

# TESTING TREATMENTS

## Chapter 4, 4.3 EARLIER IS NOT NECESSARILY BETTER

screening with screening by a special sort of computed tomography (CT) scan called a spiral CT. Both groups were assigned to three annual screening procedures. Spiral CT diagnosed lung cancers at an even earlier stage than did chest X-rays, and in a small proportion of patients this was sufficiently early (stage A in Figure) for treatment to be of benefit (346 deaths from lung cancer in the spiral CT group vs 425 in the chest X-ray group). But this beneficial outcome came at the expense of a large proportion of people wrongly labelled with lung cancer. Overall, for every 1,000 heavy smokers who had three annual X-rays or scans, over eight years of follow-up, three fewer died of lung cancer. But 13 still died of lung cancer despite earlier detection, and 233 received a false-positive result that required further investigation.<sup>19</sup>

### **Genetic tests: sometimes useful, often dodgy**

Not so long ago ‘genetic testing’ was more or less confined to generally rare, single-gene disorders – for example, the childhood-onset muscle-wasting disease Duchenne muscular dystrophy, or Huntington’s disease, a progressive nervous system disorder that usually starts to affect people in middle age. Genetic tests are done to diagnose such conditions but can also be used to screen healthy people whose family history indicates that their chances of developing the disorder in question are above average, and to guide their family plans.

However, most diseases cannot be attributed to a single faulty gene. Usually, diseases depend on the way in which risk variants in several genes interact, and on the interaction of these genetic risk variants with environmental factors. Only when there is a ‘critical’ combination of genetic risk variants and environmental factors will a disease become apparent.<sup>1</sup>

Despite the complexity of ascribing most conditions to aberrant genes, media and promoters of direct-to-consumer genetic testing extol the supposed virtue and simplicity of genetic risk profiling. All you need to do is send off a saliva sample to a company for DNA analysis and they will take your money and send you your profile. But the information you receive is unlikely to help you – or your clinician – make any sensible predictions

## **DON'T PLAY POKER WITH YOUR GENES**

'Acting on the knowledge of a single (or even a few) gene variants is similar to betting all your money on a poker hand when you've only seen one card. You don't know what hand genetic factors has dealt you, nor what effects your environment will have, and here, instead of 5 cards, there are over 20,000 genes and many thousands of environmental factors. And the effect of one gene may be cancelled out by the effect of lifestyle, family history or by the presence of other, protective genes. Many of us carry faulty genes without them ever causing disease.'

*Sense About Science. Making sense of testing: a guide to why scans and other health tests for well people aren't always a good idea. London: Sense About Science 2008, p7. Available from [www.senseaboutscience.org](http://www.senseaboutscience.org)*

about your risk of disease, let alone what might be done about it, if anything. This 'do-it-yourself' approach clearly does not meet the criteria for a useful screening test (see below). However, the result may well make you anxious and decision-making difficult, and may have wider implications too – on members of your family, for example. As one Australian health journalist put it 'For anyone concerned about the creeping medicalisation of life, the marketplace for genetic testing is surely one of the latest frontiers, where apparently harmless technology can help mutate healthy people into fearful patients, their personhood redefined by multiple genetic predispositions for disease and early death.'<sup>20</sup>

**What screening aims to achieve and why evidence matters**  
The examples we have already given show that, before rushing headlong into widespread screening, it is worth pausing a moment to consider the key features of screening programmes and to remind ourselves what they aim to achieve. People being offered screening do not have, or have not noticed, the symptoms or signs of the condition being tested for – they have not sought medical attention for the disorder in question. The purpose of screening