US$ 100,000,000. This method utilized for a novel approach to health allows safer and more effective clinical practices in addition to promoting the improvement of the quality-of-life and rational use of health resources. The United Kingdom[9] (100.000 Genomes), USA[10] (All of Us), Singapore[11] (Singapore 10K Genome Project) and Australia[12] (Australian Genomics Health Alliance) have already developed and implemented initiatives in precision medicine which has robust potential to improve health systems.

Genomic analyzes-based genetic tests can help to identify cancer genetic syndromes in individuals with family oncologic history, indicating the predisposition to different types of tumors. The preventive intervention can significantly reduce the risk of developing hereditary breast cancer (HBC), which accounts for 10% of breast cancer cases in carriers of genetic mutations[13]. Next-generation sequencing identifies potentially genetic alterations for the development of cancer cells[14] and specific tumor associated biomarkers for own tissue tumor antigens therapies[15], further to patient or group-centered therapies based in specific genetic data.

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Studies concluded that biomarkers of a certain disease can vary widely among populations, the findings for a certain group do not necessarily apply to distinct ethnically individuals[16,17]. Most genomic data currently available at databanks refer to low genetic heterogeneity populations. A great limitation to expand and consolidate precision medicine in Brazil is the lack of genomic data representative of an ethnical-diverse population with one of the highest miscegenation rates[18,19] in the world.

From this background scenario, the "*Departamento de Ciência e Tecnologia da Secretaria de Ciência, Tecnologia e Insumos Estratégicos*" of the Ministry of Health (Decit/SCTIE/MS) has launched on August 4, 2020, the National Program of Genomics and Precision Health – *Genomas Brasil* through Directive 1,949[20]. *Genomas Brasil* is a program of innovation, technology and science to ensure the Brazilian population the access to precision medicine products and services. One of its major strategies is the creation of a Genomics and Health National Database – GenBR database – which will host genome sequencing and health information of 100 thousand Brazilians in the upcoming years. It will allow the identification of the main genetic characteristics of the Brazilian population through novel studies for diseases prediction and prevention and to develop new healthcare treatments, therapies and strategies.

The patient, whose genomes will be sequenced will be enrolled from primary and specialized attention network and will be granted access to an accurate and continuous genetic diagnosis and counseling as needed. *Genomas Brasil* aims to encourage and strengthen the research, innovation and training of the national health workforce in precision medicine; in addition, robust furtherance in prognosis and cancer treatment by the National Health System (SUS) with support to projects able to identify potential biomarkers and analysis of the correlation between the genomic, epidemiological, clinical and family profile as strategy of disease therapy.

Genomas Brasil presents a proof of concept which will offer important data to demonstrate the worth of precision medicine for SUS, supporting future decision-making for implementation, estimation and organization for the public health system. The program is pivotal for a firm transition to a precision-medicine based health system, a critical step to adjust the strategies, methodologies, protocols and management of risks and potential hurdles to be dealt with. It intends to revolutionize SUS through cutting-edge knowledge strategies to provide the most accurate and modern healthcare to the Brazilian population, with solid transformations for health caring workforce to foster scientific knowledge and the national health industry.

REFERENCES

1. Collins FS, Fink L. The human genome project. Alcohol Health Res World [Internet]. 1995 [cited 2022 May 27];19(3):190-5. Available from: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6875757/pdf/arhw-19-3-190.pdf
2. Bertier G, Carrot-Zhang J, Ragoussis V, et al. Integrating precision cancer medicine into healthcare-policy, practice, and research challenges. Genome Med. 2016;8(1):108. doi: https://doi.org/10.1186/s13073-016-0362-4
3. Levit LA, Kim ES, McAneny BL, et al. Implementing precision medicine in community-based oncology programs: three models. J Oncol Pract. 2019;15(6):325-9. doi: https://doi.org/10.1200/JOP.18.00661
4. Bray F, Ferlay J, Soerjomataram I, et al. Global cancer statistics 2018: GLOBOCAN estimates of incidence and mortality worldwide for 36 cancers in 185 countries. CA Cancer J Clin. 2018;68(6):394-424. https://doi.org/10.3322/caac.21492
5. Siegel RL, Miller KD, Fuchs HE, et al. Cancer statistics, 2021. CA Cancer J Clin. 2021;71(1):7-33. doi: https://doi.org/10.3322/caac.21654
6. Sung H, Ferlay J, Siegel RL, et al. Global cancer statistics 2020: GLOBOCAN estimates of incidence and mortality worldwide for 36 cancers in 185 countries. CA Cancer J Clin. 2021;71(3):209-49. doi: https://doi.org/10.3322/caac.21660
7. De Maria Marchiano R, Di Sante G, Piro G, et al. Translational research in the era of precision medicine: where we are and where we will go. J Pers Med. 2021;11(3):216. doi: https://doi.org/10.3390/jpm11030216
8. Wetterstrand KA. DNA sequencing costs: data from the NHGRI Genome Sequencing Program (GSP) [Internet]. Bethesda (MD): National Human Genome Research Institute; [last updated 2021 Nov 1; cited 2022 May 27]. Available from: https://www.genome.gov/about-genomics/fact-sheets/DNA-Sequencing-Costs-Data
9. Trotman J, Armstrong R, Firth H, et al. The NHS England 100,000 Genomes Project: feasibility and utility of centralised genome sequencing for children with cancer. Br J Cancer. 2022. doi: https://doi.org/10.1038/s41416-022-01788-5
10. Ramirez AH, Sulieman L, Schlueter DJ, et al. The All of Us Research Program: data quality, utility, and diversity. MedRxiv [Preprint]. 2020 June 3. doi: https://doi.org/10.1101/2020.05.29.20116905
11. Vimal M, Devi WP, McGonigle I. GenomeAsia100K: Singapore builds national science with Asian DNA. East Asian Sci Technol Soc. 2021;15(2):238-59. doi: https://doi.org/10.1080/18752160.2021.1896138

12. Stark Z, Boughtwood T, Phillips P, et al. Australian genomics: a federated model for integrating genomics into healthcare. Am J Hum Genet. 2019;105(1):7-14. doi: https://doi.org/10.1016/j.ajhg.2019.06.003
13. Kuchenbaecker KB, Hopper JL, Barnes DR, et al. Risks of breast, ovarian, and contralateral breast cancer for BRCA1 and BRCA2 mutation carriers. JAMA. 2017;317(23):2402-16. doi: https://doi.org/10.1001/jama.2017.7112
14. Schrader KA, Cheng DT, Joseph V, et al. Germline variants in targeted tumor sequencing using matched normal DNA. JAMA Oncol. 2016;2(1):104-11. doi: https://doi.org/10.1001/jamaoncol.2015.5208
15. Sanmamed MF, Chen L. A paradigm shift in cancer immunotherapy: from enhancement to normalization. Cell. 2018;175(2):313-26. doi: https://doi.org/10.1016/j.cell.2018.09.035
16. Hindorff LA, Bonham VL, Brody LC, et al. Prioritizing diversity in human genomics research. Nat Rev Genet. 2018;19(3):175-85. doi: https://doi.org/10.1038/nrg.2017.89
17. Martin AR, Kanai M, Kamatani Y, et al. Clinical use of current polygenic risk scores may exacerbate health disparities. Nat Genet. 2019;51:584-91. doi: https://doi.org/10.1038/s41588-019-0379-x
18. Bentley AR, Callier S, Rotimi CN. Diversity and inclusion in genomic research: why the uneven progress? J Community Genet. 2017;8(4):255-66. doi: https://doi.org/10.1007/s12687-017-0316-6
19. Marrero AR, Leite FPN, Carvalho BA, et al. Heterogeneity of the genome ancestry of individuals classified as White in the State of Rio Grande do Sul, Brazil. Am J Hum Biol. 2005;17(4):496-506. doi: https://doi.org/10.1002/ajhb.20404
20. Ministério da Saúde (BR). Portaria nº 1.949, de 4 de agosto de 2020. Altera a Portaria de Consolidação nº 5/GM/MS, de 28 de setembro de 2017, para instituir o Programa Nacional de Genômica e Saúde de Precisão - Genomas Brasil e o Conselho Deliberativo do Programa Genomas Brasil [Internet]. Diário Oficial da União, Brasília, DF. 2020 ago 5 [acesso 2022 maio 27]; Seção 1:87. Disponível em: http://bvsms.saude.gov.br/bvs/saudelegis/gm/2020/prt1949_05_08_2020.html

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