What is sudden cardiac arrest?

Sudden cardiac arrest (SCA) is showed by a rapid pulse absence and an unconsciousness state caused by the inability of the heart to pump blood into the brain, and into the body, in an effective way. Usually sudden cardiac arrest is caused by potentially lethal arrhythmias and by some cardiac electric system abnormalities. It's defined "sudden" because of its nature, so it can affect any person in any place without any notice, even individuals who have never been previously diagnosed cardiac disease or critic clinical conditions.

What are cardiomyopathies?

Cardiomyopathies are a group of heart muscle diseases, often genetically determined with different modes of transmission, which can show symptoms that limit the functional capacity of the heart, and that involve complications such as atrial fibrillation, heart failure, stroke and, rarely, malignant ventricular arrhythmias and sudden death.

GENOMA Group: a great patrimony of technologies and human resources, a model of quality, professionality and competences

GENOMA is a highly specialized diagnostic centre of national relevance, recognised for its contribution to the molecular diagnostic process. Research projects and collaborations with scientific networks confer it an international esteem.

Founded in 1997, **GENOMA** works as reference point for high technology exams, executing mainly "service" activities of **genetic, cytogenetics and molecular analysis** of elevated specializations, both in prenatal and postnatal field.

GENOMA is considered one of the most innovative European centre of **molecular diagnostic**, and has two main headquarters in **Rome** and **Milan**. Laboratories are situated in a modern and *high tech* structure, and they develop on a total surface of over **4000 square meters** and they are characterized by instrumental equipment and high technologies, as well as elevated qualitative standards.

GENOMA's activity extends to the whole **national territory**, in **Europe**, **USA** and **Middle East**, providing specialized diagnostic support for healthcare structures, publics and privates, analytics labs, nursing homes, hospitals, private clinics, outpatient clinics, medically assisted procreation centres and doctors specialised in different disciplines.

There are more than **5.000** Italian and foreign **Hospital** and **Healthcare structures** that deliver biological samples to our laboratories daily. The integrated organization of the Centre is structured to develop impressive amount of work: the annual number of services, which is constantly growing, passes **100.000** genetic tests.

One of the characteristics that marks the Group is represented by an articulate services diversification, which is one of the most complete on national and international territory, and allows offering to patients and their doctors, innovative diagnostic solutions for every clinical need. With a list of more than **1.500 genetic tests** that can be done in the headquarter, distributed in more than **20 application areas**, **GENOMA** is able to satisfy always more specialized demands in molecular and cytogenetics diagnostic field, providing its clients a rapid, precise, effective and high specialized service.

The quality and the diversification of performance, the efficiency and reliability of services, along with a policy of continuous investments in high technological content and innovative sectors, allowed **GENOMA** to become a leader company in the genetic diagnosis field, great assets of technology and human resources. **GENOMA** means a model of quality, professionality and competencies to imitate and export, a reference point for multiple national and international healthcare structures, both public and private.

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Cardioscreen®

A sophisticated test that allows us to do a multiple genetic analysis to evaluate the presence of mutations associated with sudden cardiac death and with inherited cardiomyopathies

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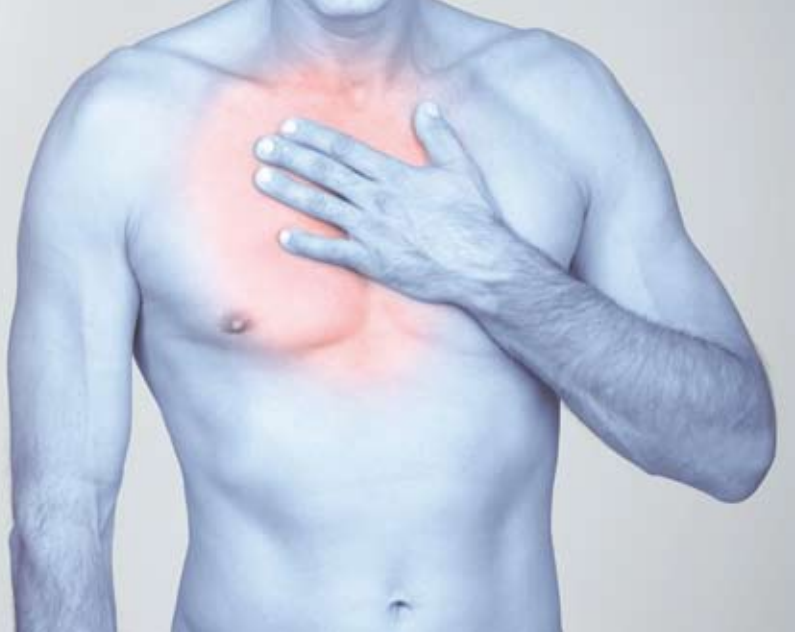
Cardioscreen®

The test that identifies patient **at risk** of **sudden cardiac arrest** or **inherited cardiomyopathies**



Cardioscreen® is a genetic test available in two editions:

Cardioscreen® - Cardiomyopathies

useful to evaluate the presence of mutations associated with sudden inherited cardiomyopathies and allows us to identify patients at genetic risk of potentially mortal cardiac events.

Cardioscreen® - Prevention of sudden cardiac arrest

is dedicated to the evaluation of the presence of mutations associated with sudden cardiac death *sine materia* (without evident pathological cardiac aspects).

Who should submit the Test?

Both **Cardioscreen®** tests are recommended for those who knows about a case of sudden cardiac death in their family (included sudden infant death), heart failure or transplant, which suggest inherited cardiac pathological substratum.

Furthermore is particularly useful to do a genetic screening as a prevention instrument in case of:

- Professional or amateur agonistic activity, also for individuals with no familiarity;
- Young individuals (younger than 40 years) with idiopathic cardiac symptomatology;
- Children and teenagers with a suspect clinical picture for QT anomalies or cardiac rhythm.

What are the benefits of the test?

The possibility to identify an at-risk individual for inherited cardiomyopathies or for sudden cardiac arrest, represents today the best method to express an early diagnosis of a potential pathology, and, therefore, to reduce related mortality and morbidity. Members of inherited high-risk families, and in particular who is affected by an idiopathic cardiac symptomatology, can ask for a genetic consultation and discuss the own genetic-clinical situation with the geneticist. This evaluation will be able to promote the genetic test to verify if the patient is carrier of a mutation associated to an inherited cardiomyopathy and sudden cardiac arrest. If the test is positive, the examination will be extended to patient's relatives to identify at-risk individuals of the nuclear family.

The information obtained from the genetic test can generate remarkable **benefits**, such us:

- the identification of family members at **high risk of inherited cardiomyopathy**;
- the organisation of an adequate **medical examination program** reserved for high risk individuals so to facilitate the adoption of the most effective preventive measures (for example implantable defibrillators or antiarrhythmic pharmacologic therapies);
- the knowing of the possibility of **transmission of genetic mutation** to the progeny and the identification of individuals children with germinal genic mutations at high risk.

How is the test done?

The test is performed through the taking of a haematic sample. By means of a complex laboratory analysis, the DNA is isolated from nucleated cells and amplified by **Polymerase Chain Reaction (PCR) technique**. Later, thanks to an innovative technological process of **massive parallel sequencing (MPS)**, which employs **Next Generation Sequencing (NGS)** techniques using **ILLUMINA** sequencer, they completely sequence at an elevated in – depth reading, **43 genes** (exons and adjacent intragenic regions, ± 5 nucleotides) connected to inherited cardiomyopathies for "Inherited cardiomyopathies", and **157 genes** (exons and adjacent intragenic regions, ± 5 nucleotides) connected to inherited cardiac pathologies correlated to sudden cardiac arrest for "Prevention sudden cardiac arrest".

Achievable test results

"POSITIVE" - Presence of one or more mutations: it indicates the test has revealed one or more mutations of one (or more) gene related to sudden cardiac arrest. Our geneticist, during genetic counselling, will explain in a detailed way the meaning of the test result.

A positive result doesn't mean that the patient to whom the mutation has been found will have a critic pathological cardiac event, but it means that the patient shows a greater risk compared with the general population.

In a suspect situation, the test is useful for confirming diagnostic hypotheses that has to be verified.

"NEGATIVE" - Absence of mutations: it indicates the test didn't detect the presence of mutations in the analysed genes.

However is important to underline that a negative result doesn't mean the patient has zero risk to meet with a potentially critic cardiac event or to develop a cardiomyopathy in the own lifetime; the risk for this people is the same as for the general population, this because not all forms of cardiomyopathy and sudden cardiac arrest has to be connected to genetic causes.

Accuracy of Cardioscreen® test

Current DNA sequencing techniques produce precise results and its accuracy is estimated to be superior to 99%.