

CHAPTER 2

Health Inequalities

Key Points:

- Health inequalities between the richest and poorest segments of the population exist between countries and within countries.
- Injuries occur in all populations and may be unintentional (like road traffic injuries, falls, drowning, poisoning, and burns) or intentional (violence that is self-directed, interpersonal, or collective).
- Non-communicable conditions such as heart disease and cancer are the most common causes of death in higher-income populations and are becoming more common in low-income populations.
- Infectious diseases and nutritional deficiencies are common causes of death in lower-income populations.

Kofi Annan, former Secretary-General of the United Nations, spoke hopefully at an African summit in 2001: “There has been a worldwide revolt of public opinion. People no longer accept that the sick and dying, simply because they are poor, should be denied drugs that have transformed the lives of others who are better off.”¹ If we truly have accepted that no one should die from easily preventable and treatable illnesses, then why are so many people still suffering and dying?

Bill Gates was moved to become involved in global health work after learning that preventable diseases killed millions of children every year and that few organizations were working to alleviate this injustice. Gates recalled that he “couldn’t escape the brutal conclusion that—in our world today—some lives are seen as worth saving and others are not.”² Other world leaders in politics and business, entertainment stars, and other influential people have begun to speak often and openly about their belief that health inequalities are wrong and about their commitment to do something to mitigate them. Entertainers, for example, have supported fundraisers for AIDS and other causes, and the television series *e.r.* has raised awareness about global health issues by featuring several episodes set in central Africa. This chapter provides an overview of some of the most common causes of illness, death, and disability in the world, and highlights some of the health inequalities that have spurred people to take action.

HEALTH INEQUALITIES

The constitution of the World Health Organization (Appendix II) states that health is a fundamental right of all people regardless of “race, religion, political belief, economic, or social condition.” We could add to that list other categories like age, sex, nationality, immigration status, educational level, occupation, ethnicity, health insurance type, and marital status. Yet significant differences in health continue to exist between groups. Black Americans are much more likely than white Americans to have strokes, hypertension, congestive heart failure, and diabetes.³ Married British and American men live longer and are healthier than unmarried men.^{4,5} A baby born in Sierra Leone in 2005 has a life expectancy of 38 years, while a baby born in Japan can expect to live more than twice as long, to 82 years.⁶ These differences are largely a function of social, political, and economic environments, and not innate biological differences.

Inequalities in health, differences in health experience and health status, exist at many levels. Some of these differences are avoidable, unfair, and unjust and are referred to as **inequities**.⁷ The poor are often more susceptible to illness and injury because of a limited diet, existing infections, and dangerous work and home environments. They are less likely to have the means to prevent illness by, for example, being screened for early-stage cancer or buying a bednet to prevent the mosquito bites that cause malaria infections. Limited money or insurance coverage, inability to take time off from work and home responsibilities, and lack of access to transportation may delay diagnosis and treatment, so by the time poor people see a doctor, their disease conditions tend to be at an advanced stage that is harder to treat. People from marginalized populations may receive inadequate explanations of their health conditions due to language barriers, and often cannot afford referrals for second medical opinions or specialized care.

Each year the World Health Organization publishes updated population and health statistics by country and region and uses those indicators to highlight a particular global health concern.⁸ The 1995 World Health Report focused on poverty and health inequalities. The authors noted that advances in modern medicine have created some of the longest-living and healthiest populations in history, yet the benefits of these discoveries are not available globally:

Beneath the heartening facts about decreased mortality and increasing life expectancy, and many other undoubted health

advances, lie unacceptable disparities in health. The gaps between rich and poor, between one population group and another, between ages and between sexes, are widening. For most people in the world today every step of life, from infancy to old age, is taken under the twin shadows of poverty and inequity, and under the double burden of suffering and disease.⁹

Consider the disparities in the deaths of children during their first five years of life, what health workers call “under-5 mortality.” In high-income countries, 6 of every 1000 children die before their 5th birthday, while in developing countries about 88 of every 1000 children die before reaching this age. In the poorest countries, 120 of every 1000 under-5 children die.¹⁰ The gap in child death rates between income groups has been increasing. Between 1970 and 2000, under-5 mortality rates decreased by 71% in high income countries but by only 40% in low-income countries.¹¹ Figure 2-1 is a world map of life expectancy at birth and Figure 2-2 is a bar graph of life expectancy at birth in each of the six regions of the World Health Organization. (The countries in each region are listed in Appendix I.) The average baby born in the African region can expect to live less than 50 years (and has a high risk of dying in infancy or childhood), while the average baby born in the European, American, or Western Pacific regions can expect to live more than 70 years.

If we look at individual countries, life expectancies are even more disparate. Babies born in 2005 had a life expectancy of less than 40 years in several countries, all in Africa—Swaziland (35 years), Botswana (36), Zimbabwe (37), Lesotho (38), Sierra Leone (38), and Zambia (39).¹² They face a high risk of death during infancy and childhood, and those who survive to adulthood face a high risk of contracting HIV infection and dying prematurely from AIDS. In contrast, babies born in 2005 in more than a dozen European nations and Japan have a life expectancy of 80 years or more.

Survival rates are low in developing countries in part because of limited access to life-saving and life-extending drugs and therapies. Figure 2-3 shows that although North America (the United States and Canada), Europe, and Japan represent only about 18% of the world’s population, these regions account for nearly 88% of drug sales. Although Asia, Africa, the Middle East, and Oceania make up nearly 73% of the world’s population, only about 8% of drug sales are to these regions.

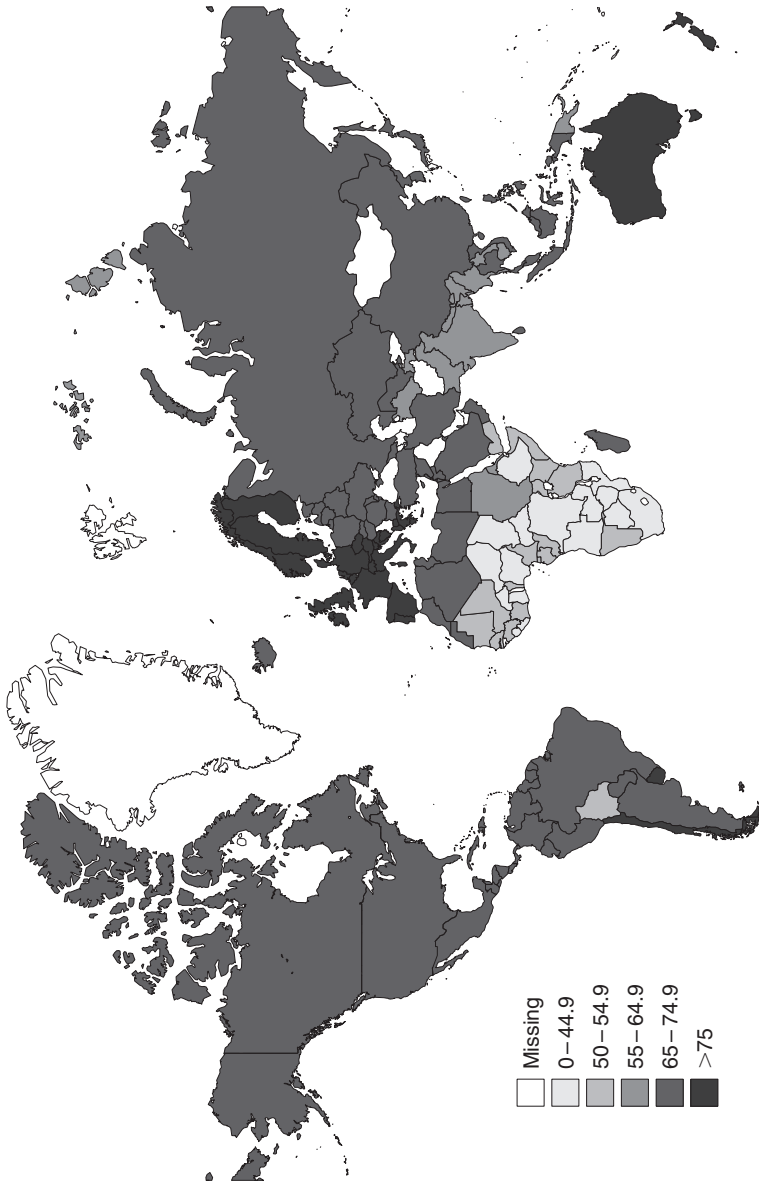


Figure 2-1. Life Expectancy at Birth, 2004.*

Data Source: World Bank, World Development Indicators Database, 2006.

*Map projections may not reflect true differences in land areas between countries.

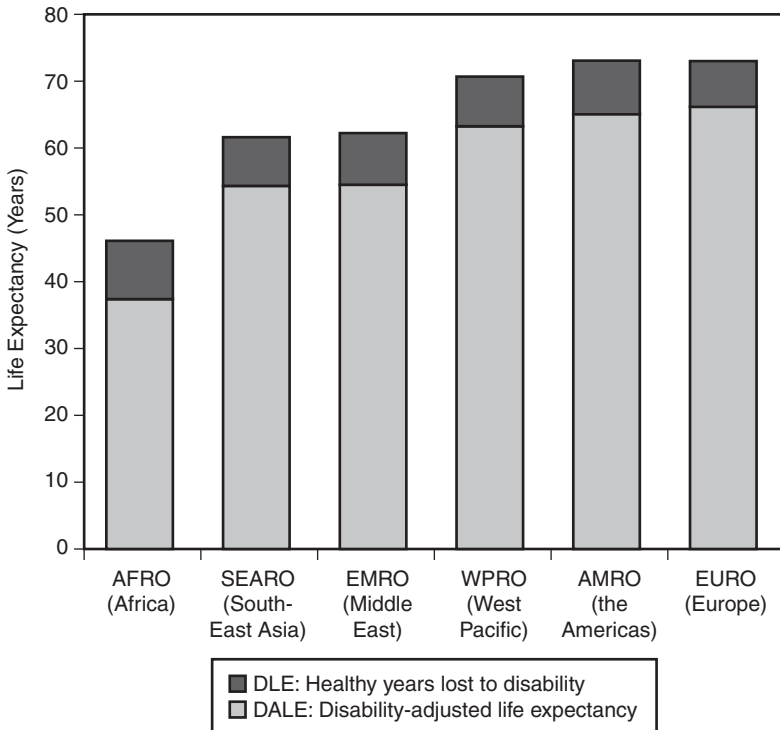


Figure 2-2. Life Expectancy and Healthy Life Expectancy in Each WHO Region, 1999.

The total height of each bar represents total life expectancy at birth. The lighter portion of the bar represents the number of years the average person in the region is expected to live a healthy life and the darker portion of the bar represents years the average person is expected to live with disability.

Data Source: Mathers CD, Sadana R, Salomon JA, Murray CJL, Lopez AD. Estimates of DALE for 191 countries: methods and results. WHO Health Systems Performance Report, Global Programme on Evidence for Health Policy Working Paper No. 16. Geneva: World Health Organization, June 2000.

CAUSES OF DEATH

Pathogenesis is the study of the process of disease. Many different processes can lead to disease, including infectious agents, injuries, malnutrition, and genetics. These exposures are called etiologic agents, or causal agents, and **etiology** is study of the origin of disease. Some diseases

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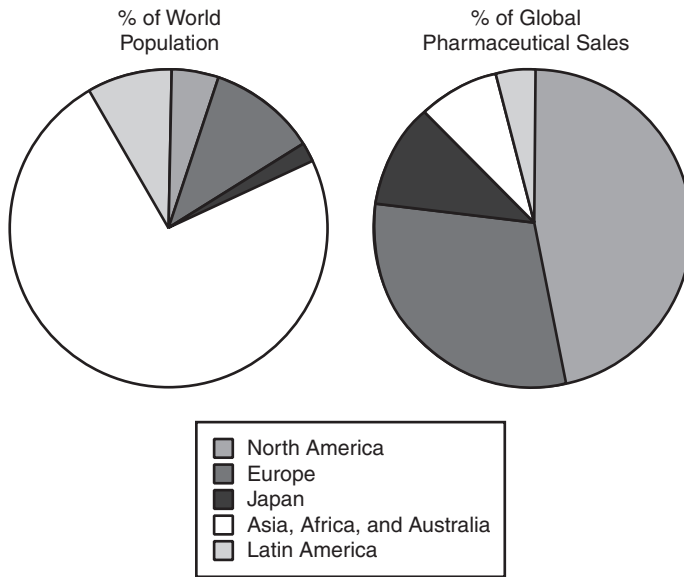


Figure 2-3. Comparison of the Proportion of Global Pharmaceutical Sales by Region to the Proportion of the World Population that Lives in Each Region.

Data Sources: Population Reference Bureau, “Population, Mid-2006.” IMS Health, “Global pharmaceutical sales by region, 2005.”

are classified as **idiopathic**, which means that we do not (at least not yet) know what causes them. The etiology of many neurological conditions like Parkinson’s Disease and epilepsy are unknown in most cases, as are the causes of many mental illnesses, autoimmune diseases, and other conditions.

The World Health Organization uses three main classifications for causes of death and disability: (1) injuries, (2) non-communicable conditions, and (3) communicable (infectious) diseases, maternal (pregnancy-related) conditions, perinatal conditions (diseases of newborns), and nutritional deficiencies.

Injuries

An **injury** is physical damage to the body. An injury may be a fracture of a bone, a strain or sprain of a joint, a brain or spinal cord injury, wounds to the skin, or damage to internal organs. Injuries may be unintentional, such

as a traffic accident, fall, burn, or drowning, or intentional, whether self-inflicted or suffered during an act of violence or war. Intentional acts are further classified as self-directed violence (self-mutilation or suicide), interpersonal violence against family members, intimate partners, or community members, or collective violence such as war, mob violence, gang violence, or terrorist acts (Table 2-1).¹³

Figure 2-4 shows the proportion of deaths due to various types of injuries. About 68% of injury deaths worldwide in 2005 were from unintentional injuries (road traffic accidents, poisoning, falls, fires, drowning, and other unintentional injuries) and 32% from intentional injuries (self-inflicted injuries, violence, and war).

Poor people are at increased risk of injury because they more often live, work, and go to school in unsafe environments. They use overcrowded and poorly maintained vehicles for transportation, are pedestrians on crowded streets, live in homes that are vulnerable to fire, do not have access to preventive tools, and have less access to treatment and rehabilitative services.¹⁴ Environmental exposures to chemicals, like poisons, alcohol, smoke, and heavy metals, or to physical trauma, radiation, heat, cold, or allergens can also cause illness and injury. In addition to immediate health problems that can be caused by exposure, these exposures may cause

Table 2-1. Examples of Types of Injuries.

<i>Unintentional Injuries</i>	<i>Intentional Injuries</i>
<ul style="list-style-type: none"> ● Road traffic injuries ● Falls ● Burns ● Drowning ● Poisoning 	<p><i>Self-directed violence</i></p> <ul style="list-style-type: none"> ● Suicide ● Cutting <p><i>Interpersonal violence</i></p> <ul style="list-style-type: none"> ● Spousal / intimate partner abuse ● Child abuse ● Elder abuse <p><i>Collective violence</i></p> <ul style="list-style-type: none"> ● War ● Terrorist acts ● Gang violence

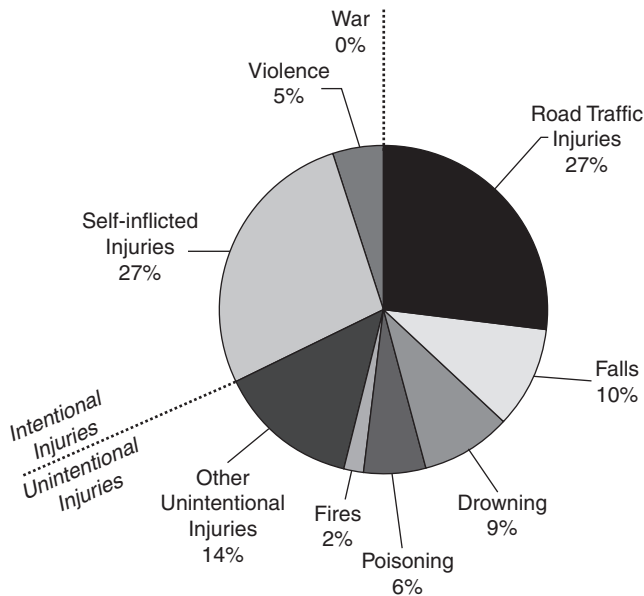


Figure 2-4. Proportion of Various Causes of Death Worldwide due to Injury, 2005 Estimates.

Data Source: WHO, Global Burden of Disease database, 2006 update.

long-term disability and lead to the development of other chronic conditions. For example, radiation exposure can cause cancer and repeated alcohol exposure can cause cirrhosis of the liver. Harmful exposures are a common problem in low-income nations where there is limited access to clean water and trash removal, where food must be prepared over an indoor fire, and where enforcement of safety laws is minimal. (Chapters 10 and 11 explain the environmental context of health.)

Males are significantly more likely than females to die from all types of injuries other than burns and self-inflicted injuries (Figure 2-5). Men are at greater risk of injury because they are more likely than women to work in hazardous occupations, to participate in dangerous recreational activities, to spend time on the road, to use alcohol, and to be involved in armed conflict. Because women spend more time cooking, they are more likely to fall into fires or scald themselves by dropping a pot of boiling liquid. Females are also more likely than males to be abused by intimate partners and family members, and the rate of injuries due to physical and sexual violence is probably under-reported.

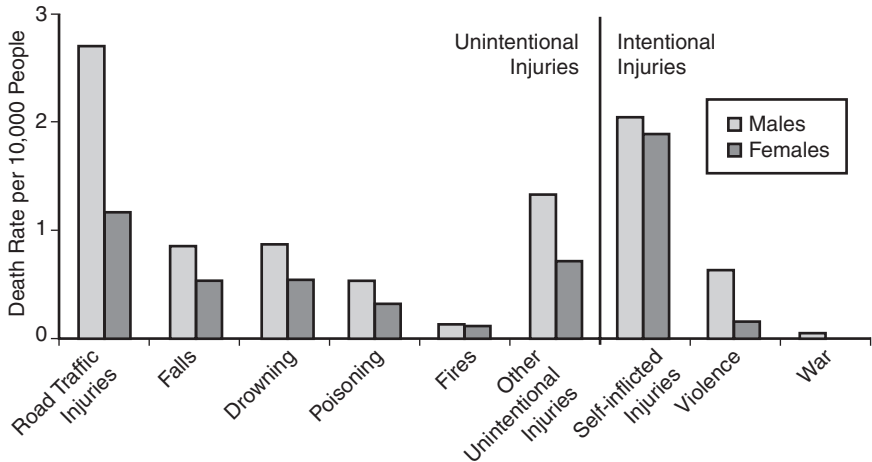


Figure 2-5. Sex-specific Death Rate for Specific Injury-related Causes Worldwide, 2005 Estimates.

Data Source: WHO, Global Burden of Disease database, 2006 update.

Most people who are injured survive their injuries, but many are left with permanent disabilities. Although some injuries are the result of accidents, many injuries could have been prevented by the use of safety belts in motor vehicles, child car seats, helmets, designated drivers who have not consumed alcohol, flame-resistant clothing, smoke detectors, fencing around water bodies, swimming lessons, protective eyewear, use of safety harnesses when working at dangerous heights, and locked storage of weapons and ammunition.¹⁵

Non-communicable Conditions

Non-communicable conditions are diseases that are not contagious (Table 2-2), such as cancers (sometimes called “neoplasms” in health reports), endocrine disorders, neurological and psychiatric disorders, heart disease and other cardiovascular disorders, chronic respiratory diseases, digestive diseases, kidney diseases, and musculoskeletal and skin diseases. Non-communicable conditions may be caused by genetic disorders or congenital abnormalities or may be related to aging processes or autoimmune disorders. The causes of many non-communicable diseases are not yet

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Table 2-2. Examples of Non-communicable Conditions.

Cancers (Neoplasms)	<ul style="list-style-type: none"> ● Lung cancer ● Stomach cancer ● Colorectal cancer ● Liver cancer ● Breast cancer ● Esophageal cancer 	<ul style="list-style-type: none"> ● Lymphoma ● Mouth and throat cancer ● Prostate cancer ● Leukemia ● Cervical cancer ● Pancreatic cancer ● Bladder cancer 	<ul style="list-style-type: none"> ● Ovarian cancer ● Uterine cancer ● Melanoma and other skin cancers
Endocrine and nutritional / metabolic disorders	<ul style="list-style-type: none"> ● Diabetes mellitus 	<ul style="list-style-type: none"> ● Thyroid disease ● Disorders of the pituitary gland 	<ul style="list-style-type: none"> ● Adrenal disorders (Addison Disease, Cushing's Syndrome)
Neuropsychiatric disorders	<ul style="list-style-type: none"> ● Alzheimer's and other dementias ● Epilepsy ● Parkinson's disease ● Multiple sclerosis ● Migraine ● Cerebral palsy ● Schizophrenia 	<ul style="list-style-type: none"> ● Depressive disorder ● Bipolar disorder ● Panic disorder ● Obsessive-compulsive disorder ● Huntington Disease ● Alcoholism 	<ul style="list-style-type: none"> ● Drug addiction ● Eating disorders ● Insomnia ● Post-traumatic stress disorder ● Tourette Syndrome ● Autism
Sense organ disorders	<ul style="list-style-type: none"> ● Blindness ● Cataracts 	<ul style="list-style-type: none"> ● Glaucoma ● Pterygium 	<ul style="list-style-type: none"> ● Deafness

clear. Non-communicable conditions are sometimes called chronic diseases because many of them develop gradually and then last for a long time.

Some non-communicable conditions are caused by immune system dysfunctions. The immune system helps the body to fight disease by recognizing and attacking invaders like infectious agents and allergens. Autoimmune disorders occur when the body has trouble distinguishing between “self” and “non-self” and begins to attack its own cells. Lupus (systemic lupus erythematosus) and rheumatoid arthritis are autoimmune diseases. Allergies are another type of immune dysfunction in which the body is hypersensitive to substances that are usually not harmful. Another “internal” disease process is inflammation. Although the process is not yet well understood, it appears that inflammation plays a role not just in arthritis and other conditions in

Table 2-2. Examples of Non-Communicable Conditions—cont'd.

Cardiovascular diseases and blood disorders	<ul style="list-style-type: none"> ● Ischemic heart disease (heart attacks) ● Cerebrovascular disease (strokes) ● Hypertension 	<ul style="list-style-type: none"> ● Inflammatory heart disease ● Congestive heart failure ● Hemophilia ● Anemia 	<ul style="list-style-type: none"> ● Hemochromatosis ● Sickle cell ● Thalassemia
Respiratory diseases	<ul style="list-style-type: none"> ● Chronic obstructive pulmonary disease (COPD)—emphysema, chronic bronchitis, and chronic asthma 		<ul style="list-style-type: none"> ● Asthma ● Cystic fibrosis
Digestive diseases	<ul style="list-style-type: none"> ● Appendicitis ● Liver cirrhosis ● Gallstones 	<ul style="list-style-type: none"> ● Hernias ● Peptic ulcer disease 	<ul style="list-style-type: none"> ● Hemorrhoids ● Inflammatory Bowel Disease
Oral diseases	<ul style="list-style-type: none"> ● Dental caries (cavities) 	<ul style="list-style-type: none"> ● Periodontal disease 	<ul style="list-style-type: none"> ● Edentulism (toothlessness)
Genitourinary diseases	<ul style="list-style-type: none"> ● Nephritis (kidney disease) 	<ul style="list-style-type: none"> ● Benign prostatic hypertrophy (BPH—enlarged prostate) 	<ul style="list-style-type: none"> ● Pelvic inflammatory disease (PID)
Skin diseases	<ul style="list-style-type: none"> ● Albinism ● Acne 	<ul style="list-style-type: none"> ● Eczema 	<ul style="list-style-type: none"> ● Psoriasis
Musculoskeletal diseases	<ul style="list-style-type: none"> ● Muscular dystrophy ● Osteoarthritis 	<ul style="list-style-type: none"> ● Rheumatoid arthritis 	<ul style="list-style-type: none"> ● Osteoporosis

which swelling is easily observed, but also in heart disease and possibly in diabetes, cognitive disorders, and many other conditions. Poor nutrition and some infections (like HIV) can also cause immune system suppression and lead to other illnesses.

Genetic make-up also plays a key role in health.¹⁶ Many genetic disorders are present at birth, and some of the symptoms caused by these genetic disorders can be avoided through early testing and lifestyle adjustments. A person with cystic fibrosis can receive percussion therapy, which involves “drumming” the chest to help clear out mucus. A boy known to have hemophilia can be treated with injections of blood clotting factors. Blood tests at birth can identify metabolic disorders, so that potentially harmful foods can be avoided. For example, infants can be tested for phenylketonuria

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(PKU), a metabolic disorder in which infants and children with a particular version of a gene found on the 12th chromosome do not make an enzyme that is needed to metabolize phenylalanine, an amino acid that is found in some proteins. A blood test done just after the baby is born can let parents know if their child has PKU, so they can help the child avoid phenylalanine and develop normally. If parents do not know that their child has the genetic disorder and feed their children foods that contain phenylalanine, the children can develop mental retardation. A simple blood test for PKU is mandated for newborns in many high-income countries, but is not available in most low-income countries.

Other conditions are caused by genetic **mutations** (permanent changes in the sequence of bases that make up DNA) that occur after birth. These mutations may result from exposure to radiation, chemicals, pollutants, or other substances. Cancers generally involve a series of mutations that can lead to tumor development. A chemical that can cause a mutation and lead to cancer is called a **carcinogen**, and the process of cancer development is **carcinogenesis**.

Birth defects, developmental disabilities, and other disabilities are common in both high- and low-income areas, but the challenges for disabled individuals and their families are greater in resource-poor settings. Furthermore, as the global population ages, the burden of non-communicable diseases such as heart disease, stroke, vision and hearing loss, and cancer will increase. When a person in a developing country develops a non-communicable disease like cancer, it is often diagnosed at an advanced stage and the treatment options that are available are limited. As a result, the survival rate following diagnosis is significantly lower in developing countries than in developed countries. (These inequalities are discussed in more detail later in the chapter.)

Infectious Diseases and Other Conditions

The third WHO category for causes of death is a broad one, and includes infectious diseases, pregnancy-related conditions, diseases of newborns, and nutritional deficiencies.

Communicable diseases are caused by infectious agents such as bacteria, viruses, fungi, and helminths (worms), and can be passed from an infected person to a susceptible person. Deaths from infectious diseases like tuberculosis, sexually transmitted infections, diarrheal diseases, meningitis,

malaria, and respiratory infections are all in this category. In the early and middle part of the twentieth century, the use of microscopes and the development of new laboratory techniques led to the identification of many specific microbes and the development of vaccines and antibiotics like penicillin. These discoveries generated a great deal of optimism and confidence in our ability as humans to control and eradicate communicable diseases, leading the U.S. Surgeon General to declare in 1967 that “the war against infectious diseases has been won!” Unfortunately, this declaration was premature. Although modern science has given us a good understanding of the infectious disease process and allowed us to develop therapies and cures for many types of infection, we now know that microbes continue to adapt, to develop, and to emerge. Even with improved prevention and therapeutic techniques, infectious diseases continue to be a health risk in all populations. Developing new vaccines, treatments, and prevention methods remains an important part of global health. (Infectious diseases are discussed in detail in Chapters 6, 7, and 8.)

Maternal deaths are those related to pregnancy, childbirth, and the hours after giving birth. Perinatal conditions include low birthweight, premature (early) birth, birth asphyxia, and birth trauma. (Maternal health and infant health are discussed in Chapter 4.)

Malnutrition may result when a person does not consume nutrients in the right quantities and combination. Taking in too few, or too many, calories or nutrients can be a cause of poor health. Common nutritional deficiencies include protein-energy malnutrition and diseases related to particular vitamins and minerals such as iron, vitamin A, zinc, and iodine. Undernutrition, deficiencies in energy or nutrients, contributes to more than half of child deaths worldwide each year.¹⁷ (Nutrition is discussed in more detail in Chapter 9.)

INEQUALITIES IN CAUSES OF DEATH

Figure 2-6 shows the breakdown of deaths by cause in each of the WHO regions. The proportion of deaths due to injuries is roughly equal across groups, but less developed regions like Africa have a very high proportion of deaths due to infectious disease and undernutrition while the large majority of deaths in Europe are due to non-communicable conditions like heart disease and cancers. Everyone will eventually die of something, but because infectious diseases are usually associated with premature death and noncommunicable

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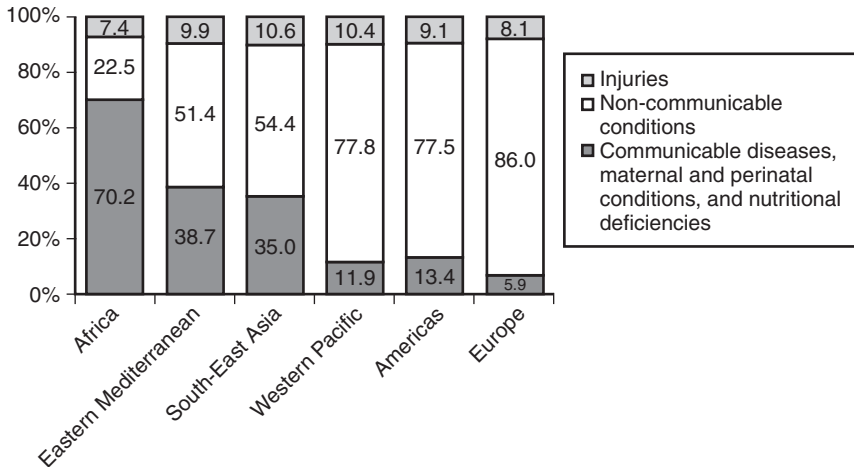


Figure 2-6. Percent of Deaths due to Injury, Non-communicable Disease, and Communicable Disease by WHO Region (2005 Estimates).

Data Source: WHO, Global Burden of Disease database, 2006 update.

conditions are associated with aging, a region with a large proportion of non-communicable deaths is generally considered to be healthier than a region with a majority of deaths from infectious diseases. There are also proven ways to prevent and cure many infectious diseases, so a large portion of infectious disease deaths could have been averted. (Immunization and the boiling of contaminated water are specific and immediate methods of preventing some infectious diseases. Some cases of cancer and other chronic conditions can be prevented, too, by avoiding tobacco products, eating a healthier diet, and exercising, but these are inexact and long-term methods of staying healthy.)

Figure 2-6 makes it look like less developed regions of the world (Africa, the Middle East, and South-East Asia) bear the burden of infectious disease, while industrialized regions (Europe, the Americas, and the western Pacific) bear the burden of non-communicable conditions. This is a bit misleading, because most of the world's population lives in less developed countries.

Figure 2-7 presents the same data as Figure 2-6, but shows the *number* of deaths by region and cause rather than the *percent* of deaths. A comparison of these graphs shows, for example, that although the percent of deaths from non-communicable diseases is lower in South-East Asia (54.4%) than

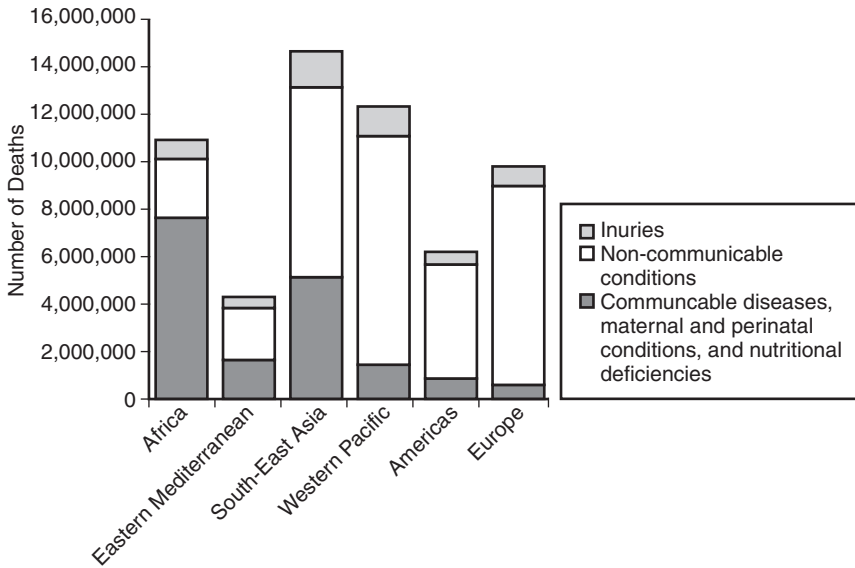


Figure 2-7. Number of Deaths due to Injury, Non-communicable Disease, and Communicable Disease by WHO Region.

Data Source: WHO, Global Burden of Disease database, 2006 update.

in the Americas (77.5%) many more people who live in South-East Asia (7.9 million) die from non-communicable diseases each year than people who live in the Americas (4.8 million). In fact, nearly 80% of the deaths from chronic disease¹⁸ worldwide each year are in low- and middle-income countries, including more than 50% of cancers.¹⁹ Cardiovascular (heart) disease, a non-communicable condition, is already the leading cause of death in developing countries.²⁰

The difference between the incidence rates of various types of cancer (the percent of people in a population who are diagnosed with cancer in one year) and the number of deaths from these cancers in low-income and high-income regions is particularly striking. The percentage of people who live in high-income countries who develop cancer is higher than the percentage of people who live in low- and middle-income countries who develop cancer (Figure 2-8), partly because cancer is more common in older adults and more people in high-income countries reach older ages. But the number of cancer deaths in low- and middle-income countries is higher than the number

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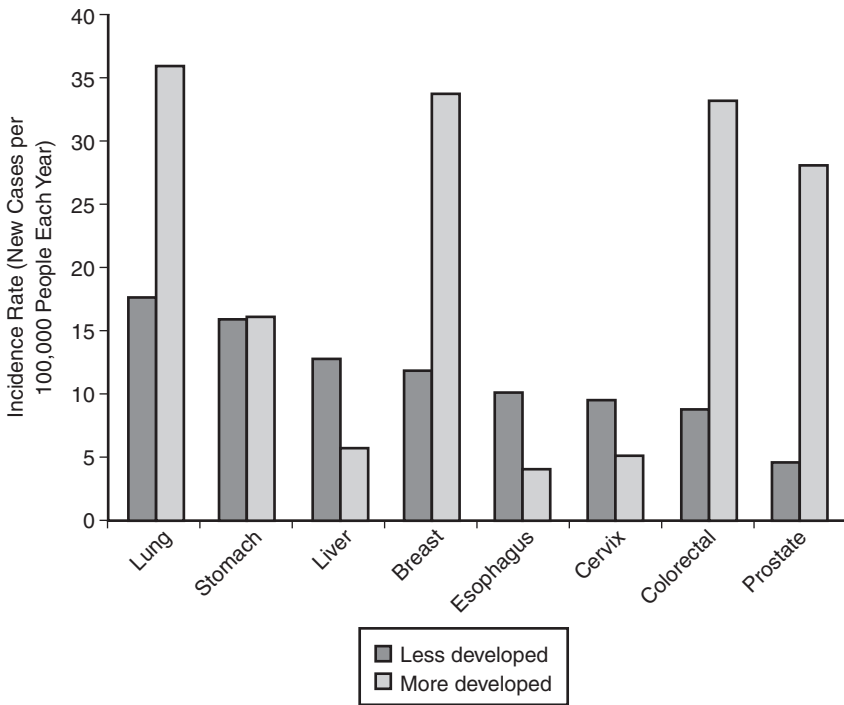


Figure 2-8. Cancer Incidence Rates (Number of New People in a Year Who Are Diagnosed with the Disease per 100,000 People in the Population) in 2002 in Less Developed and More Developed Countries.

For most types of cancer, the incidence rate is higher in more developed countries.

Data Source: Kamangar F, Doros GM, Anderson WF. Patterns of cancer incidence, mortality, and prevalence across five continents: defining priorities to reduce cancer disparities in different geographic regions of the world. *J Clin Oncol* 2006;24:2137–2150.

of deaths in high-income countries (Figure 2-9). This means, for example, that although a person who lives in a more developed country is more likely to be diagnosed with lung cancer or breast cancer than a person who lives in a less developed country (Figure 2-8), the total number of deaths each from these cancers is very similar in less developed and more developed regions (Figure 2-9).²¹

If cancer patients in low-income countries had access to the same screening and diagnostic tests (which allow for detection of cancer at an earlier,

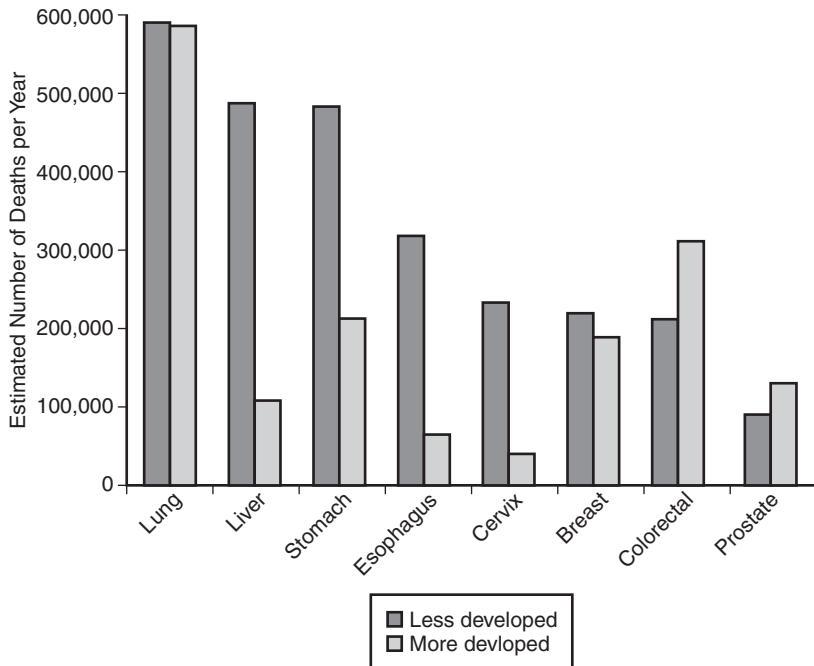


Figure 2-9. Estimated Average Number of Cancer Deaths in 2002 in Less Developed and More Developed Countries.

For most types of cancer, more deaths each year occur in less developed countries than in more developed countries.

Data Source: Kamangar F, Doros GM, Anderson WF. Patterns of cancer incidence, mortality, and prevalence across five continents: defining priorities to reduce cancer disparities in different geographic regions of the world. *J Clin Oncol* 2006;24:2137–2150.

more treatable stage) and advanced therapies (like chemotherapeutic drugs, surgical techniques, and radiology regimens) as cancer patients in high-income nations, many more people with cancer would survive. At present, the survival rate for people diagnosed with cancer in developing countries is much lower than the survival rate in industrialized nations (Figure 2-10).

Life expectancy will increase when more people gain access to technologies that allow for early diagnosis and effective treatment. One of the goals of global health is to increase healthy life expectancy, so it is important to make technology more widely available in all countries.

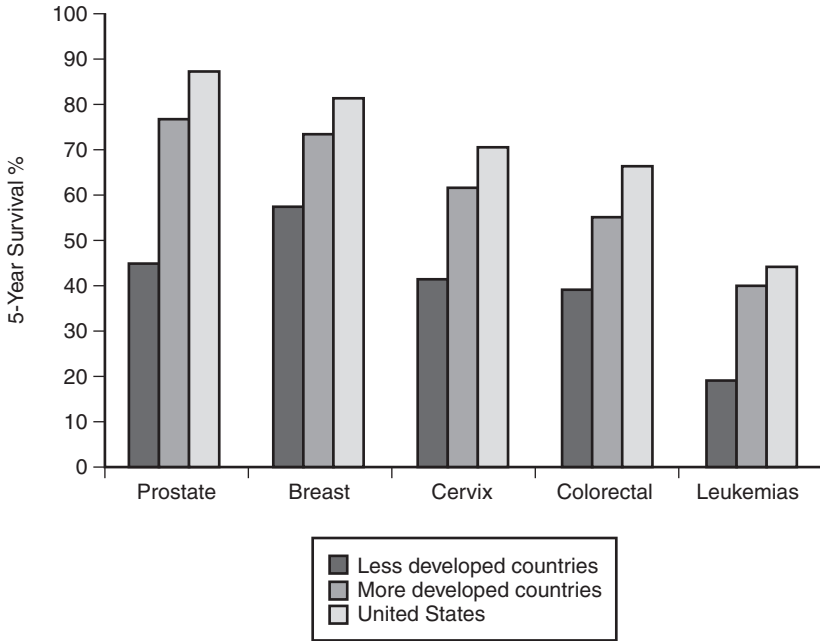


Figure 2-10. Estimated Age-adjusted 5-year Cancer Survival Rate for Selected Cancers.

Data Source: Parkin DM, Bray F, Ferlay J, & Pisani P. Global cancer statistics, 2002. *CA Cancer J Clin* 2005;55:74-108.

Global cancer statistics are compiled by IARC, the International Agency for Research on Cancer. Cancer statistics from the United States are compiled by the SEER (Surveillance, Epidemiology, and End Results) registry sponsored by the National Cancer Institute of the National Institutes of Health.

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16. Supplemental information on genetics and genetic disorders:

Genes are sequences of nucleic acids that are part of the chromosomes that are found in the nucleus of every cell in our bodies. This genetic material directs every function of the body, including cell replication, which is important for healing as well as for growth and development. Each cell contains identical DNA (deoxyribonucleic acid), although only parts of the code are active in certain cells, which is why cells in your heart form different kinds of tissue than the cells that line your intestine. Your genotype is the version of a gene you carry and your phenotype is the way a characteristic that results from having a particular allele, or version of a gene, is expressed. Phenotype can refer to physical appearance, the way a person develops or functions physiologically, or disease status.

Some alleles are dominant, which means that inheriting a copy of that allele from either parent will cause you to display the phenotype associated with that allele. Huntington Disease, which causes a progressive degeneration of brain cells, is the result of inheriting a copy of a defective gene found on chromosome 4 from one parent. Some alleles are recessive, which means that a person must inherit a copy of the allele from both parents to display the phenotype associated with the allele. Cystic fibrosis is a recessive disorder carried on the 7th chromosome that causes excessive mucus production in the lungs and digestive tract. Sickle cell and thalassemia are genetic blood disorders that may confer some protection against malaria but can cause death if not treated. A person who inherits the sickle cell allele from both parents develops sickle cell anemia because the red blood cells, which carry oxygen to all the cells in the body, become misshapen and impede blood flow. A person in sickle cell crisis experiences extreme pain and possible organ damage due to oxygen deprivation.

Some conditions are caused by the presence of an extra chromosome or a missing part of a chromosome. Down Syndrome (trisomy 21) is the result of an extra 21st chromosome. Most humans have 23 pairs of chromosomes: 22 pairs of autosomal chromosomes and one pair of sex chromosomes, usually XX for females and XY for males. Sometimes there are extra copies of an X or Y chromosome or a missing X. Males may have an extra X (Klinefelter Syndrome) and develop female characteristics or have an extra Y (Jacob Syndrome), which may cause speech and reading problems. Females may have an extra X (Poly-X Syndrome) or a missing X (Turner Syndrome) and have developmental delays.

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- Some sex-linked genetic disorders are carried by healthy women who can pass the trait on to their sons. Hemophilia, a blood clotting disorder, and Duchenne muscular dystrophy, which causes progressive muscle weakness, are examples of X-linked recessive disorders. Some genetic disorders are caused by deletions, duplications, translocations, or inversions of parts of chromosomes. Cri du Chat Syndrome is the result of missing a portion of the 5th chromosome and causes a malformed larynx and mental retardation.
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