GENETIC TESTING PROGRAM INFORMED CONSENT
INHERITED RETINAL DEGENERATION (IRD)

Thank you for getting tested at Blueprint Genetics (we). This consent form includes essential information about your genetic test and is designed to help you to make important decisions impacting you and your family. Please go through this form carefully with your physician or genetic counselor.

This test is provided at no cost to you or your healthcare provider. The costs of this test are covered by Blueprint Genetics and the Foundation Fighting Blindness. We may share parts of your de-identified data, that is, data that does not include information that could identify you, with Scientific Collaborators, as explained on page 4.

Purpose

The goal of this test is to provide information that may be helpful to you and your physician to understand more about your retinal disease. It is also to provide information to Scientific Collaborators, who are working to understand more about IRD and how to improve the lives of affected people.

About this genetic test

The purpose of this test is to analyze selected genes associated with inherited retinal diseases (IRD). IRDs are a group of disorders affecting the retina of the eye. Common symptoms include night or color blindness, tunnel vision and subsequent progression to complete blindness. More information on IRD, this test and its exact target genes is available from your healthcare provider and can also be found at www.blueprintgenetics.com/sponsored-retinal-testing.

This test requires a blood or saliva sample from you. The sample will be processed by your healthcare provider and sent to Blueprint Genetics where it will be tested. Blueprint Genetics will generate a clinical report from the testing.

Our testing process produces more data about you than is provided in your clinical report. You have the right to access all genetic data we hold about you. More information about your rights can be found on page 3.

There is no-cost genetic counseling available for you after the test as part of the testing program. Please ask your healthcare provider for more information. If you do not understand what a genetic test is and wish to receive professional genetic counseling prior to signing this consent form, please let your healthcare provider know.

Please note that this test is only intended for patients who have not had a genetic test that meets any of the following criteria in the last three years: 1) A test that examined more than 32 genes relevant to inherited retinal disease, 2) A whole exome genetic test, or 3) A whole genome genetic test. If you are not sure whether you meet this criteria, please consult your healthcare provider.

Remember: Participating in the Genetic Testing Program is voluntary and you have the right to decide whether you want to take this test or not.

Meaning of the results

The results from your genetic test may be:

Diagnostic (positive), meaning that	Inconclusive, meaning that	Negative, meaning that
• We have detected a genetic change that may be connected to your retinal disease • You may have an increased risk of developing a genetic retinal disease in the future • You may be a carrier for inherited retinal disease • The results may have implications also for your family members	• We have detected a genetic change, but it is currently unknown whether the change is linked to your retinal disease • Advances in science and technology may later show the change to be harmless or disease-causing • Additional testing, including testing of your family members, may help to clarify the results	• We have not detected any retinal disease-causing genetic changes with the test performed • Additional testing may be beneficial for you

Our test reports are written for medical professionals and may contain complex language. Please ask your healthcare provider to explain the results. Depending on the results, you or your family members may be recommended to undertake further testing.

Future scientific discoveries may alter the meaning of your test result

While we always look at your genetic variants against the latest scientific evidence, the field of genetics research keeps evolving rapidly. With future scientific evidence, we might issue an updated analysis of the results to your healthcare provider unprompted, which may alter your diagnosis.

If your results are inconclusive, your healthcare provider may also ask us to reanalyze your data in a later point in time, in which situation the reanalysis may provide clarification for your test results. We encourage you to discuss with your physician beforehand whether you wish to be informed of any reanalyzed results.

The test may reveal incidental and undesired information about you and your family

There is a possibility that your test might reveal something that is not directly related to the reason for ordering your test and/or something you did not want to know. Such information might include, for example:

- Previously unknown biological relationships; for instance, you might be adopted
- Risk for developing additional, non-retinal symptoms that you are unaware of
- Difference in the number or rearrangement of sex chromosomes

This unexpected additional information may have significant psychological and/or social implications. If you have concerns about this risk, we encourage you to discuss this with your physician and family before taking the test. We may report such incidental information to your physician, if it is likely to impact your further testing or medical care.

Some genetic information can help predict future health problems of you and your family. A federal law, called the Genetic Information Nondiscrimination Act (GINA), generally makes it illegal for health insurance companies, group health plans and most employers to discriminate against you based on your genetic information. However, it does not protect you against discrimination by companies that sell life insurance, disability insurance or long-term care insurance.

Test limitations

The accuracy of genetic testing is less than 100% and is subject to some limitations. The test may not detect certain genetic alterations that are difficult to detect with our current testing methodologies. The test does not detect alterations located outside of the test's target genes. Also, some variation is inherent in the testing process caused by different factors, like the sample quality. For the full test limitations, please visit www.blueprintgenetics.com/sponsored-retinal-testing.

The test aims to find a genetic explanation for your IRD, and therefore, it is important that we receive sufficient clinical information to support the interpretation of your data. Relevant information includes, for example, symptoms, age of onset of the retinal disease, previous tests, and family history with respect to any similar retinal diseases. Too little clinical information may result in inconclusive test results.

In addition, the interpretation of the genetic causes of diseases may change in the future, as explained in section "Future scientific discoveries may alter the meaning of your test result" above.

Privacy and Data Protection

We process your personal data based on your consent. Your personal data has been provided to us by your healthcare facility. To participate in the Genetic Testing Program at Blueprint Genetics, you need to agree to the following:

- Your biological sample and relevant health information selected by your healthcare provider are transferred to Blueprint Genetics and the test results and your personal data may be processed and stored by Blueprint Genetics and your healthcare provider.
- An anonymized summary of your results may be presented, for example at meetings, scientific publications and/or databases in order to improve the understanding of retinal diseases. No information will be presented that can identify you.
- Blueprint Genetics may share your healthcare provider's name and contact information and your de-identified information with Scientific Collaborators, as explained below.
- Blueprint Genetics may use your sample and clinical information internally, to improve the understanding of genetics behind IRD, as explained below.

After your testing has been completed, we will send the test results to your healthcare provider and genetic counselor, through our secure online system. **Unless otherwise authorized by you, we will not share your personally identifiable information with anyone else.**

We have implemented a number of safeguards to secure your information. These include the masking and encryption of personal data, regular training and refreshing of our staff in HIPAA compliance handling personal information, physical safeguards, third-party audits and regularly testing our measures for ensuring the security of our data processing. In spite of these safeguards, there is a chance that someone unauthorized could gain access to your information.

You can exercise your rights under the Health Insurance Portability and Accountability Act of 1996 (HIPAA) and other applicable privacy laws by contacting us. We also apply the EU Regulation 2016/679, which gives you the rights to request access to, rectify or erase your personal data, to limit or oppose the processing of your personal data and the right to data portability. More information on our privacy practices and your rights can be found at www.blueprintgenetics.com/sponsored-retinal-testing.

Should you have any questions or concerns regarding privacy at Blueprint Genetics, you can contact us at privacy@blueprintgenetics.com.

How we share de-identified information with Scientific Collaborators

Scientific Collaborators are companies and organizations engaged in research and treatments for IRD. These Scientific Collaborators can be:

- Biopharmaceutical companies and their affiliates and partners
- Non-profit patient advocacies for IRD
- Medical research organizations for IRD

A list of current Scientific Collaborators can be found at: www.blueprintgenetics.com/sponsored-retinal-testing.

Information we may share with Scientific Collaborators

Information	Example
Your **healthcare provider's** name and contact info	John Smith, MD, Big City Medical Center, john.smith@work-email.com; 555-123-123
Your diagnosis	Retinitis pigmentosa
Your age range	20–29 years
Your sex	Male/female/other
Your genetic findings	- Gene(s) associated or potentially associated with your retinal disease - Genes that may affect the therapy or management of your retinal disease - Medically relevant variants (mutations) in those genes

We will **not** share your name, contact information, raw genetic information or any other identifiable information with any third parties without your separate prior written consent.

What will the shared information be used for?

Scientific Collaborators may contact your healthcare provider regarding the approved treatments for IRD, clinical trials and ethically approved studies.

In addition, Scientific Collaborators may use your de-identified information to guide the research and development of novel treatments for inherited retinal diseases. If you want to know more about the data sharing, please visit www.blueprintgenetics.com/sponsored-retinal-testing.

How Blueprint Genetics may use your sample and information internally

In addition to providing your clinical report, we may use your sample and information internally to improve the understanding of genetics behind retinal diseases. By signing this consent form you give us your permission to the long-term storage of your DNA sample in the diagnostic laboratory of Blueprint Genetics for the use of the DNA sample and the related information in the following:

- Scientific research to improve diagnostics and treatment of genetic IRD
- Research and development of diagnostic methods and products for IRD

The research data concerning you will be treated as confidential information and coded in such a way that your identity cannot be discovered without the key code in possession of the Blueprint Genetics research physician. Your sample and the related data will be preserved for a maximum of 50 years.

You may cancel your consent and withdraw your participation at any time. The data collected up to the date of your withdrawal will be used as described in this consent form, but will not be shared with Scientific Collaborators after your withdrawal. Your refusal to take part in or withdraw from the research project will not in any way affect your further treatment.

How Blueprint Genetics will not use your sample and information

- We will **not** sell your name or contact information.
- We will **not** sell your DNA, blood or saliva sample.
- We will **not** sell your clinical records.

SIGNATURE

Please sign below to confirm the following:

- You have read this form and understand and agree to its content
- You consent to the use of your sample and personal data as explained in this form
- You have had the opportunity to ask questions about this form, and your questions have been answered
- You will not seek reimbursement for this no-cost genetic test from any third party
- You have not had a genetic test within the last three years that meets any of the following criteria:

  1) A test that examined more than 32 inherited retinal disease relevant genes;
  2) A whole exome genetic test; or
  3) A whole genome genetic test.

Please make sure to fill in ALL fields. If you consent on behalf of your child or another person, please make sure to clearly indicate your relationship to the patient.

SIGNATURE*:
PATIENT NAME*:
PATIENT DATE OF BIRTH*:
IF NOT SIGNED BY THE PATIENT, SIGNATORY NAME AND RELATIONSHIP TO PATIENT*: ☐ PARENT/LEGAL GUARDIAN ☐ OTHER, PLEASE SPECIFY

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