This volume provides detailed and comprehensive coverage of population structures, human genomics, and genome variation—with particular emphasis on medical and health issues—in the emerging economies and countries of the developing world. With sections dedicated to fundamentals of genetics and genomics, epidemiology of human disease, biomarkers, comparative genomics, developments in translational genomic medicine, current and future health strategies related to genetic disease, and pertinent legislative and social factors, this volume highlights the importance of utilizing genetic/genomics knowledge to promote and achieve optimal health in the developing world.

ADVANCE PRAISE FOR Genomics and Health in the Developing World

"This is a book that will be of interest to a broad range of audiences and will, of course, point to important areas of research, comparison between countries, and the development of policy. Genomic science and its technology is crucial to the social and economic development in developing countries as well as to their health care systems."
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—MUIN J. KHOURY, MD, PHD, Director, Office of Public Health Genomics, Centers for Disease Control and Prevention, Atlanta, Georgia, USA

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INTRODUCTION

The Global Burden of Disease Project showed birth defects as one of the leading causes of disability (Murray and Lopez, 1998).

The practice of assessing a population's health from the mortality data may lead to misallocation of health resources and underestimate the needs of patients with chronic disabilities (Modell and Kuliev, 1998). Congenital anomalies or birth defects are one of the most common causes of disability in developed and developing countries. Data on birth defects from population-based studies originating from developing countries in the Asia-Pacific region are lacking (Penchaszadeh, 2000). The March of Dimes estimated 7.9 million infants are born each year with a serious birth defect; of these births, 94% occur in middle- and low-income countries (Christianson et al., 2006). The estimated birth prevalence ranges from 40/1000 live births (high-income countries) to 82/1000 live births in low-income countries (WHO-MOD, 2006). Annually, over 3.3 million children under age five die from birth defects. This chapter deals with birth defects and related health issues in the Asia-Pacific region, which includes countries in East Asia and Southeast Asia with a combined population of two billion, accounting for one-third of the global population.

In many developing countries in the Asia-Pacific region, many genetic tests and chromosome studies are not available routinely or easily. Therefore, the main diagnostic tool in clinical dysmorphology in developing countries rests on clinical evaluation (Christianson and Modell, 2004). Often this leads to the second major issue in clinical dysmorphology in developing countries—the paucity of clinical photographs and information on dysmorphology in the diverse population in the Asia-Pacific region. The majority of patient photographs in dysmorphology databases and textbooks contain patient data largely derived from Western or Caucasian populations (Hall et al., 1989; Jones, 1997; Gorlin et al., 2001). Therefore, subtle dysmorphic features in Asians are often difficult to interpret, and many dysmorphologists and clinical geneticists in this region often have to rely on personal experience and clinical acumen to make the syndromic diagnosis (Thong et al., 2005). Furthermore, in addition to the limited expertise in the field of clinical dysmorphology, there are limited resources for investigations and management of these conditions.

A major birth defect (also known as congenital disorder) is an abnormality of prenatal origin that, if uncorrected or uncorrectable, significantly impairs normal physical or social function or reduces normal life expectancy. This may be structural defect, i.e., related to problems of the body parts or functional birth defects, such as disorders involving the central nervous system (learning and behavioral difficulties, speech or language disorders, seizures and movement disorders), inborn errors of metabolism, and degenerative conditions. The etiology for birth defects is heterogeneous (ICBDSR, 2017, Table 62-1).

A minor birth defect is an abnormality of prenatal origin that is present at birth and does not have surgical, medical, or cosmetic importance at the time of examination. Examples are preauricular pits and umbilical hernia. The presence of three or more minor birth defects may suggest the possibility of a syndrome (Leppig et al., 1987).

Multiple birth defects are found in 1% of all newborns and the majority are associated with dysmorphic features. Of these, about 30%–40% can be diagnosed as having a specific or recognizable syndrome, while the remainder have unknown entities that will require more time and further testing before they may be delineated (Figure 62-1). The study of human birth defects or congenital malformations, in particular with its emphasis on structural abnormalities of development, was first known as dysmorphology as described by Dr. David Smith (Jones, 1997). Clinical dysmorphology refers to the study and care of individuals with multiple birth defects and their families. There are nearly 300 well-described syndromes in Smith's Recognizable Patterns of Human Malformation (Jones, 1997) and close to