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MEDICAL SELF-TEST KITS

The Human Genetics Commission will shortly publish advice to Government on issues related to the supply of genetic tests direct-to-the-public. However, it is not only genetic tests that are increasingly available over the counter and via the internet, but also a variety of other medical tests. This briefing describes the range of tests available and discusses the implications of self-testing for individuals and for the NHS.

Background

A range of medical tests is available direct-to-the-public outside the conventional medical referral system. This growing market is driven by the development of new technologies that have made testing cheaper and easier to carry out and by public interest in personal health. Companies see opportunities to market their test kits through the internet and by mail order, where some 1000 tests are available, and in pharmacies and shops, where a more restricted range is marketed. They point to pregnancy testing as an example of how self-testing can become a normal part of the health service. It is now common for doctors not to repeat a home pregnancy test.

The simplest tests can be completed at home, typically using a sample of urine, blood or faeces. Usually, the test will take a matter of minutes, with the results shown by a colour change in a test material. For tests that require more complex technology or expertise for completion, or where more precise results are needed, the sample is sent by post to a laboratory for testing, with the results subsequently returned to the customer.

The tests can be divided into four overlapping categories:

- **screening tests** aim to identify if someone is likely to have a disease (see box opposite). To give a firm diagnosis, further investigation by a doctor is needed
- **diagnostic tests** aim to give a definite answer as to whether an individual currently has a particular disease or condition (see box on page 2)
- **monitoring tests** track the progress of a known existing condition or the response to therapy. For example, the prostate cancer, osteoporosis and diabetes tests described above are all recognised monitoring tools
- **predictive tests** use genetic and other information to assess whether a healthy individual is at an increased risk of developing disease (see box on page 2).

Screening tests – some examples

Prostate cancer
Prostate cancer is the most common cancer to affect men in the UK. This test looks at the concentration of prostate specific antigen (PSA) in blood, which can be linked with prostate cancer. One home-based test (~£15) detects whether the PSA concentration is above a set threshold. If the test is positive, customers are advised to visit a doctor. A laboratory-based test (~£95) is also available and measures the relative concentrations of the two different forms of PSA, which are thought to give a more useful indication of the presence of cancer. The use of the PSA test to screen people who show no symptoms of disease is controversial because the PSA test often gives a positive result to healthy individuals; it does not differentiate between different types of prostate cancer – some cancers develop slowly and there is no consensus over whether treatment in these cases is appropriate; and the only treatments currently available are highly invasive and can cause incontinence and impotence.

Osteoporosis
An estimated 3 million people in the UK have osteoporosis, which causes bones to fracture easily. This laboratory-based test (~£115) measures the concentration of a biochemical marker (deoxypyridinoline, dpd) in urine. This indicates how quickly bone is being broken down as part of the natural cycle of renewal. Those with an above average result are advised to visit a doctor. The National Osteoporosis Society does not consider that this test should be marketed as a screening tool, but does recognise that it can have a role in monitoring disease.

Other examples include bowel cancer (tests for blood in faeces) and diabetes (tests glucose levels in urine or blood).

Diagnostic tests – some examples

Sore throat
Bacterial infection accounts for some 10% of sore throats and antibiotic treatment is an option in these cases. This home-based test (~£15) detects Streptococci bacteria in a sample taken using a throat swab. If the result is positive, customers are advised to consult a doctor. If it is negative, the advice is to soothe symptoms with products available over the counter. Similar tests are marketed to doctors but there is debate over whether antibiotics are an appropriate routine treatment for bacterial sore throats as evidence suggests that treatment shortens illness by only one day.

Food intolerance
Some chronic conditions such as irritable bowel syndrome have been linked to a sensitivity to specific foods. An initial laboratory-based blood test (~£20) identifies if consumers have a food intolerance; a more in-depth analysis (£125 to £245 depending on the number of foods that are tested) can then identity which foods could be cut out of the diet to alleviate symptoms. The package includes telephone advice from a nutritionist. The test is based on measuring levels of certain antibodies, an approach not yet proven in clinical trials. However, on the basis of a survey, which found that the test benefited a majority of customers, the patient group Allergy UK has endorsed the test provided by one company.

Other examples include tests for Chlamydia, cystitis and for some genetic diseases such as cystic fibrosis.

Regulating the quality of test kits

The Medicines and Healthcare products Regulatory Agency (MHRA), an agency of the Department of Health (DH), is responsible for regulating the safety, quality and performance of self-test kits.[1] The ultimate sanction has, until now, been prosecution under the Consumer Protection Act or the Trade Descriptions Act. However, from December 2003 (2005 for products already in the supply chain), all kits sold in the UK will have to comply with new Medical Devices Regulations (see box on page 3).[2] The regulations deal with kits used to test human samples and are intended to ensure that all test kits are safe to use and that they perform as intended by the manufacturer. To show that their tests conform, manufacturers will be required to apply a CE marking (the EU's 'quality kitemark') to their products.[3]

Issues

Customers might reasonably expect that using a self-test will bring them some benefit, and certainly no harm. For this to be the case, a test needs to perform as described, the result needs to have a valid link to a disease and the customer needs to know how to respond to the result.

Test performance

CE marking should offer a presumption that a test will perform as claimed by the manufacturer. This relies on:

- the effective enforcement of regulations by the MHRA and its EU counterparts. The Consumers' Association has previously said that poor enforcement is allowing CE marking to be used as a marketing gimmick
- an awareness by users that only CE marked tests fulfil the requirements of the Medical Devices Regulations
- the correct use of a kit by the user.

Predictive tests – some examples

General health
Health is influenced by environmental (lifestyle, diet) and genetic factors. This test looks at these factors to give advice about how to improve long term health. The customer sends a sample of cheek cells, collected at home using a swab, to a laboratory and completes a lifestyle questionnaire. DNA is extracted from the cells and common mutations (each found, typically, in at least 10% of the population) in nine different genes are identified. As a result, customers may, for example, be advised to alter their diet, to take vitamin supplements or to stop smoking. Information about specific medical conditions is not given although there are plans to offer a parallel service that would give a more clinical interpretation of the results for use by doctors. This test was briefly available direct-to-the-public in 2001 for £120. Poor sales, and widespread negative publicity about the validity of the test and the value of the advice given to customers, led to its withdrawal from the direct-to-the-public market. The test is now available only through health practitioners registered with the company.

Deep vein thrombosis
Long haul travel and some contraceptive pills can increase the risk of thrombosis (blood clotting). This laboratory-based blood test (~£155) aims to identify individuals at particular risk. It analyses a number of chemical and cellular factors linked with blood clotting, including looking for mutations in two of the genes that code for blood clotting factors. If customers are identified as being at higher risk of thrombosis, advice is given on how to reduce risks during long haul flights, by for example wearing support stockings. They are also advised to contact their doctor.

Not all predictive tests involve genetic analysis. For example, cholesterol level and blood pressure tests can be used to predict an increased risk of heart disease.

Usefulness of the test result

A test result is useful only if there is firstly a valid link between the result and the condition that it is claiming to test for, and secondly if it leads to some benefit for the customer. These issues are not considered as part of the CE marking process, nor are they covered by any voluntary self-regulation within the industry, although the British In Vitro Diagnostics Association are considering this option.

A possible template for assessing the usefulness of any test is the criteria used by the UK National Screening Committee (NSC, see box on page 3), which provides advice to the four UK countries. These criteria are used to assess whether routine testing by the NHS of sections of the population is likely to bring overall benefit.

While the basic principles can be applied to self-testing, individual circumstances play a greater role in a decision to take a self-test. For example, on the basis of the NSC criteria, the use of the PSA test for prostate cancer screening (see box on page 1) has been rejected. However, where individuals are already showing some symptoms of prostate cancer, the PSA test is used in the NHS for both diagnosis and monitoring. With access to this type of information, individuals can make an informed choice about whether testing is useful for them.

Medical Devices Regulations

The regulations divide tests into categories according to the degree of perceived risk. This is based on who the user is and on the impact of the test failing to perform as intended.

- **self-testing kits** where a test is carried out at home form a discrete category. The manufacturer submits details of their test to an independent body designated and monitored by MHRA, which should ensure that the test performs as claimed when used by a non-professional
- **specific tests** that carry higher risks are listed separately and manufacturers must undergo a more stringent audit. The criteria for inclusion consider whether the result of the test might be used directly to inform medical action (as for diabetics who self-test their blood glucose levels) and how serious the impact of a false negative or false positive result would be. **Annex II List A** (highest risk) includes tests for HIV, Hepatitis and to determine blood groups. **Annex II List B** includes tests for prostate specific antigen (PSA) and blood glucose. These lists are kept under review at the EU level
- **all other tests** are included in the lowest risk category. Manufacturers self-declare conformity with regulations.

Possible criteria for assessing self-tests

Recognised criteria for assessing self-test kits would enable transparent decisions to be made about whether self-test kits should be placed on the market. The UK National Screening Committee (NSC) uses set criteria to advise whether population screening programmes should be introduced.[4] Amongst the criteria most relevant to self-tests are:

- the test should be simple, safe, precise and validated
- the test should be acceptable to the population
- the benefit from the test should outweigh physical and psychological harm (caused by the test, diagnostic procedures and treatment)
- there should be agreed policy on follow-up for individuals with a positive test result
- information explaining the consequences of testing, investigation and treatment should be made available to potential participants.

Counselling and support

The decision to take a test and then to determine what action to take in the light of the results is often not straightforward. Issues to consider prior to taking any test include the meaning of the result, the treatments available and the implications for family members. After a test, people may need help with interpreting the test result and advice on the options available.

Industry and consumer groups suggest that tests can be placed on a spectrum of 'seriousness' that determines the level of support to accompany them. This could be written information, either printed or on a website, or counselling, either face-to-face or by telephone. Key considerations in deciding the 'seriousness' of a test might include whether the disease is potentially life threatening, whether the test is predictive and whether it has implications for other family members. At one end of the spectrum lies pregnancy testing, a well established self-test. At the other, highly predictive genetic tests such as those for Huntington's chorea and some breast cancers are widely seen as appropriate only where offered in tandem with professional counselling – although they are currently available direct-to-the-public. This creates two options:

- either such tests would not be sold direct-to-the-public but be available only via health professionals – as is currently the case for HIV testing (see box on page 4).
- or there would be a requirement or expectation that industry would provide both pre- and post- test counselling as part of a testing service.

The middle ground is more difficult to define and there is no consensus over the level of support appropriate, for example, for screening tests for cancers or tests that determine whether an individual is carrying, but not affected by, a genetic disease. There are even concerns about pregnancy testing, which is now so sensitive that pregnancy can be detected at two weeks. Consequently women become aware of early miscarriages that might otherwise have passed unnoticed.

Implications for the NHS

There are differing opinions on how self-testing might affect the NHS. Industry representatives claim that self-testing might provide reassurance to the 'worried well', who would otherwise have visited their GP. They also believe that self-testing will encourage people who would otherwise not go to the doctor to screen themselves for disease. They point out that there are, for example, an estimated 1 million undiagnosed diabetics and that early intervention could reduce long-term health problems, representing a cost saving for the NHS.

On the other hand, the advice given to users of self-tests is to visit a doctor if they are concerned about the result, raising the prospect of increased demand for GP consultations. Industry representatives point out that this should not be regarded as a problem if the tests are providing useful information – raising the question of how this can be ensured. Most GPs will require training if they are to discuss the results of genetic tests with patients – DH is expected to publish a green paper on genetics shortly that will discuss the provision of genetic testing services in the NHS. One way to reduce the demand on GPs would be if other professionals, such as pharmacists, were to take on the role of providing initial interpretation of results. This raises questions about who should pay for their training and for time spent on advising customers. This could be a commercial decision for individual pharmacies or manufacturers. Alternatively the NHS could meet or contribute to costs as a means of decreasing the burden on its staff.

Issues specific to genetic tests

Genetics and health

The relationship between genetics and health is complex. Some diseases, such as cystic fibrosis, are caused exclusively by mutations in a single gene. In these cases, a genetic test can give a clear diagnostic result. Such tests are available direct-to-the-public but the market is likely to be small because the diseases are relatively rare. Most diseases, however, cannot be linked to mutations in one particular gene. Rather, the action of numerous different genes, combined with environmental factors such as diet, together determine whether an individual is likely to get a disease. The relative contribution of

genetic and environmental factors to any disease varies between diseases and between individuals.

As genetics is usually only one of the contributing factors to disease, some argue that genetic tests should be considered as no different from any other methods used to assess an individual's health. Others argue that genetic tests are special for reasons including:

- a genetic test may not tell only the individual about their health status, but could also have implications for blood relatives
- genetic tests can sometimes be highly predictive, raising issues about how individuals might respond to the results before a condition manifests itself
- people may believe that genetic information gives clear cut answers and may not recognise that genes are only one of many contributors to most conditions. This means that a higher level of support is needed.

As discussed below, this has led the Human Genetics Commission (HGC) to consider issues specifically as they relate to the supply of genetic tests direct-to-the-public.

Predictive tests

Industry representatives claim that there is now sufficient understanding of the genetic contribution to complex conditions such as heart disease and osteoporosis to allow them to draw valid conclusions about disease susceptibility from gene analysis. However, several consumer and public interest groups disagree and point out that scientific studies often produce conflicting results. Until large scale, long term research studies are carried out this is likely to continue to be the case.[5] The Consumers' Association argues that, at the moment, genetic tests predicting future health are of little value and are effectively an expensive way of getting commonsense advice – for example to give up smoking. Industry representatives argue that people want personalised advice and that this is a more effective motivator for change.

Regulatory issues

The HGC is to publish advice to Government on the supply of genetic testing services direct-to-the-public.[6] The issues it considers are not unique to genetic tests but there has been no equivalent consideration given to the supply of other medical tests. The HGC looked at how the validity and utility of any test could be assured, how advertising could be regulated and how internet sales could be overseen. It explored whether voluntary or statutory regulation would be most appropriate and is likely to recommend that the MHRA and other regulatory bodies establish a system for regulating genetic tests that is comparable with that for medicinal products. Some tests would be available only through a qualified medical practitioner, others could be administered by pharmacists or other health professionals such as dieticians, while others would be available direct-to-the-public. To implement such a system, the MHRA would need to work together with professional bodies, the NHS and industry to establish criteria for placing tests into these different categories.

HIV testing

The advertising and sale of HIV testing kits directly to the UK public was banned in 1992.[7] This was primarily because of concerns over the performance of tests emerging on the market and the need for pre- and post- test counselling to be provided.

A similar ban had been imposed in the US in 1988. This was lifted in 1995 when the Food and Drug Administration (FDA) decided that access to self-testing might encourage previously unreachable groups to test for HIV – although the overall impact has been low. There is currently one self-test HIV kit available in the US (~$60) that has been approved by the FDA. It is a laboratory-based test that offers customers counselling over the phone both before taking the test and when they are given their results. Several US companies market and sell unapproved HIV test kits and the FDA is particularly concerned about the performance of those that are carried out at home.

Industry representatives claim that there is demand for HIV self-testing from the UK public and there are tests currently on the market. The Terence Higgins Trust believes that there is a place for self-testing, particularly for people who test themselves regularly, but regard access to face-to-face counselling and follow-up to be important for others. The Department of Health wants to encourage uptake of HIV testing by a wider range of people and sees the best way to achieve this as offering testing in a wider range of settings rather than through self-testing.

Overview

There is an active debate about the supply of genetic tests direct-to-the-public. Less attention has been given to other medical tests, although many of the issues are the same. Measures are already being put in place to ensure that all tests perform as intended by the manufacturer. However, there is no system for considering wider issues such as whether a test is likely to give useful information to the user or what support an individual might need to interpret any test results. Such issues are critical in considering whether tests are appropriate for marketing direct-to-the-public.

Endnotes

1 The MHRA will be created in April 2003 and takes over the functions of the Medical Devices Agency and the Medicines Control Agency.
2 The Medical Devices Regulations (SI 2002 No 618) bring the In Vitro Diagnostic Devices Directive (98/79/EC) into UK law.
3 CE marking is required for a range of products from toys to electrical goods and is a declaration by a manufacturer that their product complies with appropriate EU directives.
4 Available via www.nsc.nhs.uk
5 See POSTnote 180, The UK Biobank, July 2002.
6 Genes Direct: ensuring the effective oversight of genetic testing services supplied directly to the public. *Human Genetics Commission*. Expected publication date: 9 April 2003.
7 HIV Testing Kits and Services Regulations 1992 (SI 1992 No 460).

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