www.carrieradvance.it

GENOMICA
Next Generation Genetics

The most advanced carrier screening test

An advanced carrier screening test allowing identification of couples who are at risk of passing inherited disorders to their children.

CARRIERADVANCE

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CarrierAdvance is an innovative genetic test that allows identification of couples at risk of transmitting the most common genetic diseases to their children, by detecting DNA mutations of which one or both parents may be unwitting carriers.

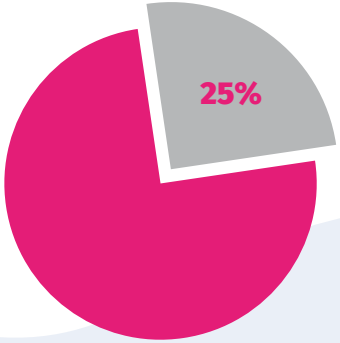
Each person is born with genetic characteristics that differentiate him/her from others and that make this person unique.

While most of the differences in the DNA sequence between different people is harmless, some changes, kown as **"mutations"**, can alter genomic functionality and make that person a carrier of a specific genetic disease, transmissible to their children.

In most cases, carriers of genetic diseases are healthy individuals and show no symptoms, have no known family history of the disease, and are unaware that they are at risk of passing this DNA "error" to their children.

If both partners of the couple carry mutations in the same gene with autosomal recessive inheritance, there will be an increased risk (25%) of conceiving a child with that specific disease. If a woman is a carrier of a X-linked disorder, 50% of male children will be at risk of having that specific genetic disease.

The risk of conceiving a child with a specific genetic disease is 25%



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INDICATION FOR TESTING

- **For patients who are pursuing pregnancy with assisted reproductive technologies.**
- **Couples requiring gamete donation, in order to select the most appropriate donor for each recipient (i.e. a donor that doesn't carry the same mutation as the member of the couple who will provide the gametes), minimizing the reproductive risk.**
- **Anyone who wants to know if they are carrier of any condition included in the panel.**
- **Individuals with a family history of a genetic disease, who are therefore at higher risk of being carriers for those diseases**

Test is intended to be used as a family planning tool, allowing patients to be tested individually or with their reproductive partner for their risk of having children with various genetic conditions.

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3 LEVELS OF SCREENING

LEVEL	GENE	DISEASES INVESTIGATED
CARRIERADVANCE F *Focus*	30	**31** recessive and X-linked **genetic disorders**, including the most common in the European population
CARRIERADVANCE P *Plus*	925	**1467** recessive and X-linked **genetic disorders**, including the most common in the European population and those recommended by prestigious international societies, such as ACMG and ACOG
CARRIERADVANCE X *Exome*	4000+	Clinical exome sequencing, including **5000+** recessive and X-linked **genetic disorders**, compatible with the majority of carrier screening test available in the market

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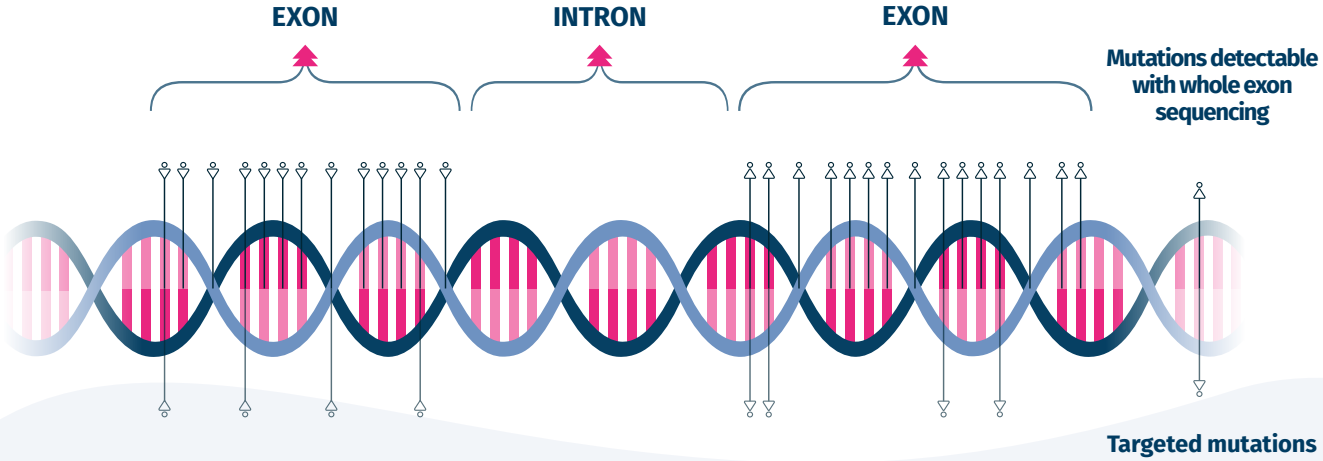
A GROUNDBREAKING TECHNOLOGY COUPLED WITH AN ADVANCED BIOINFORMATIC ANALYSIS TO DELIVER GREATER ACCURACY



CarrierAdvance uses a state-of-the-art technological process, named **Next Generation Sequencing (NGS)**. Unlike other carrier screening tests using targeted sequencing, CarrierAdvance is carried out performing full-exon sequencing of all the genes included in the panel, which allows a more comprehensive assessment of each gene and related diseases.

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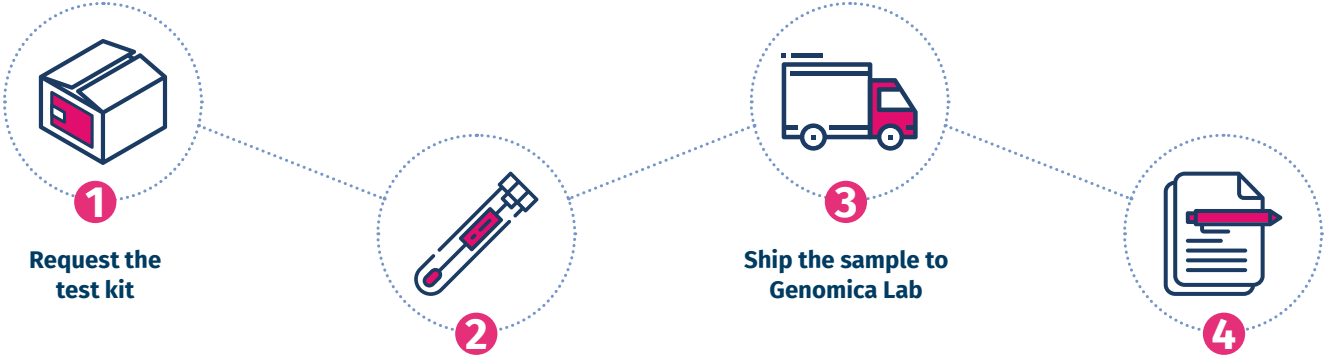
4 EASY STEPS



Types of samples	Peripheral blood	Buccal swab

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TEST RESULTS

Identification of one or more mutations: *this test result indicates that one or more mutations have been detected in the targeted genes screened, identifying the patient as a* ***carrier****.*
Mutations reported in a POSITIVE CarrierAdvance test may be classified under the following prognosis categories:
Known pathogenic: *clinical relevant mutations causing well-established syndromes;*
Likely pathogenic: *variants that are likely clinical relevant and may cause well-established syndromes.*
Classification follows the recommendations of the international reference guidelines[1]

Variants of uncertain clinical significance (VUS): *this test result indicates that one or more variants of uncertain clinical significance have been detectedin the targeted genes screened. These are variants with insufficient evidence available for unequivocal determination of clinical significance. VOUS will only be reported if both partners perform the test and one of them is found to be carrier of a known pathogenic mutation.*

No mutations identified: *this test result indicates that not known or likely pathogenic mutations have been detected in the targeted genes screened. A single test cannot always detect all possible genetic changes. Hence, a negative test results do not completely rule out the the possibility of carrying a mutation located in a region of the genome not investigated by the test.*

1 Richards et al. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genetics in Medicine. 2015;17(5):405-423.

Turnaround time **15 days**

GENOMICA
Next Generation Genetics

Advanced molecular diagnostics solutions using state-of-the art technologies

GENOMICA is recognized as one of the most advanced molecular diagnostics laboratory in Europe, both for the state-of-the-art instruments and technologies, as well as for its high quality standards.

With a **comprehensive portfolio of over 10.000 genetic tests**, GENOMICA is able to satisfy increasingly specialised requests in the field of molecular genetics, providing physicians and their patients with innovative and highly specialised diagnostic solutions for any clinical need.

- **Professionals with 20+ years experience** in the field of genetics and prenatal molecular diagnostics
- Laboratories with **groundbreaking technologies** and high quality standards
- Over **100.000** genetic tests/year
- Fast TAT 15 days
- **Test performed in Italy** (Rome or Milan)
- Dedicated R&D team
- International **Partnership**
- **Personalized genetic counseling** with genetic counselors experts in discussing genetic test results and familial risks

LABORATORIES

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