EGAN
European Genetic Alliances' Network

Social and Psychological Aspects of Medical Testing

Foreword

For patients and families an accurate diagnosis, delivered in a timely manner with user-friendly explications, can be the key to understanding their condition. It can enable them to make effective plans for the management of their condition. This is important not only for the person affected, but also for those around them who may also find themselves at risk – be it because the condition may be genetic or infectious, or because it may impose demands related to the care and support that the affected individuals may need as their disease progresses.

Recent advances in diagnostics increasingly permit a precise and nuanced opinion as to a patient's condition and their likely prognosis. This greater scientific and clinical clarity allows for a more personalised approach to healthcare. At the same time, it implies a growing need for social and psychological support that patients and families require if they want to fully understand their condition and effectively address the consequences that may arise in non-medical areas.

This leaflet aims to provide ideas and guidance for patients and families who are offered diagnostic or predictive medical testing of one sort or another. It builds on the shared experience of patients and professionals who seek to explain the many and varied social and psychological consequences that may be generated by diagnostic testing programmes. In this way we hope that it will be possible for patients and their families to engage with their doctors as equal partners, making better, more informed choices when facing the suggestion of diagnostic or predictive testing that result from awareness of medical risk.

The booklet is the result of a collaboration between the patient group EGAN (European Genetic Alliance Network) and Roche. EGAN has collected commonly asked questions from patients and Roche has provided the answers with the verification of experts and independent specialists. In our effort to keep everything clear and accurate, we will gladly welcome any comment or feedback with suggestions for additional topics or other improvements that may be incorporated in future editions.

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1. What kinds of test are used to make a diagnosis possible?

Whenever medical testing is mentioned, many tend to refer to testing in the very limited sense of molecular or genetic testing, meaning the analysis of a person's DNA, RNA, proteins or protein products. Diagnosis is not limited to molecular analysis, however; there are many options, some of which are so well-established we barely consider them diagnostic tests anymore.

Information about a person's medical background can be established in a number of ways:

- **Appearance:** some diseases can change the way a person looks e.g. one symptom of acute hepatitis can be the slight yellowing of the skin or eyes
- **Consultation:** a patient explaining their symptoms and personal background e.g. earlier diseases, recent travels, allergies, etc., may lead to the correct diagnosis
- **Family background:** the medical history of a person's relatives may reveal the pattern of a genetic disease
- **Clinical examination:** a disease might be discovered using X-ray, ultrasound or similar means
- **Analysis of body tissues or fluids:** biological and chemical analysis may reveal anomalies which suggest a particular disease (e.g. bacteria in the blood or urine can indicate an infection)
- **Chromosomes:** chromosomal analysis (e.g. studying a person's set of chromosomes to identify differences) can confirm or exclude a suspected chromosomal defect
- **Molecular genetic analysis ("genetic test"):** a suspected gene mutation can be confirmed or excluded with an analysis of a person's DNA, RNA or proteins

The first three methods of diagnosis have been utilised for a long time whilst clinical examinations as well as analysis of body fluids or tissues are comparatively new. While raising a few controversial issues, most discussions focus on the even more recent introduction of chromosomal and genetic analysis.

2. What can tests show?

Tests may reveal a number of things. First, there are tests related to diseases, disease relapse, and monitoring of treatment reactions. Tests may:

- reveal unusual results that might suggest a particular medical condition (e.g. a blood parameter departing from the norm)
- give, confirm, refine or refute a diagnosis
- help adapt a treatment to the special needs of a patient (e.g. adjust the dose of a medication for a patient dependent on their metabolic rate or help determine when a treatment can be stopped)
- reveal a person's risk of being affected by a disease
- tell potential parents whether they are at risk of passing a disease on to their children
- show whether a child is likely to be born with a certain condition

Other tests are intended to answer questions that are not related to diseases. They may:

- reveal whether a person has taken illegal substances (doping, drugs)
- confirm or refute a pregnancy
- reveal the identity of a child's father
- reveal characteristics of a child before birth (e.g. gender)

3. Why would someone want to take a medical test?

In the case of someone taking a disease-related test, reasons may be:

- the person is feeling ill and would like to learn more about the cause
- a certain disease regularly occurs within a person's family. They might want to know whether they also carry the disease , or are at risk of passing it on to their children
- the person would like to know if a treatment is working
- a routine screening test to establish an early diagnosis, e.g. regular mammography
- to help an individual / family prepare and plan medically, financially and personally dependent on a particular diagnosis.

There are also reasons why someone may not want to take a test, ranging for instance from having no desire to learn of the result (see also Q 6 & 7), not trusting the test to deliver accurate results (see Q 5) or not wanting to pay for a test, if the costs are not reimbursed (see Q 6). Alternatives to testing should be discussed between patient and physician to ensure the patient has the full picture before making up their mind.

4. Do medical tests hurt?

As there are various tests and therefore also various ways to obtain a result, it is difficult to generalise as to whether a test hurts; it would depend on the specific test and should be discussed with the physician.

For example, a test might involve temporary physical discomfort; hardly anyone would consider it a pleasant experience to have a heavy lead protective apron draped around their shoulders while their jaw is being X-rayed.

Likewise, tests that analyse a human sample – such as blood, tissue or hair – might involve temporary pain. To take blood, a Physician has to prick the patient's skin. Collecting a tissue sample might be more complex, ranging from the removal of a small area of skin to local anaesthesia so that a physician can, to name just one example, collect bone marrow cells from the hip bone.

In addition to physical pain, a test might cause emotional discomfort; both nervousness prior to taking the test and the anxious wait for a result could put serious strain on a person.

5. How accurate are the different types of tests?

Several characteristics can be used to describe the quality and usefulness of a test; accuracy is one of them. However, there is more than one way to measure the accuracy of a test. For instance, accuracy can be measured by the likeliness of a true positive result, or alternatively on the overall probability of a correct result, be it positive or negative.

In addition, the specific accuracy of a test not only depends on how accuracy is measured, but also on how and when the test is taken. For example, many home pregnancy tests claim an accuracy of 99%, but accuracy is significantly lower if the test is taken very soon after a missed period/menstruation. It is also possible that the test shows an inaccurate negative result because an insufficient amount of urine was utilised.

Regardless of a test's specific accuracy, an incorrect result can never be completely excluded. If a test reveals a result that would entail far-reaching consequences for the tested person, it is a good idea to repeat the entire test with a new sample and confirm the result. Medical tests are not fool-proof. Sometimes it is better to take the same test twice with the same result, than to risk significantly changing one's lifestyle only to find out at a later point that it was unnecessary or inadequate.

In any case, critical test results should be discussed with a physician, whose experience should help to analyze accuracy of the test results. This also applies to tests bought online.

6. How can a person understand what a proposed test is intended to reveal?

Nobody should take a test unless they are confident that they understand all aspects of it, as well as its potential implications. Physicians are responsible for informing their patients' not just about potential benefits, but also about the exact procedure, the risks and the potential consequences that accompany a particular test. For instance, studies have indicated that out of one thousand women participating in regular mammography-screening, to detect breast cancer in early stages, four die of breast cancer. Without regular screening, it would be five. Whether this reduced risk is worth the annual exposure to X-rays , and what alternative methods of prevention there are is a matter physicians should discuss with each individual patient.

In order to get a good idea of the test and its implications, it is worth considering asking the following questions:

- Why does the physician suggest taking this test?
- Are there costs?
- Will my insurance cover the costs?
- What is the procedure? Does it hurt?
- Are there any risks?
- Are there alternatives, e.g. other, less invasive tests?
- What, precisely, is the potential benefit?
- How long will it take to get a result? How will I be notified of the outcome?
- How accurate is the result?
- Who will know that a test has been performed? Who will learn about the test results?
- What would a positive or negative result entail for the patient?
- What would a positive or negative result entail for the patient's family, partner or possibly even friends?

In the case of tests taken at home (e.g. home pregnancy test), a patient should discuss the test with the physician afterwards or, even better, ensure there are no questions before actually taking the test.

7. Will someone want to learn the results of a medical test even if it brings bad news?

That is a highly personal decision that no one else can, or should, make in the place of the affected person. Every person has a right to know, just as much as they have a right not to know.

In some cases, the result of a medical test might be valuable in that it allows a physician to suggest lifestyle changes that might prevent a disease from developing, or might limit its negative impact. For instance, if a patient's blood test reveals abnormally high levels of cholesterol, a physician can provide that patient with dietary advice to help reduce cholesterol levels. Here, the bad news in the form of a test result may ultimately help to prevent worse consequences. It is for this very reason that a patient might want to know a result even if it's a bad one given it is of immediate medical value.

On the other hand, there are cases when the bad news implied by a test result does not come with a treatment option.

To take a rather prominent example, Huntington's disease can be detected with a genetic test which can be performed at any age, well before the occurrence of the physical symptoms of the disease which typically manifest between the ages of 35 and 45. Its causes lie on a single gene. Unfortunately, the knowledge that a person carries the dominant disease gene does not come with the option of a cure as none is currently available. On the other hand, the knowledge that an individual carries the gene might play a role in the person's decision to have children or not, as each child of an affected parent has a 50% risk of inheriting the mutated gene and therefore being affected with the disorder. For others, the knowledge that they tested positive for Huntington's might make them decide to have children at a younger age already.

As far as children of affected parents are concerned, some might prefer not to be weighed down with the certainty of carrying this disease as well. Others might go the opposite route and take a test so as to escape the constant uncertainty of not knowing. If the test shows they do carry it, the knowledge allows them to take the necessary personal steps, to plan and make decisions. For some, reducing anxiety and uncertainty can, in and of itself, already be of value.

The question of whether to take a test, particularly one that might reveal a critical disease, is a complex and highly personal one, and the answer will depend on individual circumstances. Everyone has a right to know, and, simultaneously, the right not to know.

Either way, a decision should not be rushed. A person should be well-informed about what the test is intended to reveal, as well as the potential consequences. They should be able to fully grasp the psychological risk so as to make a decision that is in line with personal goals and values as well as potential consequences for their family.

Conversations with confidants, a physician or a genetic counsellor can only offer guidance; the final decision whether or not to take a genetic test is entirely up to the person taking that test. There is no right or wrong choice, only a choice that is right for one person and, ideally, their family.

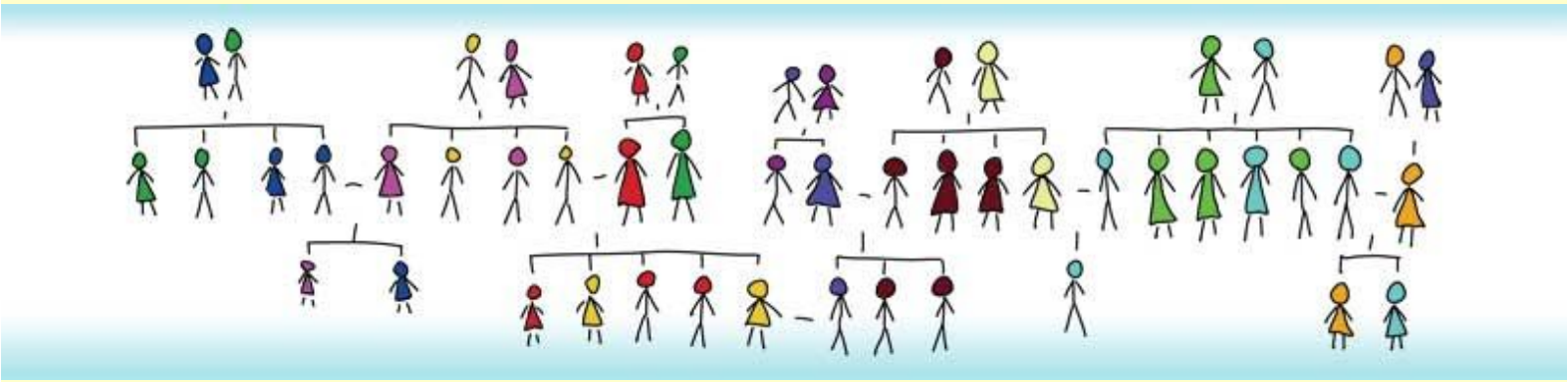
8. What can a person do to understand the results of a test after taking it?

A test should not be taken unless the person is certain of what it is supposed to reveal, the procedure and circumstances as well as the potential implications of different results (see Question 6). In some cases, people have to sign an informed consent form before taking a test, stating that they have been adequately informed about all relevant aspects of the test.

However, given that medical tests are complex and the results often not as straight-forward as one might expect, it frequently happens that new questions, or a need for clarification, arise after receiving the results. In that case, one competent source of information should be the physician; physicians should be able to help with the interpretation of a result, and to explain its consequences. Other sources of information are specialists in the disease field or genetic counsellors in the case of genetic tests.

A person might also wish to talk to other patients, so as to get a better understanding of the result implications and what these mean in daily life. Usually, a physician will be able to provide the patient with addresses of support groups that bring together people suffering from the same condition. Hospitals and local health authorities might also be of help. Another tool for finding people suffering from the same condition can be through trustworthy internet pages. For instance, see the Open Directory Project (see links at the end of this brochure)

In any case, it is important to have any open questions answered. Only a full understanding of the test results will enable a person to make the right decisions for their life and gauge the potential consequences for their family.



9. Will there be implications for a person's family if the results show them to potentially suffer (or not suffer) from the disease the person was tested for?

Since potential implications for a person's family depend on the disease in question, the matter should be discussed with the physician both prior to taking a test and after having the results.

There are several scenarios for diseases that could affect a person's relatives:

- While some conditions come without any direct consequences, others might require a change of lifestyle or a severe treatment which, by extension, could also affect a person's family.
- There is also the case of infectious diseases, such as HIV or tuberculosis. A result revealing such an infection will impact family and friends, as certain precautions have to be taken to protect them.
- Also complex are the consequences of monogenetic diseases. A parent with a disease-causing gene mutation may have passed that mutation on to a child, or might pass it on to future grand-children. Also, should a person be diagnosed with a genetic disease, their siblings may also carry or suffer from the same condition.

Therefore, genetic or infectious diseases are likely to affect a patient's close relatives more directly than diseases that are not genetically inherited or transmittable. This raises the question of informing them about the test and its result (see Question 10).

10. Who has to inform people about the result of a test? Is it possible to get help?

There is generally no obligation to tell anyone about the result of a medical test, or even about taking a test in the first place. Exceptions exist, of course:

- When it comes to infectious diseases such as HIV or tuberculosis, the obligation to inform others may not only be a moral obligation, it may also be of legal relevance. For instance, some countries or states oblige a person who tested positive for HIV to disclose this to sex or needle-sharing partners; otherwise, they can be charged with a crime.

- A disease that might lead to a person endangering others during the course of their work should be disclosed (see also Question 11)
- Another exception are life insurance companies, which may require the disclosure of previously taken genetic tests when policies are requested (see also Question 11).

Putting aside those few instances when disclosure is mandatory, there are a number of reasons why a person might want to tell relatives and friends about taking a test of their own accord:

- In the case of a diagnosis revealing a genetically inheritable disease, the patient should be aware of what the diagnosis implies for close relatives, who might be affected by the same disease (see also previous question).
- In fact, with genetic tests, it is common to involve the family beforehand, e.g. to compile a proper family tree with information on diseases occurring within the family.

The most important reason for telling family and friends about any kind of critical test, however, is that it often helps to confide in someone who will listen and share one's hope, pain and sorrow. Confiding in someone means not having to bear the burden alone.

That said, informing their close ones is not something a patient has to do alone. Usually, a physician will be willing to sit down with the patient and a family member, or friend (or a small number of family members or friends), to discuss the findings of a test and answer questions which may arise. Other patients may prefer a family conference to reveal the news instead of informing friends and relatives one by one, or they may ask a fellow patient, who might also be a member of a support group for the disease in question, to accompany them when telling people.

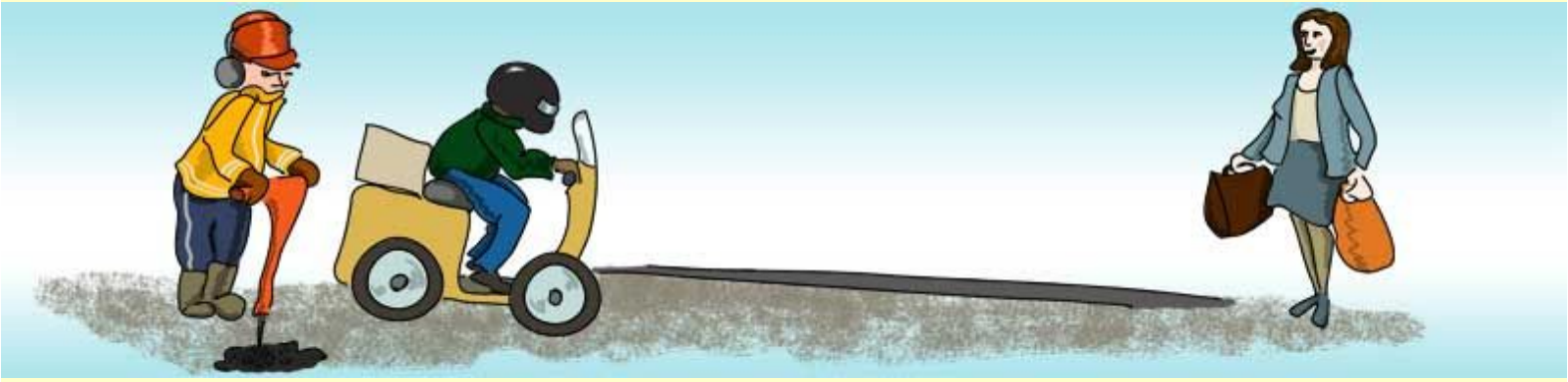
To summarise, each patient will have to find his or her individual way of dealing with the decision, and decide whether or not to tell people. A physician or another healthcare professional, e.g. a nurse or a genetic counsellor, should help the patient to make that decision by supplying them with the necessary information and giving support without pressuring the patient in any way.

11. Who will see the results of any tests carried out?

This is a matter that should be discussed with the physician before taking the test.

Unless exceptions have been made by the patient; physicians and other medical personnel are bound to professional discretion, meaning they are not allowed to share a person's test results with third parties without the patient's consent. The only people who should have access to the outcome of a particular test is the patient's physician and the medical and laboratory personnel involved in analysing the sample. In most cases, however, laboratories receive samples in an anonymous manner.

Employers or an insurance company are not normally informed of test results (see also next question).



12. Will a diagnosis affect employability or insurance?

Different countries come with different legal situations, and thus with differing answers to this question. Typically, however, the laws against discrimination as well as data privacy regulations apply to people diagnosed with a disease just as much as they apply to gender and race. On the other hand, it is true that insurance companies might be tempted to insure healthy people rather than those diagnosed with a disease. Likewise, employers might prefer a healthy employee over one likely to fall ill. For that reason, a person might wish to keep a test result secret.

In general:
The result of a test will become part of a patient's medical record. However, physicians and other medical personnel are bound to professional discretion (see also previous question).

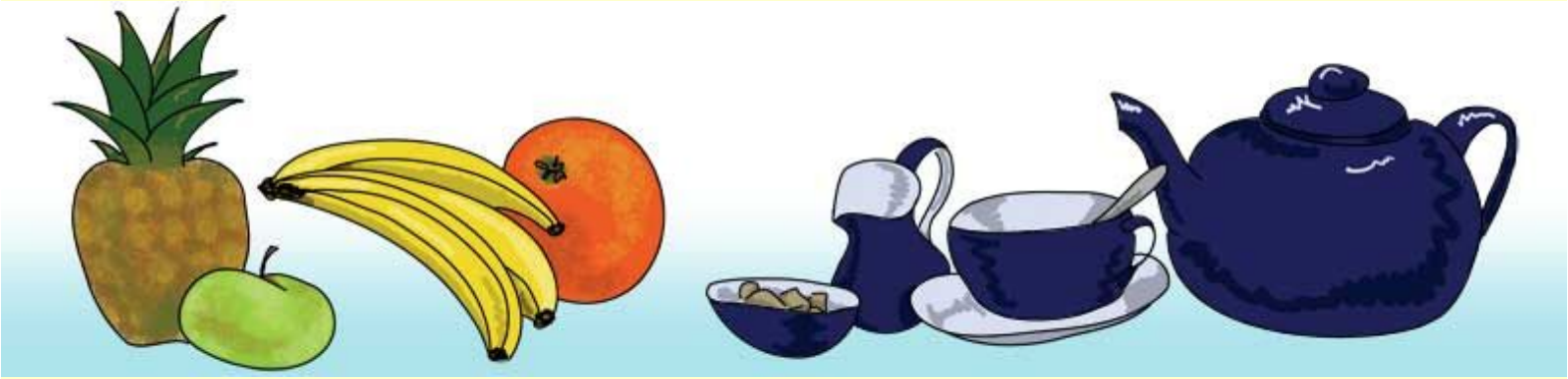
Health insurance:
A distinction must be made between basic and additional health insurance:

- *Basic insurance* generally does not require the disclosure of test results. While an insurance company might get notified that a test has been taken, simply because it has to pick up the bill, the results are strictly confidential.

In some cases, the simple information that a test has been taken might already be sensitive in nature as it suggests a potential risk. Taking a test anonymously is possible, but has to be specifically arranged with the responsible physician prior to the fact. The result will, once again, become part of the person's medical record, unless they explicitly state they don't want that to happen.

- If someone requires an insurance that goes *beyond a basic health insurance* (e.g. a private life insurance that exceeds a certain sum, mostly according to national legislation), that person is obliged to truthfully answer questions about previous tests and their results. Otherwise, the insurance company can reduce the benefits of a policy or even cancel the contract if a person has been diagnosed with a high risk for a certain disease and that disease later surfaces, at least if the insurance company can prove the insured person knew about the high risk when signing the contract.

Employment:
While there is no obligation to inform an employer about a test result, it is highly recommended if it means a risk for others while the diagnosed person is performing his or her job, e.g. flying a plane.



13. Will a positive result entail lifestyle changes?

Generally, being diagnosed with a condition doesn't mean having to put one's life on hold, but it might require some adjustments.

For example, a bus driver diagnosed with epilepsy will not be able to continue driving a bus; someone diagnosed with diabetes will most likely have to adjust their diet accordingly. It could also mean changing one's lifestyle, e.g. doing more physical exercise if diabetes goes hand in hand with being overweight, or not driving when diagnosed with epilepsy.

Therefore, a test revealing a certain medical condition can very well mean having to change certain habits. On the other hand, changing one's lifestyle might improve a person's condition, and with that their quality of life, or keep it from deteriorating. In those cases, a change of lifestyle could also be seen as a chance that might not always be easy to take, but that is ultimately worth it.

14. If persons suspect they might suffer from a specific disease, should they consider buying a test off the internet rather than going to see a physician?

While there are tests, such as pregnancy tests, that are so common that they can be bought in supermarkets, the same is not true for more invasive or critical tests such as genetic tests. Generally, medicinal products bought off the internet have a significantly higher risk of being counterfeit suggesting that they might not work, or in some cases even hold the potential of physical harm. Official distribution chains such as pharmacies are much better controlled.

In nearly all cases, consulting a physician is the best option. Not only can physicians ensure that the test a patient takes is genuine, they also supply additional information. It is the physician's responsibility to explain the purpose of the test and what will happen with it, as well as what the consequences of a positive, meaning affirmative, result might entail. Buying a test off the internet does not come with such additional advice and support. Especially when dealing with a test that might entail severe consequences for a person's life, adequate preparation is of great help and will hopefully soften the impact of a potentially positive result.

15. If more than one company is selling tests for the same disease, how can one choose between them?

Ideally, this is a decision that is made with the aid of a health professional such as a Physician, who is likely to have some prior experience with the handling and accuracy of a test. In the case of over-the-counter tests, a pharmacist might also be able to offer advice.

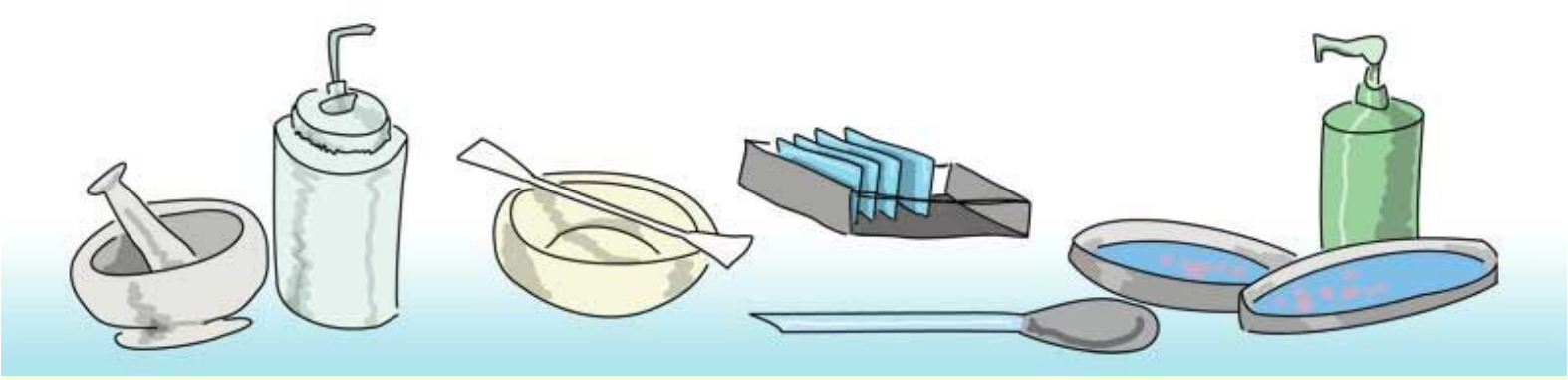
One important point that should be noted is that the cheapest test is not always the best choice; in some cases, handling is more difficult or accuracy is relatively low. The test might thus have to be repeated, leading overall to higher costs. Again, health professionals should be able to assess the different aspects of a test and help with a decision.

16. If a person refuses to take a test, what will happen to their medical care?

In general, no one can force a person to take a medical test. Like any form of medical examination, taking a test is a personal decision, and the patient's autonomy must be guaranteed. A refusal should therefore come without any negative implications for the person's care.

On the other hand, it is possible that a medical test might help a physician to optimize a patient's medical care. Without it, only part of the picture might be visible, which could lead to treatment decisions that are suboptimal.

Insurers usually cannot force a person into taking a test or having a medical examination by a physician. There are country-specific exceptions when it comes to uncommonly high insurance sums, however; e.g. in Switzerland, a person taking out a life insurance of more than 300'000 Swiss Franks has to go through a thorough examination.



17. What happens to the samples taken for the test afterwards? Can they be used for other purposes?

No, a sample may be used only for purposes that the donor has explicitly agreed to. It is therefore not feasible for a physician or a laboratory to run it through other tests, or offer it to a human specimen repository (a so-called biobank) without first retrieving the express consent of the sample owner (see also link related to FAQ on biobanks at the end of this leaflet).

18. If someone is diagnosed with a specific disease, they will want to see an expert for their condition. How can they find out who that expert is?

A patient's general physician should be able to provide names of specialized physicians and should, if necessary, help the patient to arrange an appointment.

Other good sources of disease-specific information are people affected by the same disease. Given, they too faced the same questions, after their diagnosis, they often have valuable information. In particular, they will most likely have seen different experts and will thus be able to recommend someone. There are also self-help groups and patient networks for a wide variety of medical conditions (see also Q 8). If the physician cannot supply a patient with addresses, trustworthy internet pages, for example the Open Directory Project (see links at the end of this brochure), may be of assistance.

19. Persons diagnosed with a specific disease might want to participate in research. How can they find out what is going on?

There are two ways for patients to be directly involved in research. The first option is to donate a sample to a human sample repository, often referred to as a biobank. Analysing a quantity of these samples might help researches to discover a new treatment or to develop improved tests for a particular condition.

The second possibility is to take part in a clinical trial. Clinical trials focus on new, innovative treatment methods that have shown the potential to improve the condition of people diagnosed with a certain type of disease. At the stage where patients can participate in a clinical trial, treatment tends to be fairly advanced.

A patient who wants to find out whether there is a clinical trial that fits their condition can ask their physician or search via various websites. These generally list ongoing trials as well as the results of finished trials or provide other relevant websites (see links at the end of this brochure).

For more information on clinical trials and biobanks, two other FAQ leaflets might be beneficial and already address some additional questions (see links at the end of this brochure).

Another way for patients to become active is through a patient organisation. These interest groups exist for most medical conditions (see links at the end of this brochure). Some of them have set themselves up not only for providing patients with a network, but also for informing and influencing society so as to benefit their members. These groups are interested in moving research forward and will provide anyone seeking involvement with addresses as well as advice on ways to further their cause.

20. Links

- Open Directory Project – List of health support groups: http://www.dmoz.org/Health/Support_Groups/
- Clinical trial websites: http://www.clinicaltrials.gov
- International search Portal for clinical trials: http://clinicaltrials.ifpma.org
- The portal for rare diseases and orphan drugs includes a search engine for clinical trials and research projects: http://www.orpha.net
- Clinical Trials FAQ: http://www.biomedinvo4all.com/en/publications-and-downloads/faq-on-clinical-trials/ (German Translation: http://www.biomedinvo4all.com/en/publications-and-downloads/klinische-studien-n-informationsbroschure/)
- Biobanks FAQ: http://www.biomedinvo4all.com/en/publications-and-downloads/faq-on-biobanks/ (German Translation: http://www.biomedinvo4all.com/en/publications-and-downloads/biobanken-n-informationsbroschure/)

More FAQ leaflets

FAQ leaflet on Biobanks:
http://www.biomedinvo4all.com/en/publications-and-downloads/faq-on-biobanks/

FAQ leaflet on Clinical Trials:
http://www.biomedinvo4all.com/en/research-themes/clinical-trials

Leaflet with Glossary of terms:
http://www.biomedinvo4all.com/en/research-themes/clinical-trials

Political Information on PHC

Euractiv.com – European Union Information Website
http://www.euractiv.com/en/health/personalised-healthcare/article-171796

International

U.S. Department of Health & Human Services
http://www.hhs.gov/myhealthcare/

illustrations by www.rebeccajkent.com