

## **EPIDEMIOLOGY: HEALTH AND DISEASE IN POPULATIONS**

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### **Summary**

Epidemiology concerns the study of health and disease in populations and is a central discipline in both clinical medicine and public health. It is the study of the distribution, frequency, and determinants of health-related conditions in populations and the application of this study to control disease. Although some basic notions concerning epidemiology have a long history, epidemiology as a defined discipline is quite recent and its methods and uses have been rapidly evolving, especially since about 1980.

Epidemiology provides essential concepts and tools needed to understand the causes and underlying contributing factors to disease such that the most effective methods for promoting health and preventing or treating disease can be achieved.

To solve the many issues of public health requires the interaction of epidemiology with many disciplines including the clinical sciences, bio-statistics, economics, and the social sciences. This article takes a broad view of the boundaries of epidemiological methods and tools. Much of the demand for new epidemiological tools and stronger epidemiological evidence for action derives from the broad array of forces that are driving health sector reform throughout the globe.

The role of epidemiology in the health sciences is manifold. It provides the vocabulary and central concepts to describe the pattern of health and disease in a population and its changes over time. It provides the basis for defining and measuring the health status and the burden of disease within a community and the consequences that ensue. Disease states must be defined, classified, and counted. Knowledge of frequency, duration, and consequences of disease in terms of morbidity and mortality are required. An adequate description must include the who, when, and where and how these factors change over time and in response to what.

Epidemiological methods have been the principal means to help understand causes of diseases and the factors that may influence their outcome. "Cause" is a term that has many levels of meaning in epidemiology. To have a rational basis for preventing or intervening in the development of disease, an understanding of the cause or risk factors of the disease is essential.

Another major domain of epidemiology is interventions and systems of interventions directed at safeguarding and promoting health and preventing or overcoming disease and its consequences. Epidemiological methods are needed for assessment of new vaccines for prevention, and treatments for curing a disease. Epidemiology is vital in guiding and evaluating health planning, resource allocation, and health policy decisions. This article reviews the concepts and methods of epidemiology as used for these many purposes and points to some of the new directions the discipline is taking.

## **1. What is Epidemiology?**

Epidemiology, consisting of the principles and methods for the study of health and disease in populations, is a central discipline in public health. A widely used definition is that epidemiology is the study of the distribution, frequency, and determinants of health-related states or events in specified populations and the application of this study to control health problems[1]. Although important approaches to measuring the health of populations have roots dating back many centuries, epidemiology as a defined discipline is quite recent and has been rapidly evolving in its methods and uses, especially since the early 1980s. Epidemiology provides essential concepts and tools for understanding the causes and underlying contributing factors to disease such that the most effective methods for promoting health and preventing or treating disease can be achieved.

Public health is usually viewed in somewhat broader terms than epidemiology; the United States Institute of Medicine's 1988 report *The Future of Public Health* defined public health as a mission, "the fulfillment of society's interest in assuring the conditions in which people can be healthy" and stated that the substance of public health consisted of "organized community efforts aimed at the prevention of disease and the promotion of health"[2]. There has been some controversy as to the exact role epidemiology as a scientific discipline plays in public health so defined. But there should be no controversy that to solve the many issues of public health requires the interaction of epidemiology with many disciplines including the clinical sciences, biostatistics, economics, and the social sciences. This article takes a very broad view of the boundaries of epidemiological methods and tools including the use of epidemiology and its methods in the planning, managing, and assessment of health care provision. Indeed much of the demand for new epidemiological tools and stronger epidemiological evidence for action derives from the broad array of forces that are driving health sector reform throughout the globe.

## 2. Purposes of Epidemiology

All epidemiology can be divided into three parts:

1. Epidemiology provides the vocabulary and central concepts to describe the pattern of health and disease in a population and its changes over time. What is the burden of disease within a community and what is the best way to express it? In order to discuss these issues one must have clear definitions of health and disease and of the population in which they occur. Disease states must be defined, classified, and counted. Knowledge of frequency, duration, and consequences of disease in terms of morbidity and mortality are required. And an adequate description must include the who, when, and where and how these factors change over time and in response to what. How is infection transmitted from one person to another? What is the natural history and prognosis of disease? These issues are discussed in **Section 3. Defining and Measuring Health and Disease** and **Section 4. Descriptive Epidemiology**.
2. Epidemiological methods have been the principal means to help understand causes, risk factors, and contributing or predisposing conditions that influence disease. Knowledge of how an infection is transmitted from one person to another is central to knowing how to reduce transmission of many diseases. Much of epidemiology is devoted to understanding the etiology of disease. "Cause" is a term that has many levels of meaning in epidemiology that are discussed further in **Section 5. Epidemiological Approaches to Understanding Causal Relations**. Factors are said to be necessary causes if a specific factor is always present before disease initiation, but it may not be sufficient. For example, pulmonary tuberculosis requires infection with *Mycobacteria tuberculosis*, but many are infected with *M. tuberculosis* without having disease. On the other hand, a factor may be sufficient if disease always occurs when it is present, but it may not be necessary if disease can also occur in its absence. An example is severe iron deficiency that will always result in anemia, but anemia may be "caused" by many other factors even when iron levels are normal. To have a rational basis for preventing or intervening in the development of disease, an understanding of the cause or risk factors of the disease is essential.
3. The third major domain of epidemiology is interventions and systems of interventions directed at safeguarding and promoting health and preventing or

overcoming disease and its consequences. Epidemiological methods are required for assessment of new interventions or combinations of intervention programs through clinical and field trials, which are discussed in **Section 6. *Experimental Epidemiology: The Randomized Trials***. Further epidemiological methods and information are important in guiding and evaluating health planning, resource allocation, and public health policy decisions, particularly as tools making use of composite indicators that combine measures of morbidity and mortality are developed. These measures can provide a common denominator for comparing the impact that different diseases may have on populations. New approaches to managing health programs also are requiring new ideas and methods to measure and assure that health program interventions are indeed being carried out according to evidence-based guidelines and that they are being applied equitably to all who would benefit from receiving them (see **Section 7. *Epidemiology for Health Systems: Use in Policy, Planning, and Assessment***).

### 3. Defining and Measuring Health and Disease

The first step towards understanding the basis of epidemiology is to agree on definitions for health and disease. In its charter in 1948, the World Health Organization (WHO) defined health as not merely the absence of disease, but rather in positive terms as “A state of complete physical, mental and social well-being and not merely the absence of disease or infirmity.” Although this is an important ideological conceptualization, it has not been operationally useful. For most epidemiological purposes, objectives of health programs are more readily defined in terms of prevention or treatment of disease.

Disease has been defined in many ways. Generally illness, sickness, and disease are used interchangeably, but some make distinctions. For example, Susser[3] recommends that *disease* represents a physiological or psychological dysfunction, while *illness* is a subjective state of the person who is experiencing a state of not feeling well, and *sickness* is a state of social dysfunction that the individual assumes when not feeling well. But for purposes of defining and measuring disease in general, a broad definition may be most useful. Disease is anything that an individual (or population) experiences that causes, literally, “dis-ease” (i.e. anything that leads to discomfort, pain, distress, disability, or death constitutes disease for any reason including injuries or psychiatric disabilities).

Epidemiological notions and tools are used to elucidate the natural history of disease. Most specific diseases have a characteristic pattern from onset through progression of the disease process to termination of the process either through recovery or death. There is wide variation in the patterns of disease evolution. The onset of disease usually will be dated from the start of symptoms or signs as determined by the individual afflicted, a family member, a medical practitioner, or as the result of a lab test. Figure 1 illustrates healthy life lost from disability and from premature death due to typical cases of cirrhosis, polio, and multiple sclerosis in terms of onset, extent, and duration of disability and termination. The conclusion of the disease process depends on a host of factors from correct diagnosis to appropriate treatment. Possible outcomes include clinical recovery with complete disappearance of clinical signs and symptoms, recovery from the acute phase of disease but with residual effects such as paralytic polio, or death

primarily as a result of the disease. The latter includes death directly caused by the disease and that indirectly brought about as a result of complication such as heart disease as a complication of diabetes. Termination of a disease state may also be marked by recovery followed by progression to another disease such as cirrhosis following hepatitis infection.

### 3.1. Disease Nomenclature and Classification

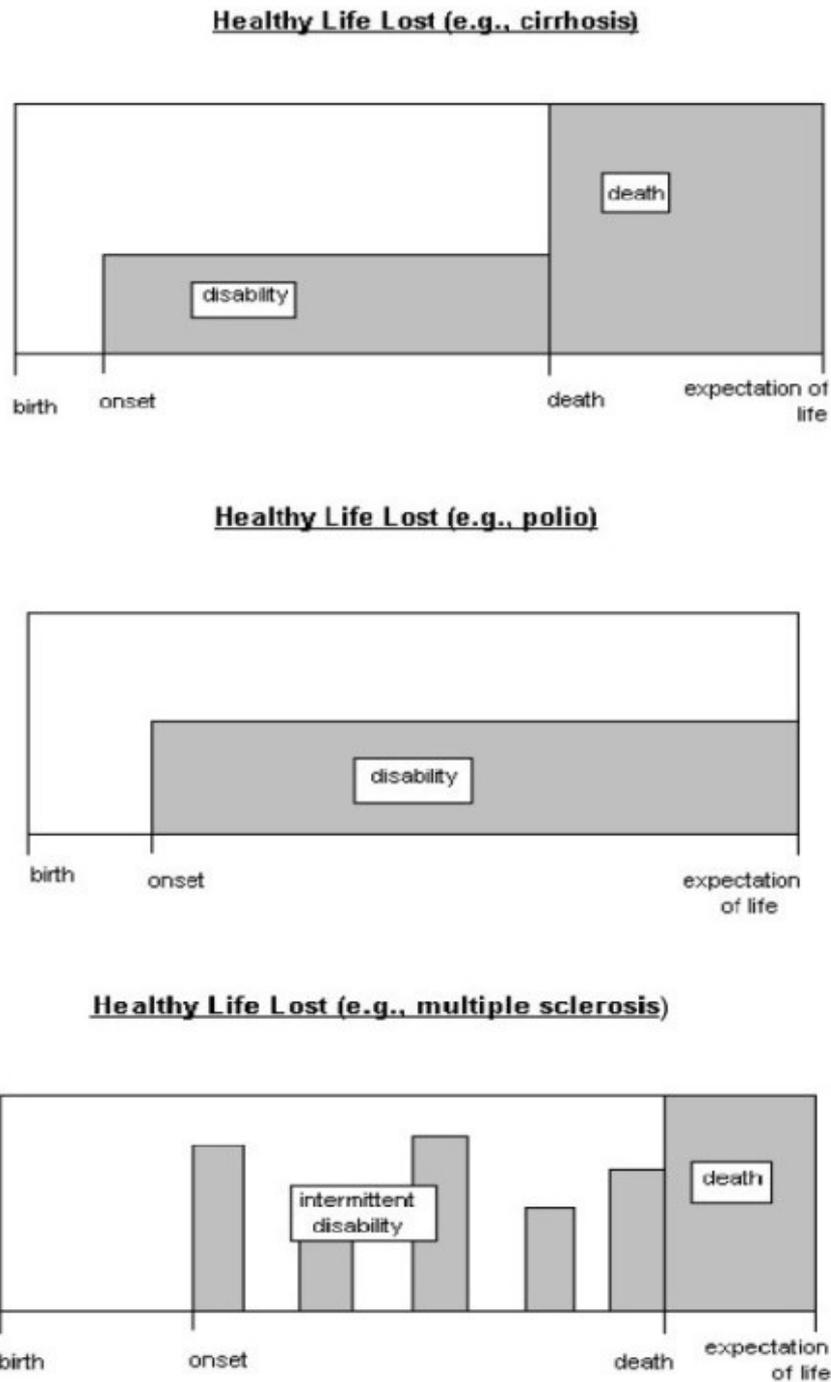


Figure 1. Patterns of healthy life lost

Diagnosis and classification of specific diseases is central in determining what health intervention programs would be most useful. Understanding the pathogenesis of the disease process and defining (classifying, categorizing, and/or diagnosing) disease is critical to understand and classify “causes” so that the most effective prevention and treatment strategies for reducing the effects of a disease or risk factor can be selected.

The nomenclature and classification of diseases undergo continuing change as our understanding of disease processes and their causes advances. In general, diagnostic classification is based upon two general, quite different, characterizations of disease. The first is a description of the pathological features of the disease process. Examples include pneumonitis (inflammation of the lung) or pancreatic carcinoma. The second depends upon the underlying causal factor. Examples include influenza and cholera. Often a diagnosis will include descriptive terms combined with a causal factor such as meningococcal meningitis or rheumatic heart disease.

The International Statistical Classification of Diseases and Related Health Problems (ICD) is the most widely used classification system. It is published by WHO in a book that is revised every 10 years or so by an internationally representative committee of experts that meets regularly and reviews advances in the understanding of diseases. It is now in its tenth edition (ICD-10), which came into effect in 1993 exactly 100 years after the original. Every disease entity is assigned an alphanumeric code number in a hierarchical arrangement; there are now 21 major disease divisions and each has a series of subdivisions. Some of the divisions are based on etiology, such as Chapter I (A00-B99): Certain Infectious and Parasitic Diseases; some on body organ systems, such as Chapter VI (G00-G99): Diseases of the Nervous System; and others on classes of conditions, such as Chapter XIX (S00-T98): Injury, Poisoning and Certain Other Consequences of External Causes. The classification is now increasingly used for death certification and for hospital inpatient discharge coding. When properly used, it is a very valuable tool for epidemiological studies.

### 3.2. Counting Disease

Counting the frequency of disease can be done in several ways; it is important to understand what these different methods of counting actually mean. The most useful way may depend upon the nature of the disease and upon the purpose for which it is being counted. The starting point for any counting must be a clear definition of the disease or event that is to be counted as discussed above. Further, it is important to be clear whether the number of cases refers to individuals or episodes or attendances. For a particular disease a person may have several separate attacks in one year and may attend a clinic two or three times for each attack. Only one person has been ill but has had several episodes and attended a health service several times for each episode.

The three basic measures of disease occurrence are the *incidence density*, often simply termed the *incidence rate*; the *cumulative incidence*; and the *prevalence*.

*Incidence* is the fundamental building block for epidemiological inference. Incidence is a measure of events (i.e. transition from a non-diseased to a diseased state) and can be considered as a measure of risk. This risk can be looked at in any population group,

defined by age, sex, place, time, socio-demographic characteristics, occupation, or indeed, for example, by exposure to a toxin or any suspected causal factor.

The incidence rate (incidence density) is defined as the number of new cases of disease onset per person-time. There are three critical components: a definition of the onset of the event, a defined population, and a particular period of time. The essential point is new cases of disease—the disease develops in a person who did not have the disease previously. The numerator is the number of new cases of disease (the event) and the denominator is the number of person-time units at risk for developing the disease. Everyone included in the denominator must have the potential to become part of the group that is counted in the numerator. To calculate incidence of prostate cancer, the denominator must include only men, because women are not at risk of developing prostate cancer. The third component is the period of time or time-unit. Any time-unit can be used so long as all those counted in the denominator are followed for the same period as those who are counted as new cases in the numerator. *Incidence density* directly incorporates time into the denominator and it is generally the most useful measure of disease frequency, often expressed as new events per person-year or per 1000 person-years. For a variety of reasons, including loss to follow-up, individuals in the denominator may not be followed for the full time period specified, so that different individuals are observed for different lengths of time. In such situations, an incidence rate uses a denominator consisting of the sum of the different times each individual was at risk expressed as person-years.

*Cumulative incidence* or *incidence proportion* is the number of new cases of a disease that occur in a population at risk for developing the disease during a specified period of time (during which all of the individuals are at risk). It is the proportion of people who develop new disease in a specific period of time.

Note that the expression *rate* in biology or physics generally refers to time in terms of how fast a process occurs and usually is expressed per instant of time. In epidemiology, *rate* is often loosely used to express a proportion and to provide a denominator of the population in which new cases are occurring during a period of time, as in cumulative incidence rates above. Use of rate as in incidence density is closer to its more general meaning of how fast new cases are arising and can indeed be expressed as per instant of time, often referred to as the *force* of mortality or morbidity.

Occasionally, time may be implicitly rather than explicitly specified. In acute epidemics of very limited duration such as most food poisoning outbreaks, most cases occur within a few hours or days after exposure. Cases that may develop months later are not considered part of the same outbreak. But in most situations in which current knowledge of the biology and natural history of the disease does not clearly define a time frame, time must be stated explicitly.

*Prevalence* is a measure of present status—the number of people who currently have the disease—rather than of newly occurring disease. It measures the proportion of people who have defined disease at a specific time. Thus it is a composite measure made up of two factors: the incidence of the disease that has occurred in the past that continues to the present or to some specified point in time, that is, prevalence equals incidence rate

of the disease times the average duration of the disease. For most chronic diseases prevalence rates are more commonly available than are incidence rates.

### 3.3. Severity of Disease

Measures of disease frequency are central for epidemiological investigations concerning etiology. To understand the importance of a disease in a population, however, it is necessary to consider not only the frequency of the disease but also its severity as indicated by the extent of disability and premature mortality that it causes. Premature mortality is defined as death before the expectation of life at the age of death, had the disease not occurred.

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He obtained his B.A. in economics at Swarthmore College, his M.D. at Washington University, St. Louis, his M.P.H. at Harvard School of Public Health, is board certified in Internal Medicine and a Fellow of the American College of Medicine and Member of the American College of Epidemiology. He has worked extensively in clinical and epidemiological research in Ghana, Uganda, and globally with the United Nations Development Programme, the World Health Organization, and the World Bank Special Program in Research and Training in Tropical Diseases (TDR) and served as a member of the faculty at Makerere University Medical School, University of Ghana Medical Faculty, and Harvard University School of Public Health before Johns Hopkins. He now has a special interest in the application of epidemiological methods to health policy and planning in developing countries.