

INFORMED CONSENT

WHOLE EXOME SEQUENCING PATIENT / FAMILY MEMBER

For more information on genetic testing for patients and family members, please visit: <https://blueprintgenetics.com/resources/whole-exome-sequencing-guide-for-patients-and-families/>

I confirm that the information below has been explained to me concerning the test:

1. The results of this test may show that I and/or my family members have an inherited disease or are at an increased risk to be affected by a genetic disease. I understand that this test may detect previously unrecognized biological relationships, such as non-paternity.
2. I am aware that the results of this test might be inconclusive about my genetic status. While some genetic variants are known to be disease causing and others are known to be benign, a portion of genetic variants found are of uncertain significance. Depending on the results of this test, my physician may recommend genetic counseling or further testing of myself and/or my family members.
3. I understand that an anonymized summary of results from this test may be presented for example at meetings, scientific publications and/or DNA variant databases in order to improve the understanding, diagnostics and treatment of similar clinical conditions. No identifying information will ever be presented.
4. If I have selected the patient insurance billing option, I authorize my health plan or insurance provider to pay my insurance benefits directly to Blueprint Genetics. I authorize Blueprint Genetics to release information concerning my testing to my insurer. I understand that I am legally responsible for sending Blueprint Genetics any money received from my insurance company for performance of this genetic test. If my insurance does not cover these services or only covers part of the amount, I am responsible for remaining costs of this test.
5. I am aware that not consenting to any of the sections to follow will not in any way affect my further treatment. If no box is checked in a section, it is assumed that no consent is given.
6. **Separate consent for sample storage at Blueprint Genetics for 3 years for the purposes of family member testing.** By checking the relevant box below I give my consent to the 3-year storage of the DNA sample in the diagnostic laboratory of Blueprint Genetics for the purposes of family member testing. Without this permission the sample will be stored approximately for 12 months and it is disposed of after that unless earlier disposal is required by applicable laws.

☐ I give my consent to the 3-year storage of the sample for family member testing.

7. **Separate consent for research use and long-term storage.** By checking the relevant box below I give my consent to the long-term storage of the DNA sample in the diagnostic laboratory of Blueprint Genetics (without separate consent for long-term storage the DNA samples are typically stored for approximately 12 months) for use of the DNA sample in research into hereditary Mendelian diseases and the efforts to improve the diagnostics and treatment of said diseases. The research data concerning me will be treated as confidential information and coded in such a way that my identity cannot be discovered without the key code in the possession of the Blueprint Genetics research physician. Where necessary, such coded research data may also be processed within or outside the European Union and released for use by another research group or a company participating in the study. I hereby give my consent to the use of the aforementioned research data for the purposes set out in this consent. The data will be preserved for 50 years.

I understand that my consent to the research use of the sample taken for diagnostic purposes is voluntary and that I may cancel this consent and withdraw my participation at any time prior to the completion of the study. I am aware that the data collected up to the date of my withdrawal will be used as part of the research material.

☐ I give my consent to the research use and long-term storage of the sample as set out in Section 7 above.

8. **Separate consent for reporting of secondary findings.** By checking the relevant box below I give Blueprint Genetics my consent to report to my ordering healthcare professional any possible secondary findings that are not directly related to the reason for ordering my test. Blueprint Genetics reports as secondary findings pathogenic and likely pathogenic variants in selected genes associated with various genetic disorders. The selected genes where secondary findings are reported represent those included in “ACMG Recommendations for Reporting of Secondary Findings in Clinical Exome and Genome Sequencing” published by the American College of Medical Genetics and Genomics.

I understand that secondary findings are of medical value and may have implications for my future health and for family planning purposes. I understand that the absence of secondary findings for any particular gene does not mean that there are no pathogenic variants in that gene.

Blueprint Genetics needs to receive this consent before sample is put into analysis in order to report any secondary findings. I understand that my family members can decide on their secondary findings independent of my decision.

☐ I give my consent to the reporting of secondary findings.

More information about how we process personal data: <https://blueprintgenetics.com/privacy/>

☐ I give Blueprint Genetics permission to contact me regarding further genetic research and/or other genetic services relevant to me in the future. I may withdraw from such contact at any time.

PATIENT SIGNATURE

By signing this form, I acknowledge that I have read the Informed Consent for Whole Exome Sequencing and understand its content. I have had the opportunity to ask questions about this form and my questions have been answered.	
Name (please print):	Date of birth (YYYY-MM-DD):
Signature:	Date (YYYY-MM-DD):
Name and relationship of Legal Representative, if patient is a minor (please print):	Signature of Legal Representative, if patient is a minor: