

# GENETIC TESTS WITHOUT PRESCRIPTION

Overused tests, as indicated by the Italian Society of Human Genetics

The human genome has now been completely mapped out. However, this does not mean that a genetic test is able to give us answers to everything we're looking for regarding health, illnesses, athletic capabilities and whatever else. The availability of genetic tests sold on the internet, in pharmacies, gyms and beauty salons, at relatively affordable prices, has stimulated demand, and more and more people choose to take one.

## NOT WITHOUT A DOCTOR

The results of these tests have important consequences for patients and their families. The tests may influence decisions and, when not carried out correctly, can sometimes lead to excess diagnoses and treatments. Italian law, in accordance with European law, requires that specialist doctors request genetic tests after appropriate advice. The laboratories that carry them out must comply with strict accreditation requirements, meet precise standards and employ personnel specialised in genetic medicine.

It is therefore clear, to begin with, that the so-called tests directed at consumers (purchased directly by the public) will not meet the essential requirements of these laws. Furthermore, the results may not be

clear and may cause people unjustified stress and anxiety, alarming or reassuring them based on inaccurate conclusions regarding the risk of contracting a disease.

## A PIECE OF THE PUZZLE

The potential lack of precision and inability to read and properly comprehend results are on their own sufficient reasons not to be seduced by genetic tests directly available to consumers. There is, however, another



underlying issue which relates to the role that genetic code has on diseases. It is important to understand that genetic risk is only a piece of a much bigger puzzle. Besides some rare exceptions, the diseases being studied through susceptibility testing are multi-factorial conditions in which a particular set of genes interacts with many environmental factors. From smoking to alcohol and from diet to exercise, lifestyle has a tremendous effect on the chances of contracting a disease, while the effect of genetic factors is often modest.

## LET'S UNDERSTAND WHY

In some cases, a traditional blood test tells us more about the probabilities of developing a disease than a genetic test does. For some diseases, we know rather efficient risk markers, like levels of cholesterol and triglycerides in the blood, which are associated with the risk of heart disease. Some genetic factors that influence this risk indeed act on cholesterol and triglyceride levels. These, though, are also the result of a mix of environmental and lifestyle factors. It is perhaps more useful to measure them in the blood than to analyse the genes that control them, as these would provide only partial information.

To learn more:  
Project "Doing more does not mean doing better – Choosing Wisely Italy"  
[www.slowmedicine.it](http://www.slowmedicine.it)  
[www.choosingwiselyitaly.org](http://www.choosingwiselyitaly.org)

## WHEN THEY ARE TRULY USEFUL FOR SPECIFIC ILLNESSES / DISORDERS



- Genetic tests are used in medical practice when they can provide reliable and clinically valuable answers to a precise question. This is especially true for diseases determined exclusively or predominantly by genetic factors.
- In the cases of: thalassemia, cystic fibrosis, muscular dystrophy, Down's syndrome and hereditary predisposition to breast or other kinds of cancer, the test serves to highlight (in most cases, with certainty) a genetic predisposition to the emergence of the disease.
- Since such diseases are quite rare in the general population, these tests are usually requested when there are specific risk factors, giving family history particular importance.
- Moreover, it is important to evaluate whether the test result has an impact on clinical practice. For example, a test that highlights gene alterations BRCA1 or BRCA2, responsible for breast or ovarian cancer, suggests opportunities to look to measures of prevention or early diagnosis such as frequent radiological checks or surgery.