Screening for Disease Part I

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Welcome to this four-part series focused on epidemiologic and biostatistical methods related to disease screening. In this first module, we will learn about three types of prevention, screening tests, and discuss essential components for effective screening programs.



After viewing this module, you will be able to identify three types of prevention, define screening, distinguish screening from diagnosis and will also be able to describe the criteria for an effective screening program.



In order to understand types of disease prevention, it is important to take into account the natural history of the disease in question.

Natural history of disease is the course of a disease from its beginning to its final clinical or end points.

Patients begin in a healthy state and then progress through a preclinical phase where the disease process begins but symptoms have not yet become apparent. The preclinical phase is followed by the clinical phase where the patient experiences symptoms, seeks care, receives a diagnosis and subsequent treatment, and is followed for an outcome.



Primary prevention is an action taken to prevent the development of disease in a person who is well and does not yet have the disease.

Secondary prevention involves identifying people in whom a disease has already begun but who have not yet developed symptoms of the illness.

Tertiary prevention is preventing complications in those who have already developed signs and symptoms and have been diagnosed –clinical phase of illness.

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Primary Prevention			
Healthy	Preclinical Phase	Clinical Phase	
	Disease Onset Sy	nptoms Diagnosis Treatment	Outcome
Primary	Secondary	Tertiary	
 Prevent occurrence of disease 			
Remove exposure or agent			

In public health, primary prevention of disease is considered the best approach. With primary prevention, we aim to prevent the occurrence of disease. This can be accomplished by removing the exposure or agent, altering risky behaviors, or protecting the individual from the exposure or agent. Primary prevention interventions are applied before disease occurs.

Primary prevention includes activities such as promoting a healthy diet, exercise, sunscreen use or immunizations.

Primary prevention may be active or passive.

Active prevention requires some sort of behavioral change on the part of the individual.

Examples including wearing protective devices to prevent occupational injuries and immunizations against infectious disease.

Passive prevention requires no voluntary effort. Examples include fluoridation of public water supplies or vitamin fortification of milk and bread products.



If all cases of disease cannot be prevented, the next best strategy is early detection.

Secondary prevention aims to reduce the progression and impact of disease. We aim to detect disease early and then utilize prompt treatment.

Examples of secondary prevention include cancer screening programs to detect cancer in early stages or retinopathy screening among patients with type 2 diabetes.



In tertiary prevention, the goal is to impact the progression of chronic, long-lasting disease to reduce impairments or disabilities.

Tertiary prevention includes prompt treatment, proper follow-up, rehabilitation and patient education. We aim to maximize quality of life and functional abilities among patients with

chronic diseases.

Examples of tertiary prevention include controlling insulin to prevent complications from diabetes, physical therapy for stroke victims, and fitness programs for heart attack patients.



Screening is a component of secondary prevention.

Screening involves rapid tests that identify disease in asymptomatic individuals.

Objective of screening test or program is to distinguish persons who probably have the disease from those who probably do not have the disease. Note that screening programs are not intended to be diagnostic; instead, positive or suspicious findings from a screening test are referred for diagnostic testing and based on the diagnostic testing results, are referred for subsequent treatment as appropriate.

An example of a screening test would be mammography, which if found to be positive, is followed by more definitive testing using imaging and biopsy, and if found to be positive, the patient receives treatment for breast cancer.



A screening test is meant to identify those who may have disease earlier than those who are not screened and have no idea they have a disease until symptoms arise. A key feature of screening tests is early detection.

In contrast, diagnostic tests determine the presence or absence of a disease when individuals show symptoms.

Screening tests are often cheaper, less invasive, and easier to use in practice than diagnostic tests.



To be effective, the disease screening process is implemented during the preclinical phase i.e., after the onset of disease but before the patient experiences symptoms.

Keep in mind that different diseases have different lengths of time during the preclinical phase or clinical phase, which plays a role in whether an individual should be screened for a specific disease.



Early detection involves detecting disease at an earlier stage than what would occur in standard clinical practice without a screening. The goal for screening is to detect disease earlier in order to improve prognosis or outcome and reduce morbidity and mortality related to that disease.

There is an assumption that early detection automatically leads to decreased morbidity and mortality, but this is not always the case. If there is no treatment or way of controlling a disease, then this would not be the case.



Now, we will review the World Health Organization's criteria for Effective Screening programs. These are criteria that should be considered when developing a screening program.



First, the condition being screened for must be serious. For example, most states require newborn screening for congenital anomalies including congenital hypothyroidism. If untreated, this rare condition leads to mental retardation, which is severe and costly to society.



Second, the condition being screened for must be treatable. Once the patient screens positive and is positive for subsequent diagnostic tests, there needs to be an effective treatment that can be used to treat the disease.

For example, all newborns are screened for phenylketonuria (PKU) which is a birth defect that causes an amino acid called phenylalanine to build up in the body. The condition is treatable by withholding foods that contain phenylalanine.

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The third criterion relates to the timing of the screening process. The condition must be detectable while asymptomatic and timely treatment must reduce morbidity and mortality more effectively than treatment after the appearance of symptoms.

For example, cervical cancer can be detected an average of 8 to 9 years before it becomes symptomatic. If caught early by a Pap test, cervical cancer can be treated successfully. If cervical cancer is diagnosed later, after symptoms have developed, treatment is less effective and the prognosis is poor.

WHO Criteria for Effective Screening

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4. The screening test must be accurate.

Example: Testing for fecal occult blood to detect colon cancer among asymptomatic, low-risk populations is an example of a screen with low accuracy. The sensitivity is only 50% and the positive predictive value is 5%. Thus, for every 20 people who initially screen positive and suffer the cost and anxiety of follow-up tests, only one will have colorectal cancer.

Fourth, the screening test must be accurate in distinguishing disease cases from noncases.

As an example, testing for fecal occult blood to detect colon cancer among asymptomatic, low-risk populations is an example of screening test with low accuracy. The sensitivity (which is the probability of having a positive test among those who have colon cancer) is only 50% and the positive predictive value is only 5% (which is the probability of having colon cancer among those who test positive based on the presence of fecal occult blood).

This means that for every 20 people who initially screen positive for fecal occult blood and suffer the cost and anxiety of follow-up tests, only one will have colorectal cancer.



Fifth, the screening test must be acceptable to the patient and inexpensive.

For example, sigmoidoscopy is more sensitive than testing stools for occult blood and is highly specific; however, its value as an effective screening test is questionable given the invasive nature of the test. Patients and physicians avoid using sigmoidoscopy and the cost is also high.



The final criterion is that the condition must be sufficiently prevalent to warrant screening.

For example, 50 million Americans have high blood pressure. Most patients would benefit from monitoring and treatment of their high blood pressure. Screening is warranted for hypertension because the prevalence is high, and the other conditions previously discussed would be met.



To conclude this module, let's now discuss universal versus targeted screening.

A universal screening program is one that is applied to all members of a given population. For example, newborn hearing tests are applied to all newborns. Pap tests are recommended for all women above a certain age. As a final example, blood pressure is checked for all patients visiting a primary care physician's office. These screening tests are applied universally to everyone in a defined population.

Targeted screening, on the other hand, is applied in populations who are at particularly high risk for developing the disease of interest.

For example, mammography screening is recommended for younger women only when the woman has a family history of breast cancer (high-risk subgroup). In addition, mammography screening is recommended more broadly when considering women who are older; however, among younger women, screening is focused on particular, high-risk subgroups.



In this module, we discussed three types of prevention.

We learned about the features of a screening program that distinguish screening from diagnostic testing programs. We have also discussed six criteria for defining an effective screening program.