GeneLinx

Informed Consent - Genetic Testing

The health professional requesting this test has provided the required genetic counseling to the patient regarding the purpose of the test, the procedure to be carried out, the limitations and risks, and the potential findings and implications. They also commit to resolve any queries before and after the genetic test is carried out.

Patient Full Name:	**Jane Doe**
Patient Date of Birth:	**01.01.1900**
Test being ordered:	**SCA7 (CAG expansion in *ATXN7* gene)**

1. Purpose of the test and implications

You acknowledge the following:

- A genetic test shall be performed on a biological sample provided by yourself (blood or other tissue) and a written report shall be issued with any clinically relevant findings (genetic disorders) related to the purpose of the test. The results of the genetic test may determine that you have an increased risk of suffering from or transmitting a genetic disorder, and may affect how you respond to a specific treatment.

- The results of this genetic test may have implications for yourself, your offspring and other members of your family. If this is the case, it is advisable that you inform your family members of the findings yourself.

- You have the right to not be informed of the results of this genetic test. If you choose to not be informed of the results, we may inform your family members (or legal guardians) if you give your consent and/or there are medically ethical grounds to do so, should this information be necessary to avoid serious harm to their health, as determined by the treating physician. Communication shall be limited exclusively to the information required for these purposes.

- At times, for certain genetic tests, we may also require samples from family members to interpret the results correctly. In these cases, the results may reveal parental relations that were previously unknown (e.g., non-biological paternity).

- *If applicable:* In the case of pharmacogenetic testing, bear in mind that it is designed for instructive, informational and research purposes based on the available scientific evidence. As such, its objective is to inform you about contributing factors for the treatment of certain disorders and diseases. The information that is obtained through pharmacogenetic testing is not intended to diagnose any disorders or diseases, and under no circumstances may it be used in place of specialist care provided by healthcare professionals.

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Results of the genetic test

You understand that the genetic test may return four different types of results:

- **Positive result:** One or more genetic variants (pathogenic or likely pathogenic) have been identified that are believed to be the cause of the suspected diagnosis or the clinical indication that led to the test.

- **Negative result:** No genetic variant with clinical implications has been identified. A negative result does not necessarily rule out the possibility of a genetic disorder or predisposition to a disease. Some genetic disorders have many different causes and it is not always possible to test for all of them. A negative result could also occur as a result of scientific, technical and/or knowledge-based limitations.

- **Non-conclusive result (unknown significance)**: One or more genetic variants have been identified but we do not know how relevant they are yet. These are known as "variants of unknown significance", or VUSes. These variants cannot be used as the basis for medical decisions. In certain cases, additional testing may be recommended for yourself or for other members of your family to reassess the clinical significance of these variants. The report will only include variants of unknown significance if they are considered clinically relevant by the clinical team.

- **Non-informative result:** No results have been obtained. This may occasionally happen as a result of a technical fault caused by an issue related to sample quality/quantity or sample contamination. In this case a new sample may be requested.

The identified genetic variants are classified into five different categories of pathogenicity, as per the ACMG standards (American College of Medical Genetics and Genomics; Richards et al. Genet Med 2015 17(5):405-424). Classification of genetic variants may change over time as a result of rapid advances in scientific knowledge and available clinical evidence, or due to changes in the patient's condition. For this reason, we may occasionally recommend a reanalysis of the data obtained after a certain time.

Whole Exome or Genome Sequencing (if applicable):

With exome and genome testing, as recommended by the ACMG, in addition to the primary purpose of the test, we may also analyze pathogenic (or possibly pathogenic) genetic variants from a list of genes associated with diseases that are considered clinically actionable, or in other words, diseases that can be prevented or treated. The genes or regions included in this list are associated with a predisposition to certain types of cancer, cardiovascular disease and metabolic disorders, among others. (https://www.ncbi.nlm.nih.gov/clinvar/docs/acmg/)

Findings in these genes are known as **secondary findings** and you will only be informed of them should you have explicitly stated in the informed consent form that you would like to be made aware

of them.
With genomic testing (e.g., genome, exome, or panels with a high number of genes), it is possible to detect **incidental findings (findings that were not requested)** that could have potential implications for your health but are not related to the symptoms of the disease or the clinical indication that led to the test. You will not be routinely informed of these incidental findings, but they may be provided to your doctor should they have

Limitations and risks of genetic testing

You acknowledge the following:

- If the genetic test is performed using a blood sample, the blood extraction method may cause temporary and minimal bleeding and pain around the puncture site, in addition to dizziness or fainting, and bruising may appear over the following days.
- Upon receiving the results of the genetic test, the patient (you) and/or other family members may suffer from psychological stress.
- There are various types of genetic disorders and no technique is capable of detecting all of them. Every technique has its own specific limitations, which will be duly indicated in the results report.
- In exceptional circumstances, the genetic test may return inaccurate results due to errors when taking, labeling, or processing the sample, or when analyzing/interpreting the data.
- The analysis and clinical interpretation of the genetic test is performed using the knowledge and technology that is currently available today. As the scientific knowledge expands, the analysis and interpretation of the test may change or be supplemented.
- The data may need to be reanalyzed and the patient (you) or your referring provider may be contacted again in the future should new findings be made related to the clinical indication behind the test.
- The data obtained in the test is not routinely reanalyzed by us or by the laboratory, but it is possible for certain genetic tests (e.g., clinical exome or genome testing) upon request, with additional fees required to issue a new results report.

Disclaimer:

Please note that genetic analyses are not definitive. Due to limitations in technology and/or incomplete medical knowledge, some disease-causing variants may not be detected. Therefore, it is not possible to completely exclude all risks for all possible genetic diseases.

Moreover, in some cases, the analysis may indicate a genetic abnormality when you or the Patient are/is actually unaffected (false positive) or may indicate no genetic abnormality when you or the

Patient are/is actually affected (false negative).

In cases where the underlying cause of a false-positive or false-negative finding could not be identified after investigation, the laboratory shall not be responsible for the incomplete, potentially misleading or incorrect result of an analysis.

Limitations of genome sequencing (If applicable):
Please note that this test cannot sequence a person's entire genome nor can it fully identify every single possible genetic condition. Genome sequencing will provide information about a wide array of genetic changes. However, you will not be informed about the majority of changes detected as there will likely not be enough information about their clinical relevance. You will only be informed about genetic changes that have the potential to trigger a condition related to your clinical condition. The laboratory cannot predict how serious the condition will be nor at what age symptoms may develop in the affected individual. It is also possible that the identified genetic differences may not explain the medical condition in question and they may not lead to any change in the current therapeutic and/or pharmacological treatment or approach.

Privacy, data protection, data storage (samples) and research

In accordance with the provisions of the European General Data Protection Regulation 2016/679 (GDPR), your sample and personal data will be shared with the testing laboratory to provide you with the requested service, which serves as the lawful basis to process said personal data. This personal data will also be stored and processed by the laboratory in a manner compliant with the GDPR.

This data may be used by the laboratory to respond to and follow up on any queries that you or the requesting party may have, and it may also be used for quality procedures or for contacting you in the future to update relevant clinical information. To complete the testing effectively, the laboratory may share said information with other designated centers, which also comply with the same legislation, in accordance with ethical considerations and current regulations.

Only duly authorized personnel from our organization and the laboratory will have access to your personal data. You may contact GeneLinx and the laboratory at any time to find out what data we have on you, correct said data if it is inaccurate, and request its erasure once our relationship has come to an end, should this be legally possible. You also have the right to request the transfer of your data to another entity (portability).

The genetic data obtained may be used by the laboratory for research purposes to expand scientific knowledge in scientific publications or genomic databases, based on the consent you provide below. These research activities provide new evidence for reclassifying variants, thus enabling more precise interpretation of results and enhancing diagnosis, prevention and treatment of genetic diseases.

Although the data shared with the scientific community is anonymous, the risk of identification cannot

be entirely excluded due to the unique nature of genetic information. However, the risk of this occurring is very low. It is also possible for someone to break into or gain unauthorized access to the system storing the data in the laboratory, although all appropriate measures shall be taken to minimize this risk. Also cannot other privacy risks that may not have yet been foreseen cannot be excluded. If you provide consent, you understand that you will not receive any financial benefit from research carried out or products developed by the laboratory.

Declaration of consent

By signing this form, I declare that I have received comprehensive information regarding the genetic background related to the disease in question, as well as the possibilities and limitations of genetic testing. I understand that I have the right to withdraw my consent for genetic analyses.

I have been informed, and agree, that my personal data and the data obtained in the analysis will be recorded, evaluated, or stored in a pseudoanonymized form in scientific databases, in accordance with data protection and medical confidentiality.

If the test report recipients (healthcare providers) are located in a so-called third country outside the European Economic Area (EEA), where the EU General Data Protection Regulations (GDPR) provisions do not apply, I consent to the transfer of my or the Patient's personal data to this third country and acknowledge that such third country may not provide a level of data protection equivalent to the GDPR and may grant fewer or less enforceable data protection rights, and there may be no independent data protection supervisory authority to assist in exercising these rights. This consent includes:

- providing the results of the Analysis and the raw data to the treating physician and/or the requesting laboratory.
- providing the results of the Analysis to the healthcare professionals who are involved in my or the Patient's medical counseling and/or clinical care.

I consent to the re-evaluation of my test results within the data storage period if needed (and applicable). If significant alterations become apparent, my ordering healthcare provider will be informed by e-mail.

I understand that this declaration of consent can be completely or partially withdrawn at any time.
I have had sufficient time to consider giving my consent

Optional consents:
I hereby give my consent to be informed about secondary findings (ACMG) - **only for WES/WGS**:
☐Yes ☐No

I consent to the storage of my genetic material for additional tests and/or quality control (for max. 10 years).
☐ Yes ☐ No

I consent to the storage of my test results beyond the time span of 10 years (or the legally required local time frame) by the laboratory.
☐ Yes ☐ No

I consent to the pseudo-anonymous storage and use of surplus genetic material and/or test results for scientific research and in scientific literature.
☐ Yes ☐ No

Signature of patient, mother, father, or legal guardian

By signing this document, I voluntarily authorize GeneLinx GmbH to order the indicated genetic test at the chosen laboratory. I have been adequately informed of the risks, benefits, and limitations of this genetic test.

In the case of a minor or a person without legal capacity, as mother/father/legal guardian, I authorize this genetic test. I confirm that the signatory is the sole legal guardian and that the other parent does not oppose this test being performed on our child.

Patient/Guardian Name:	
Patient/Guardian Signature:	
Date:	

Genetic Counsellor Name:	
Genetic Counsellor Signature:	
Date:	