Patient Informed Consent for GeneScreen® Carrier Screening test

The accuracy of the genetic testing and reporting methods have been determined and verified to meet required regulatory performance standards by Eurofins Genoma Group Lab ("Genoma"), a licensed and accredited laboratory.

I understand the following information regarding the general purpose and benefits of testing:

The purpose of this genetic test is to identify couples who are at risk of passing inherited disorders to their children. Expectant parents, or couples planning a family, may be at risk for passing on severe genetic diseases to their offspring. If both parents are carriers for an autosomal recessive or X-linked condition they have a 1 in 4, or 25%, chance of having an affected child. The **GeneScreen® Carrier Screen** provides a closer look at genes, to see if the couple is at risk of passing a hereditary genetic disorder to their offspring. **GeneScreen® Carrier Screening Test** allows for a comprehensive care and enables patients to make more informed reproductive decisions. Offering **GeneScreen®** to a patient before pregnancy allows her to gain knowledge about her reproductive health early.

I understand the following information regarding GeneScreen® results:

- The **GeneScreen® Carrier Screening 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. This test is intended to identify couples with high reproductive risk. Thus, concurrent testing of both prospective parents is highly recommended.
- A positive test result indicates that one or more mutations in one or more genes were detected, identifying the patient as a **carrier**. Being a carrier is relatively common, carriers do not generally involve the disease development and do not show any symptoms. A patient with a positive test result should be referred for genetic counseling and further evaluation. The patient's reproductive partner and at-risk family members may also be tested. When both parents are carrier of the same genetic disease, there is a 25% chance of transmitting this condition to their offspring. In these cases, genetic counseling is recommended to the couple.
- In some cases the result of this test may reveal DNA variations or mutations with an unknown or unclassifiable clinical significance with the current medical and scientific knowledge. Moreover, the detection of gene variations does not always imply that the person will develop a certain pathology or the severity of the related symptoms, nor when this person may have the disease.
- The interpretation of genetic variations is based upon the most updated knowledge available upon examination. Such interpretation may change in the future, when new scientific and medical information on the structure of the genome are acquired and may affect the evaluation of the genetic variations themselves.
- A negative test result indicates none of the disease causing mutation in the investigated genes were identified in my specimen. A negative result for the genes analyzed does not exclude the possibility that mutations are present in a region of the genome that was not explored during the analysis.
- While results of this testing are highly accurate, a negative result significantly reduces but does not eliminate the chance of being a carrier.
- This test analyses only genetic diseases and genes listed in technical report enclosed to the test report. The test does not detect other genetic diseases or genes that were not specifically targeted.
- In the course of performing the analysis for the indicated tests, information regarding other genetic disorders may become evident (called Incidental Findings), and that Genoma's policy is to NOT REPORT or comment on any Incidental Findings that may be noted in the course of analyzing the test data.
- Genoma may contact me for additional information or follow-up clinical history at any point after I undergo this testing.

I understand the general risks and limitations of genetic testing including the following:

- One tube of blood or a buccal swab will be collected to perform the test. Risks of having blood drawn are uncommon, but may include dizziness, fainting, soreness, pain, bleeding, bruising, and rarely, infection.
- Genetic testing should not be used as a substitute for treating and diagnosing conditions, or for the provision of health care services by a physician or other qualified healthcare professional.
- This test may not provide informative results for other reasons, such as: (1) non-genetic factors, (2) individual genetic variation, (3) insufficient scientific information about the relationship between genetic information and health outcomes, (4) various laboratory and non- laboratory technical reasons, and (5) incomplete gene sequence information.
- Other risks that may be experienced as a result of this testing include: unjustified alarm and/or false reassurance that can discourage preventive measures, related emotional issues, impact on life-changing decisions, potential genetic discrimination, and loss of confidentiality. The testing results and information may become part of my permanent medical record and may be available to individuals and organizations with legal access to such records.

Informed Consent Acknowledgement

I understand that this testing is voluntary and freely consent to this testing. My signature below acknowledges that:

- I understand written English;
- I have read and understood the front and back of this consent, all of my questions have been asked and answered to my satisfaction, and I agree to this testing. I understand that I can receive a copy of this consent by calling Client Services. (See "Questions").
- I have discussed with the healthcare provider ordering this test the reliability of positive or negative test results and the level of certainty that a positive test result for a given disease or condition serves as a predictor of that disease or condition;
- I am 18 years of age or older and have the legal authority to provide this consent and authorization for genetic testing, under all applicable laws.

Sede legale
00138 Roma - Via di Castel Giubileo, 11
C.F. e P.Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

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Web: www.genomamilano.it
E-mail info@genomamilano.it

CHIAMATA GRATUITA NUMEROVERDE 800-501651

- I understand Genoma may use my DNA and clinical information in medical research studies and for publication, if appropriate, unless I opt-out by initialing below. I understand that my name or other personally identifiable information will not be used in or linked by Genoma to the results of any studies and publications.
- I understand I can withdraw my consent to the analysis/examination with effect for the future at any time in full or in part without stating reasons.
- I have the right not to be informed about test results, to stop the testing processes that have been started at any time up to being given the results and to request the destruction of all text/examination results not already known to me.

☐ (**initial to opt-out**) **I do NOT consent** to the use of my extracted DNA sample and clinical information for anonymous medical research purposes. I understand this is deemed useful by Genoma and explained on the other side of this Consent.
☐ (**initial to opt-in**)**:** My DNA, extracted from my original specimen, and my clinical information can be retained for greater than 60 days and up to five (5) years after the completion of testing for anonymized medical research purposes as described above.

I understand that it is recommended that I obtain pre-testing and post-testing counseling to consider the purpose, meaning, risks, benefits, and limitations of, as well as any alternatives to, genetic testing in my particular situation, including medical issues based on my personal medical history. Pre-test counseling may help me better prepare to receive the test results and allow for advance consideration of medical options and the impact test results may have on me and my health. Post- test counseling provides a valuable opportunity to understand the medical interpretations of detected mutations, the psychological risks and benefits of learning my genetic test results, options for additional independent testing, among other things.
I understand that if testing results are inconclusive that I may be asked for an additional specimen(s). This consent is effective for any such additional specimen(s).
I understand the following information about confidentiality and disclosure of my personal information:

- My personal information and test results are confidential. While there can be no guarantee of privacy, Genoma has established reasonable safeguards to protect it. This information and the test results will be released to the ordering healthcare professional. I may request a copy of my lab results from Genoma' Client Services (see "Questions" below for contact information). For more information about my rights and Genoma' privacy practices, see Notice of Privacy Practices.
- This information and the results may also be disclosed if required by law, such as in response to a subpoena.
- I understand that if I share this information or these test results with anyone, I am responsible for any compromise of confidentiality that may result from such sharing.
- If I have opted out of allowing storage of my sample for research, the original specimen(s) may be securely stored for sixty (60) days from the date of collection and any remaining isolated DNA may be securely stored in accordance with applicable laws, regulations and standards. After such storage, the specimen(s) and the isolated DNA will be properly destroyed in accordance with applicable laws and regulations and the testing laboratory's standard operating procedures.

I understand the following regarding specimens for Medical Research Purposes: I understand that, unless I opt-out, as checked above, I am authorizing that my DNA extracted from my original specimen may be retained up to 5 years by Genoma as deemed useful for medical research purposes to develop new genetic tests. I understand that I may opt-in for this retention of my original specimen for up to 5 years by checking the correct box above. I understand that to protect my identity: a unique identifier will be assigned to my specimen and all resulting research data will be recorded, handled, and stored using this unique identifier. My name will be unavailable to any member of the research team and my identity will not be released or disclosed to others outside of Genoma. No compensation will be given me nor will I be owed any funds due to any invention(s) resulting from research and development using my specimen(s). I may refuse to submit my specimen for use in this way and this will not affect my results. Unless I indicate in the front page that I do not consent to anonymous medical research, I understand that my specimen(s) may be used in this manner.
I understand I may withdraw my consent: Under Italian regulations, Genoma cannot destroy medical records. However, at my written request and according to my instructions, Genoma can: a) destroy my DNA specimen(s) at the next regularly scheduled destruction cycle, b) delete my account, and c) move all medical information, including results report(s), into a into a secure, offline storage area with limited access. This means my account and results report(s) will not be searchable in Genoma systems by regular means and I, and my healthcare professional, will not be able to obtain a copy of my account information and results report(s) from Genoma. A request to withdraw my consent may be made to Genoma' Client Services (see phone number under "Questions" below).
Questions: If I have further questions about this testing, I understand that I can either contact a genetic counselor, other qualified healthcare professional or Genoma' Client Services at +39068811270, 8:00 AM to 8:00 PM, Monday through Friday. I may also call this number to set up an appointment to speak with a Genoma genetic counselor.

Patient's Name: _____ Patient's Surname: _____ Date of Birth: _____
Address: _____ City: _____ Country: _____
Signature of Patient or Legally Authorized Representative _____ Signature Date _____

Check one: ☐ Self ☐ Parent ☐ Legal Guardian ☐ Durable Power of Attorney for Health Care

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