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Editorials

Why the term "carrier screening" should be abandoned

Words used for describing medical screening activities can have a pivotal role in guiding perception as to how efficacy should be judged. An initial step in providing such guidance is accomplished when the screening test is characterised according to the medical disorder being sought. Although this step may seem self evident, there are instances in current medical practice where the purpose of a screening test is not accurately stated. Blood pressure measurement, for example, is most often described as a screening test for hypertension. Hypertension is a synonym for high blood pressure and is not itself a medical disorder. If judged only on its ability to detect hypertension, blood pressure measurement is highly effective. In fact, the purpose of such screening is to identify individuals at high risk for stroke. Efficacy, therefore, needs to be measured not by how many people with high blood pressure can be found but, rather, by how successfully individuals at high risk for stroke can be identified and their strokes avoided by treatment.

A similar confusion in screening terminology is beginning to appear, now that tests for genetic disorders are being developed. A number of recessively inherited genetic disorders can be identified or anticipated by screening tests performed either before or during pregnancy. "Carrier screening" or "screening for carriers" is now widely used in both medical publications and common parlance. Carriers of a recessive trait do not ordinarily suffer adverse health consequences; there is, therefore, no value on that basis for individuals to be informed of their carrier state. Identifying carriers of the trait in question represents one step in the screening process; it is not an end in itself. The goal of such screening is to identify couples at high risk for producing offspring with a serious medical disorder (which occurs when two copies of the recessive gene are inherited). When couples are aware of their risk, it is possible for them to choose any of several options to avoid the birth of an affected child.

"Carrier screening" or "screening for carriers" is currently used most often in relation to cystic fibrosis, but it can also be found in articles dealing with sickle cell anaemia, thalassaemia, and Tay–Sachs disease. A more accurate description would be—for example, "cystic fibrosis screening", with carrier identification classified as the screening test. Characterising the screening process in this way is an important step in directing attention towards the correct yardstick of screening efficacy: the extent to which morbidity and mortality of the medical disorder can be reduced.

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