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Editorials

What are we screening for?

There is a temptation to view the objective of screening in terms of the identification of individuals with positive results, or those with a recognisable predisposition to a disease. As a result, there is a tendency to lose sight of the fact that a screening test needs to be judged by how well it identifies individuals who actually have the disease or who, in the absence of preventive action, would develop it. The confusion may lead to the impression that certain screening tests are more effective than they are. Perhaps worse, it can create the impression that having a positive screening result is itself a disease.

Screening for stroke by regular blood pressure measurement is often incorrectly defined as screening for hypertension. This reduces screening to a meaningless tautology, in which the objective of measuring blood pressure is to identify those with high blood pressure. Viewed in this way, the screening test is necessarily perfect, even though only about 30% of future strokes will be identified in the 5% of individuals with the highest blood pressure. The possibility for confusion is made worse by calling high blood pressure "hypertension", which gives the impression that it is a separate medical entity. Hypertension is the positive test result, not the disorder for which we screen. Screening for stroke using blood pressure measurement necessarily identifies other diseases linked to high blood pressure. This does not affect the argument – it simply implies that the performance of measuring blood pressure for the detection of each disease needs to be specified separately in the same way as for stroke.

Mutations in the BRCA1 gene on the long arm of chromosome 17 indicate a high risk of breast cancer. Identification of individuals who have a mutation in this gene is sometimes defined as screening for disease predisposition. In fact, this is an example of genetic screening for breast cancer. The problem implicit in making predisposition to disease the objective of screening is the tendency to assess the performance of the test by its ability to identify individuals with the gene, rather than future mortality or morbidity in the absence of preventive action. Technical errors notwithstanding, the test will be perfect – with a 100% detection rate and a false positive rate of 0%. The value of the test should be assessed in terms of its ability to identify women who will develop breast cancer or who, in the absence of intervention, would die from the disease. The BRCA1 test would then have a much lower detection rate and a significant false positive rate, since not everyone with breast cancer will have the gene and a proportion of women who do not develop breast cancer are likely to have the gene. The test may be a poor means of discriminating individuals who will and will not develop breast cancer.

An advantage of keeping the focus on the disease, rather than the screening test, is that it encourages the development of complementary screening tests for the same disorder, which may improve the performance of screening for that disorder.

It is often convenient to describe screening programmes in terms of the test – for example, α fetoprotein screening (for open neural tube defects), mammographic screening (for breast cancer), or blood pressure screening (for stroke). There is no disadvantage in this shorthand as long as it is used simply to specify the screening method and does not lose sight of the principal objective.

It is essential to distinguish between positive screening results and the disorder that the screening test is designed to detect if people are to have a realistic appreciation of the expectations of screening.

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