

# AUTHORIZATION TO USE AND DISCLOSE HEALTH INFORMATION



I authorize the laboratory that has conducted or will conduct my genetic testing under the **RPE65 Genetic Test Program** and my physician to disclose to Spark Therapeutics and its affiliates, partners, collaborators, and others (collectively, “Spark”) the following:

- my name;
- contact information;
- date of birth;
- information regarding my condition and diagnoses and the results of my genetic testing (collectively, “My Information”) **so that Spark may use My Information for the purposes described in this form.**

I authorize Spark to use My Information for the following purposes:

- to contact me via mail, telephone, in electronic format or otherwise, to provide or offer information or services, including genetic counseling, that it believes to be of interest to me
- to help Spark develop programs and services that may be of interest to me or others with inherited retinal diseases (IRDs)
- to provide me with educational or marketing information about IRDs and their treatments.
- to contact my healthcare provider(s) about products and services that may be relevant for me, including contacting additional laboratories for further analysis

*Spark will not sell My Information or use or disclose My Information for unauthorized purposes.*

I understand that this Authorization is voluntary and that my ongoing medical care or eligibility for healthcare benefits will not be affected if I decline to sign this authorization form nor will it impact my ability to participate in Spark-sponsored programs in the future, but that I will not be able to participate in the **RPE65 Genetic Test Program** if I decide not to sign this authorization.

I understand that I may revoke this Authorization at any time in writing by sending a letter to Spark at the address listed on the following page. Revoking this Authorization will prevent Spark from further using My Information, but will not affect uses and disclosures of My Information that were already made in reliance on this Authorization.

# AUTHORIZATION TO USE AND DISCLOSE HEALTH INFORMATION



**To revoke this Authorization or to change your contact information, submit a written request to:**

Spark Therapeutics  
3737 Market Street  
Suite 1300  
Philadelphia, PA 19104  
Attn: Patient Services

I understand that once My Information has been disclosed, federal privacy laws may no longer apply or protect the information from further disclosure. Unless I expressly revoke this Authorization, it shall remain in effect for ten (10) years from the date I sign below. I may obtain a copy of this Authorization to keep for my records.

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**Signature of patient  
or parent/legal guardian of patient**  
(if under the age of 18)

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**Date**

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**Print name of patient  
or parent/legal guardian**

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**Relationship to patient**  
(if patient/legal guardian)

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# TERMS AND CONDITIONS



## THE *RPE65* GENETIC TEST PROGRAM TERMS AND CONDITIONS

The *RPE65* GENETIC TEST PROGRAM is a genetic testing program (“the Program”) that tests for the presence of *RPE65* gene mutations. The testing is supported by Spark Therapeutics, Inc. Your participation in the Program and use of Spark Therapeutics content and services is subject to the terms of the agreement between you and Spark Therapeutics set forth in these Program Terms and Conditions, which incorporate by reference the Spark Therapeutics general Terms of Use ([sparktx.com/terms-of-use](http://sparktx.com/terms-of-use)) and Privacy Policy ([sparktx.com/privacy-policy](http://sparktx.com/privacy-policy)). You may accept these Program Terms and Conditions by (1) clicking to accept or agree, where this option is made available to you, or (2) by signing this form at your physician’s office when you agree to participate in genetic testing and share your genetic test results with Spark Therapeutics. You may not participate in the Program if you do not accept these Program Terms and Conditions.

To be eligible to participate in the Program, you must (1) be a US resident at the time you are tested and receive your test results; (2) have a type of inherited retinal disease (IRD) that makes you eligible for the Program as determined by your healthcare professional; (3) have the approval of your healthcare professional to have the genetic test; and (4) authorize in writing that your healthcare professional and the genetic testing laboratory selected by Spark Therapeutics may test the genetic sample you provide and share your name, contact information and information regarding your condition, diagnoses, and results of your genetic testing (collectively, “Your Information”) with Spark Therapeutics. If you are under the age of 18, you must have the approval of your legal guardian to participate in the Program.

The genetic test provided under the Program requires you to provide a saliva or blood sample to your healthcare professional. Your sample will be analyzed by a genetic testing company selected by Spark Therapeutics, and the results will be provided to your healthcare professional and to Spark Therapeutics. The genetic testing company or companies that perform the test are independent from Spark Therapeutics and Spark Therapeutics has no control over or influence over how the test is conducted. Spark Therapeutics makes no warranty that the Program will meet your requirements, that it will be secure or error-free, that the results will be accurate or reliable, or that the quality of any of the services or information will meet your expectations. You understand and agree that by participating in the Program, Spark Therapeutics will process, use and disclose Your Information only as permitted by your written authorization and the Spark Therapeutics Privacy Policy.

The Program does not test for gene mutations other than *RPE65*, nor are the results of the testing performed in connection with the Program intended to be comprehensive. The results of the genetic test provided to your healthcare provider by the testing company may be: (1) positive (if the gene associated with the IRD has been identified by the test); (2) negative (if no genetic cause of the IRD has been identified by the test); or (3) inconclusive (if the test identified a genetic mutation, but it is unknown whether the identified mutation causes an IRD). You should consult with your own healthcare professionals about your diagnoses, genetic testing, and genetic

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testing results. Spark Therapeutics does not provide medical advice, and the results of the Program are not intended to be used by you for any diagnostic purpose or as a substitute for professional medical advice. Spark Therapeutics does not endorse, warranty, or guarantee the effectiveness of any specific course of action, resources, tests, physicians or other healthcare professionals, drugs, biologics, medical devices, products, procedures, opinions, or other information that may be offered to you or become available to you through the Program. Reliance on any information provided by Spark Therapeutics is solely at your own risk.

Through the Program, you may be offered an optional opportunity to discuss your genetic test results by telephone with a genetic counselor, in the case of certain types of results that may not be conclusive as to your *RPE65* mutation. If you choose this option, any advice provided by the counselor is independent of Spark.

If you choose to participate in the Program, you will not be responsible for the costs of the genetic test itself or the genetic counseling described in the previous paragraph. Please be aware, however, that you will be responsible for any other costs that may be incurred as a result of participating in the Program, including but not limited to the costs of visits or consultations with your healthcare professional in connection with the genetic test or the testing results.

By participating in the Program, you understand and agree that you acquire no rights in any research or commercial products that may be developed by Spark Therapeutics and/or its collaborating partners. You specifically understand that you will not receive compensation for any research or commercial products that include or result from Your Information.

**DISCLAIMER OF WARRANTIES.** You expressly acknowledge and agree that your participation in the Program is at your sole risk, and the Program is provided on an “as is” and “as available” basis. Spark Therapeutics expressly disclaims all warranties of any kind, whether express or implied, including but not limited to the implied warranties of merchantability, fitness for a particular purpose, and non-infringement.

**LIMITATION OF LIABILITY.** Spark Therapeutics does not control or endorse any actions resulting from your participation in the Program, and therefore, SPARK THERAPEUTICS SPECIFICALLY DISCLAIMS ANY LIABILITY WITH REGARD TO ANY ACTIONS RESULTING FROM YOUR PARTICIPATION IN THE SERVICES, TO THE EXTENT PERMITTED BY APPLICABLE LAW. YOU EXPRESSLY ACKNOWLEDGE AND AGREE THAT SPARK THERAPEUTICS SHALL NOT BE LIABLE FOR ANY DIRECT, INDIRECT, INCIDENTAL, SPECIAL, CONSEQUENTIAL, OR EXEMPLARY DAMAGES ARISING OUT OF OR RELATED TO YOUR PARTICIPATION IN THE SERVICES.

These Program Terms and Conditions, which incorporate by reference the Spark Therapeutics general Terms of Use and Privacy Policy, constitute the entire agreement between you and Spark Therapeutics and govern your participation in the Program.

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# Test Requisition Form – SP#047

(revised 11/22/2017)

- This form must accompany signed Spark Consent Form and patient specimen.
- Testing must be ordered by a qualified healthcare provider.

<b>Person completing form</b>	<b>Contact Information (phone or email)</b>	<b>Date of Request</b>
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Patient Information						
<b>Patient's Last (Family) Name</b>	<b>First Name</b>	<b>MI</b>	<b>Date of Birth:</b>	<b>Month</b>	<b>Day</b>	<b>Year</b>
<b>Street Address</b>			<b>City</b>	<b>State</b>	<b>Zip Code</b>	
<b>Phone Number</b>			<b>Email Address</b>			
<b>Patient ID Code (i.e., EMR #)</b>	<b>Date Collected:</b>	<b>Month</b>	<b>Day</b>	<b>Year</b>	<b>Reason for test</b> <input type="checkbox"/> Diagnosis <input type="checkbox"/> Presymptomatic/At risk	
<b>Sex</b> <input type="checkbox"/> Male <input type="checkbox"/> Female <input type="checkbox"/> Other	<b>GeoAncestry/Ethnicity</b>		<b>Specimen Source</b> <input type="checkbox"/> Whole blood <input type="checkbox"/> Saliva <input type="checkbox"/> Other:			
<b>Has patient been tested previously at PreventionGenetics?</b> <input type="checkbox"/> Yes <input type="checkbox"/> No <b>If yes, PG ID#:</b>	<b>Relevant Clinical Information (all that apply must be filled in please).</b> <b>Diagnosis:</b> <input type="checkbox"/> Retinitis Pigmentosa <input type="checkbox"/> Leber Congenital Amaurosis <input type="checkbox"/> Other:				<b>Signs/Symptoms:</b> <input type="checkbox"/> Nyctalopia <input type="checkbox"/> Nystagmus <input type="checkbox"/> Abnormal ERG <input type="checkbox"/> ERG Not Performed <input type="checkbox"/> Poor Dark Adaptation <input type="checkbox"/> Light Seeking Behavior <input type="checkbox"/> Decreased Visual Acuity <input type="checkbox"/> Other:	
			<b>Age of onset of disease:</b> <input type="checkbox"/> 0-10 years <input type="checkbox"/> 11-17 years <input type="checkbox"/> ≥ 18 years			

Test Selection		
<b>Test Code</b> #7371	<b>Test Name</b> Spark RPE65 sequencing & deletion/duplication	<b>**RPE65 testing is covered by the Spark Testing Initiative. Any requests for reflex testing to a broader vision panel will be discounted 10%, but will be the responsibility of the patient and/or institution (complete a separate Add On Test Requisition Form). For any questions or additional information about the Spark Testing Program, please visit <a href="http://www.luxturna.com">www.luxturna.com</a>.</b>

Provider Information			
<b>Institution</b>			
<b>Street Address</b>		<b>City</b>	<b>State</b> <b>Zip Code</b>
<b>Requesting Provider (First, Last, Degree)</b>		<b>NPI#</b>	
<b>Phone Number</b>		<b>Email</b>	

Genetic Counseling	
You acknowledge that if a patient's results are INDETERMINATE, Spark sponsors a program through which the patient may be offered genetic counseling.	
Test Reporting Instructions (Our preferred method of report transmission is via secure email.)	
Via Secure Email: <input type="checkbox"/> use email(s) above <input type="checkbox"/> send to the following email(s):	
<input type="checkbox"/> DO NOT email results. Instead, send via fax (provide fax #):	

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## Specimen & Shipping Instructions

### Specimen Requirements

**WHOLE BLOOD:** Collect 3-5 ml of whole blood in EDTA (purple top tube) or ACD (yellow top tube), minimum 1 ml for small infants.

**DNA:** For the NextGen panel, send in screw cap tube at least 5-10 µg of purified DNA at a concentration of at least 50 µg/ml (indicate concentration on tube label). For deletion/duplication testing via aCGH, send in screw cap tube at least 1 µg of purified DNA at a concentration of at least 100 µg/ml (indicate concentration on tube label). Generally, one tube of DNA is sufficient for both tests.

**SALIVA:** Oragene™ or Isohelix Saliva Collection kit used according to manufacturer instructions.

### Shipping/Handling Instructions

**Please label all specimen containers with the patient name, date of birth and/or ID number. At least two identifiers should be listed on specimen containers.** We accept specimen deliveries Monday-Saturday. Holiday schedules will be posted on our home page at least one week prior to major holidays.

**BLOOD:** Do not freeze. During hot weather, include a frozen ice pack in the shipping container. Place a paper towel or other thin material between the ice pack and the blood tube. In cold weather, include an unfrozen ice pack in the shipping container as insulation. At room temperature, blood specimen is stable for up to 48 hours. If refrigerated, blood specimen is stable for up to one week.

**DNA:** DNA may be shipped at room temperature. Label the tube with the composition of the solute, DNA concentration as well as the patient name, date of birth, and/or ID number. We only accept genomic DNA for testing. We do NOT accept products of whole genome amplification reactions or other amplification reactions.

Address	DNA Banking
Diagnostic Lab PreventionGenetics 3800 S. Business Park Ave. Marshfield, WI 54449, USA	DNA Banking has a reduced price of \$98 for patients if clinical testing is also being performed with us. For DNA Banking, see our DNA Banking Process and DNA Banking Forms. For questions related to DNA Banking, contact our DNA Banking Director at 715-387-0484, ext. 151 or email <a href="mailto:dnabanking@preventiongenetics.com">dnabanking@preventiongenetics.com</a> .

**DNA Genotyping Panel**

For quality control purposes, the PreventionGenetics DNA Genotyping Panel is performed on all clinical specimens. Genotyping results are **not** included in test reports.

**New York Specimen Collection**

If the patient's specimen is collected in New York, a New York State Non-Permitted Laboratory Test Request approval letter and Genetic Testing Healthcare Provider Statement must be included before testing will proceed.