

# Chapter 29

## Genetics and Public Health

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### ABSTRACT

*This chapter wraps up by discussing the crucial role played by public health specialists who must reconcile traditional public health concerns of health inequality and equity with safe and effective health interventions and diagnostics that meet individual health needs. Since most genetic diseases in the realm of public health are an interplay of different genetic, lifestyle, and environmental factors, genomic science has given greater emphasis to the importance of molecular and cellular mechanisms in health and disease. New biological knowledge must be integrated with the social and environmental models to improve health at individual and population levels. Public health specialists must now be able to integrate genome-based knowledge into public health in a responsible, ethical, and effective way and anticipate the increase in the health service requirements likely to occur in the future. The foundational pillars of bioethics (beneficence, non-maleficence, autonomy, and justice) must be protected by all public health stakeholders.*

### CHAPTER OUTLINE

- 1 The Role of Public Health and Public Policy
- 2 Big Data, Personalized Medicine, and Biomarkers
- 3 Ethical, Legal and Social Considerations
- Chapter Summary

### LEARNING OUTCOMES

- Characterize the role of public health and public policy in the use of genomic information
- Explain the meaning of personalized medicine and its potential applications
- Discuss the social, legal and ethical issues arising in the use of genetic information

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## 19.1 THE ROLE OF PUBLIC HEALTH AND PUBLIC POLICY

Connecting genes and their phenotypic manifestation is complicated necessitating the translation of information about genes and DNA sequences into facts about genetic susceptibility to disease, the interaction between these susceptibilities, modifiable risk factors, and the impact of this knowledge on population health. From a public health perspective, systematic, evidence-based evaluations and technology assessments are critical to the incorporation and use of genomics in **clinical** and **public health practice**. *Genetics* is the science of inheritance focusing on how Mendelian inherited traits with single-mutations cause a disease. The more recent term *genomics* focuses on studying complex set of genes, their expression, and how they interact with other genes and the environment to affect disease development. Genomic science has given greater emphasis to the importance of molecular and cellular mechanisms in health and disease. Since most genetic diseases in the realm of public health are an interplay of different genetic, lifestyle and environmental factors, **public health specialists** must reconcile traditional public health concerns (like **health disparities** and **health equity**) with safe and effective health interventions and diagnostics that meet individual health needs. Once the genetic and environmental factors involved in the causation of disease and how they interact (referred to as epigenomics) is understood, public health genomics' goal is to devise effective preventive interventions targeted at individuals with susceptible genotypes. New biological knowledge must be integrated with the social and environmental models to improve health both at the individual and population levels. Public health genomics is clearly a very interdisciplinary research field bringing together different disciplines that will include medical sciences, statistics, biotechnology, engineering, pharmaceutical research/industry, health policy, ethics, law, sociology, public health practitioners, genetic centres, governments, non-governmental organizations, and representatives of patient groups (Marzuillo et al., 2014; Zimmern & Khoury, 2017).

Following the completion of the Human Genome Project in 2003, the utility of genomics in improving public health education, outreach, and interventions has come into the limelight as public health practitioners in the public and private arenas reposition to provide more proactive guidance and leadership to improve population and community health outcomes. Public health specialists must now be able to integrate genome-based knowledge into public health in a responsible and effective way and anticipate the increase in the health service requirements likely to occur in the future. Public health agencies must work out feasible ways to access and analyse the results of genome-based research and technologies and identify information gaps at both individual and population levels to allow the formulation of appropriate policies to guide **evidence-based interventions**. Public health aims at improving the health of the entire population by implementing preventive strategies, but behind the entire population is the individual.

In a most inspiring characterization of the complexity of today's biomedical research problems, Zerhouni (2003) noted over 15 years ago that scientists must reorient in the new millennium to envelope the interdisciplinarity of knowledge to new organizational models emphasizing team science. This will enable a holistic understanding of the interplay between genetics, diet, infectious agents, environment, behaviour, and social structures. Ultimately, public health successes and/or failures will be measured by the level of involvement in four main areas: 1) **Translational research** to conduct systemic reviews of genomic studies, develop evidence-based policy and practice guidelines, and monitor impacts on people's health; 2) **Epidemiological studies**, to extend **genetic biobanks** and disease registries with genomics information; 3) Harmonization of genomics data with the persistent health disparities and equity concerns; 4) Evaluating the social, ethical and cultural issues arising from expanding realms of genomic data.

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