

Short Communication

Screening for Diseases in Family Practice

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Abstract

The active check-over for disease among apparently healthy people is a fundamental aspect of prevention. This is perceivable by screening, which is a search of unrecognized disease or condition by means of rapidly applied test, examination or other procedures in apparently healthy individuals. This is carried out in the hope that earlier diagnosis and subsequent management favorably alters the natural history of the disease in a significant proportion of those who are identified as 'test-positive'.

Family Practitioners have privilege to provide comprehensive and holistic health care services including preventive, curative and rehabilitative on continuous and long-term basis to all members of family irrespective of their age, sex and nature of disease and condition. Screening of disease being an important preventive strategy should be offered by Family Practitioners to their clients when ever recommended and appropriate.

However, before screening is initiated, a decision must be made whether it is worthwhile, which requires scientific, financial and ethical justification. This review summarizes the basic concepts and criteria regarding the screening for diseases.

Introduction

Family Practice is a coordinated, integrated and comprehensive care provided to individuals and their families in the broad land between health, illness and diseases. Family Practitioners are engaged with individuals and their families across the field of cure, care and prevention by using and integrating the science of biomedicine, medical

psychology, epidemiology and preventive medicine. In this respect, it also includes early detection and prompt treatment of diseases, thus, preventing and minimizing complications and disabilities.

Prevention is the best strategy to reduce disease burden as well as to decrease substantial direct and indirect cost that affects the patient, his/her family, society and health system of the country as a whole. Prevention of disease should be at all levels when it is desirable. Screening for disease is a major component of the Family Practitioner's overall preventive responsibility. It allows the physician to make age and gender specific recommendations modified by the patient's individual risk profile as determined by family history, previous health history, health-related behaviors, and environmental and occupational exposure. Screening maneuver applied to apparently well people to identify those at increased risk of a disease. Thus the basic purpose of screening is to identify earlier the persons who are diseased among those who are free from that particular disease. An implicit assumption underlying the concept of screening is that early detection of disease, before the development of symptoms, will lead to a more favorable prognosis as treatment begun at an earlier stage of disease will be more effective.

Screening of disease may be done through specific tests, history and/or clinical examination or implementing specific questionnaires for that particular condition. A screening test is not intended to be a diagnostic, it is an initial examination only, and positive responders require a second, diagnostic examination¹, that may be more expensive and painful. For example, if a woman found a lump in breast during her routine manual breast examination,

should be evaluated further for definitive diagnosis by mammography and biopsy. However, the criteria are not hard and fast and there are some tests that are used both for screening and diagnosis. Test of hemoglobin percentages for anemia is a good example in this regard.

Screening for disease at their early asymptomatic stage has played a significant role in public health practice and now considered as a routine and important aspect of a good medical care. However, inappropriate application or interpretation of screening test can rob people of their perceived health, initiate harmful diagnostic testing, and squander health-care resources.² Every test is not an ideal test for screening purpose, as every disease is not an ideal for screening. There are certain criteria which makes a test more suitable and appropriate for using as a screening test and similarly so for disease or disorder to be screened and the population to whom screening test is to be applied.

Test to be appropriate for screening

An appropriate screening test should be capable of identifying the people among the population who have a high probability of being affected by disease or defect and capable of excluding those who are not affected. In other words, screening test should identify persons correctly who are more likely to have a disease among the apparently normal population. A screening test ideally should be valid, reliable and reproducible. In addition, it should be inexpensive, easy to administer, and impose minimal discomfort on the person or population to whom it is applied.

The validity of a screening test is measured by its ability to do what it is supposed to do, that is, correctly categorize persons who have pre-clinical disease as test-positive and those without pre-clinical disease as test-negative. Sensitivity and specificity are two measure of the validity of a screening test. Sensitivity of the test is defined as the ability of the test to identify correctly those who have disease while specificity of the test is defined as the ability of the test to identify correctly those who do not have disease.³ In other words, sensitivity of the test is the proportion of true positives amongst all those who truly have disease and specificity is the proportion of true negative amongst all those who truly do not have disease. Hence, it is desirable to have a screening test that is both highly sensitive and highly specific. However, usually it is not possible, and there is generally a tradeoff between the sensitivity and specificity of a given screening test⁴, i.e. if sensitivity of test increases the specificity decreases and vice versa. In addition to sensitivity and specificity, the performance of a screening test is measured by its predictive values, which reflects the detection power of the test. The term 'false-positive' means that persons who do not have disease are told that they have the disease and 'false-negative' denotes that

persons who actually have disease are told that they do not have. The more prevalent a disease is in a given population, the more accurate will be the predictive value of a positive screening test. The predictive value of a positive result falls as disease prevalence declines. Reliability (repeatability) of a test refers to the consistency of results when repeat examinations are performed on the same person under the same conditions. Regardless of the sensitivity, specificity and predictive accuracy of a test, if the test results cannot be reproduced, the value and usefulness of the test is minimal.³

Disease to be appropriate for screening

There are some characteristics that make disease appropriate for screening. To be suitable for screening, the prevalence of disease should be high among the population screened, having a long lead time, disease should be serious enough and treatment given at an early stage should be more beneficial in terms of reducing morbidity, disability and mortality. The criterion of seriousness relates primarily to issues of cost-effectiveness and ethics. The expenditure of resources on screening must be justifiable in terms of eliminating or ameliorating adverse health consequences. Similarly, with respect to ethics, the consequences of failing to diagnose and treat early must be sufficiently grave to warrant undergoing the risk and discomfort of the screening procedure itself. For a screening test to be more fruitful, treatment given during detectable pre-clinical phase must result in a better prognosis than therapy given after symptoms develop. Type 2 Diabetes Mellitus (DM), for example, fulfills all the criteria to be a disease suitable for screening. First, type 2 DM is a pandemic disease with higher prevalence worldwide^{5,6} thus its prevalence in a screened population is likely to be high. Second, diabetes remains asymptomatic for a long period of time in majority of subjects and presentation with complications is not unusual.^{7,8} Third, it is a serious disease, which leads to greater number of short-term and long-term complications, disabilities and premature mortality.^{9,10} Fourth, early detection and prompt treatment of type 2 DM has shown to reduce and delays the long term complications.^{11,12} Hypertension and carcinoma of cervix are other diseases that are appropriate for screening in their early and asymptomatic stages. On the other hand, for example, lung cancer has a poor prognosis, regardless of when treatment is initiated thus the application of a screening test will be neither necessary nor effective for this disease. Other disorders/conditions which can give high yield for screening in Family Practice set-up includes dyslipidemia in high risk persons, perinatal screening of congenital and genetic disorders in high risk women during antenatal check-ups,

includes dyslipidemia in high risk persons, perinatal screening of congenital and genetic disorders in high risk women during antenatal check-ups, congenital hypothyroidism during infancy and measurement of intra-ocular pressure of elderly persons.

Population should be appropriate for screening

To be cost-effective and to get maximum yield, screening test should be applied for specific group of population having more probability and suspicion of disease to be present. In other words, the number of cases detected by screening can be increased by screening high-risk groups such as targeting breast cancer screening to women with family history of disease; cervical cancer screening to women with multiple sex partners and belonging to low socio-economic status. Diabetes is another good example in this regard. People who are at higher risk of developing type 2 DM should be screened when appropriate and required.

References

1. Last JM. A dictionary of epidemiology. 3rd ed. New York: Oxford University Press, 1995.
 2. Grimes DA, Schulz KF. Use and abuse of screening tests. *Lancet* 2002;359: 881-4.
 3. Gordis L. Assessing the validity and reliability of diagnostic and screening tests, 'Epidemiology' Philadelphia: W.B Saunders, 1996, pp 58-76.
 4. Hennekens CH, Buring JE. Screening. *Epidemiology In Medicine* (first ed.) Boston: Little Brown and Company, 1987, pp 327-47.
 5. King H, Aubert RE, Herman WH. Global burden of diabetes, 1995-2025: prevalence, numerical estimates, and projections. *Diabetes Care* 1998;21: 1414-31.
 6. King H. WHO and International Diabetes Federation: regional partners (Editorial). *Bull World Health Organ* 1999;77:954.
 7. Harris MI, Klein R, Welborn TA, Knudman MW. Onset of NIDDM occurs at least 4-7 years before clinical diagnosis. *Diabetes Care* 1992;7:815-19.
 8. Weerasuriya N, Siribaddana S, Dissanayake A, Subasinghe Z, Wariyapola D, Fernando DJS. Long-term complications in newly diagnosed Sri Lankan patients with type 2 diabetes mellitus. *Q J Med* 1998;91:439-43.
 9. World Health Organization. Technical Report Series No. 646. World Health Organization, Geneva, 1980.
 10. Spanheimer RG. Reducing cardiovascular risk factors in diabetes: which factors to modify first? *Postgrad Med* 2001;109:26-36.
 11. Harris MI. Undiagnosed NIDDM: clinical and public health issue. *Diabetes Care* 1993;16:642-52.
 12. Davies MJ. Is screening for type 2 diabetes justified? *The Practitioner* 1999; 243:93-100.
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