INVITAE

ID YOUR IRD
INHERITED RETINAL DISEASE GENE TESTING INITIATIVE

ORDER ID For Invitae internal use only	**Requisition Form** ID YOUR ID® TRF948-2

This requisition form can be used to submit a specimen for the **ID YOUR IRD® program**, a sponsored testing program brought to you by **Spark Therapeutics** and **Invitae Corporation**. Please confirm that the patient meets the eligibility requirements for the program, including provision of written patient consent. Submit the sample according to the kit instructions with the below **Test Requisition Form (TRF, pages 1-3)** and **Patient Authorization forms (pages 4-7) completed and signed**.

INSTRUCTIONS: Review the ordering options and then complete all sections of this form. Your ordering option will be indicated in the test selection section.

ORDERING OPTIONS

1. ID YOUR IRD® PROGRAM

For individuals that meet the eligibility criteria below and wish to receive the program specific genetic testing panels.

REQUIRED: You must select the appropriate eligibility criteria for this patient below.

This program is available to patients suspected of having an inherited retinal disease AND who have experienced one or more of the following (select all that apply, patients must have at least one to qualify):
Note: This program does not test for genes associated with age-related macular degeneration or ocular/oculocutaneous albinism.

- ◯ Nyctalopia
- ◯ Peripheral field loss
- ◯ Central vision loss
- ◯ Deterioration of color vision
- ◯ Photophobia
- ◯ Clinical or suspected diagnosis of an inherited retinal disease

2. GENE-SPECIFIC FAMILY FOLLOW-UP TESTING

For relatives of program participants who received a Pathogenic/Likely Pathogenic result or approved VUS who want to receive gene specific family follow-up testing at no additional cost. Relatives do not need to meet the eligibility criteria listed above. Learn more at **www.invitae.com/family**.

PATIENT INFORMATION

First name	MI	Last name

Date of birth (MM/DD/YYYY)	Biological sex ◯ M ◯ F	MRN (medical record number)

Ancestry ◯ Asian ◯ Black/African American ◯ White/Caucasian ◯ Ashkenazi Jewish ◯ Hispanic ◯ Native American ◯ Pacific Islander ◯ French Canadian ◯ Sephardic Jewish ◯ Mediterranean ◯ Other: _____

Phone	Email address (report access after clinician releases)

Address	City

State/Prov	ZIP/Postal code	Country

Ship a saliva kit to this patient (to submit, fax this form and Patient Authorization form to Client Services at 415-276-4164)

◯ Ship kit to address above

◯ Ship kit to alternate address: _____

SPECIMEN INFORMATION

Specimen type: Blood (3-mL purple EDTA) -OR- Saliva (Oragene™) -OR- Assisted Saliva

We are unable to accept blood/saliva from patients with:
- *Allogeneic bone marrow transplants*
- *Blood transfusion <2 weeks prior to specimen collection*

Specimen collection date (MM/DD/YYYY): ____ / ____ / ____
If not provided, the day before specimen receipt will be used

Special cases: ◯ History of/current hematologic malignancy in patient

INVITAE PARTNER CODE	SPARK

CLINICIAN INFORMATION

Organization name	

Phone	Fax

Address	City

State/Prov	ZIP/Postal code	Country

Primary clinical contact name (if different from ordering provider)	NPI

Primary clinical contact email address (for report access)

Ordering provider (select one ordering provider by marking the checkbox before the name)

	Name	NPI	Email address (for report access)
◯			
◯			
◯			
◯			
◯			

Additional clinical or laboratory contacts (optional, to share access to order online)

◯ **Share this order with the primary clinical contact's default clinical team, manage at invitae.com**

Name	Email address (for report access)

Name	Email address (for report access)

CLINICAL HISTORY

FAMILY HISTORY

Is there a family history of disease for which the patient is being tested? ◯ Yes ◯ No If yes, describe below and attach pedigree and/or clinical notes.

Relative's relationship to this patient	Maternal or paternal	Diagnosed condition	Age at diagnosis	Relative's relationship to this patient	Maternal or paternal	Diagnosed condition	Age at diagnosis

PERSONAL HISTORY

Is/was this patient affected or symptomatic?† ◯ Yes ◯ No
Provide details in the required clinical history questions (if applicable).

† Symptomatic means this patient has features or signs known or suspected to be related to the genetic testing being ordered and could include findings on physical examination, laboratory tests, or imaging.

REQUIRED CLINICAL HISTORY

Age of onset: _____

Clinical diagnosis, if known:

Ocular features	Y	N	UNKNOWN
Bone spicules/pigment clumping	◯	◯	◯
Optic nerve atrophy/optic disc pallor	◯	◯	◯
Abnormal fundus appearance	◯	◯	◯
Abnormal ERG results	◯	◯	◯
Is disease progressive in this individual?	◯	◯	◯

Other ocular features and/or detailed test results (e.g., ERG):

Extraocular features	Y	N	UNKNOWN
Developmental delay	◯	◯	◯
Intellectual disability	◯	◯	◯
Sensorineural hearing loss	◯	◯	◯
Skeletal abnormalities	◯	◯	◯
Renal disease	◯	◯	◯
Genital abnormalities	◯	◯	◯
Ataxia	◯	◯	◯
Organ laterality defect (e.g., situs inversus)	◯	◯	◯
Molar tooth sign/vermian hypoplasia	◯	◯	◯

Other extraocular features:

Y = test performed and/or medical history taken AND material finding reported; N = test performed and/or medical history taken AND no material finding; Unknown = not in medical record and/or test not performed

OPTIONAL - REQUESTED VARIANTS FOR THIS PATIENT'S REPORT, IF KNOWN

*To have the presence or absence of specific variants commented on in this patient's report, provide the details below. For gene-specific family follow-up see **Note** under Test Selection.*

Was the proband (individual with variant) tested at Invitae? ◯ Yes, Invitae Order ID: RQ# _____ ◯ No: Attach copy of lab results (*required*)

Variant(s) *(e.g. GENE c.2200A>T (p.Thr734Ser) NM_00012345) If left blank, all variants identified in the proband will be commented on.*		**This patient's relationship to proband:**
		◯ Parent ◯ Sibling ◯ Grandchild
		◯ Child ◯ Self ◯ Other: _____

TEST SELECTION – Select option 1 or 2 below:

◯ 1. ID YOUR IRD® PROGRAM – Indicate test(s) to be performed below:

Test code	Test name	# of genes	Gene list
◯ 72100	Invitae Inherited Retinal Disease Panel	248	ABCA4, ABHD12, ACBD5, ACO2, ADAM9, ADAMTS18, ADGRA3, ADGRV1, ADIPOR1, AGBL5, AHI1, AHR, AIPL1, ARHGEF18, ARL13B, ARL2BP, ARL3, ARL6, ARMC9, ARSG, ASRGL1, ATF6, ATOH7, BBIP1, BBS1, BBS2, BBS4, BBS5, BBS7, BBS9, BBS10, BBS12, BEST1, C1QTNF5, C8orf37, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CCT2, CDH3, CDH23, CDHR1, CEP19, CEP41, CEP78, CEP83, CEP164, CEP250, CEP290, CERKL, CFAP410, CHM, CIB2, CLCC1, CLN3, CLRN1, CLUAP1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL2A1, COL9A1, CRB1, CRX, CSPP1, CTNNA1, CWC27, CYP4V2, DHDDS, DHX38, DRAM2, DTHD1, EFEMP1, ELOVL4, EMC1, EXOSC2, EYS, FAM161A, FLVCR1, FRMD7, FSCN2, FZD4, GDF6, GNAT1, GNAT2, GNB3, GNPTG, GRM6, GUCA1A, GUCA1B, GUCY2D, HARS, HGSNAT, HK1, HMX1, IDH3A, IDH3B, IFT27, IFT43, IFT80, IFT81, IFT140, IFT172, IMPDH1, IMPG1, IMPG2, INPP5E, INVS, IQCB1, JAG1, KCNJ13, KCNV2, KIAA1549, KIF11, KIZ, KLHL7, LCA5, LRAT, LRIT3, LRP5, LZTFL1, MAK, MAPKAPK3, MERTK, MFRP, MKKS, MKS1, MTTP, MYO7A, NDP, NEUROD1, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NRL, NYX, OAT, OPN1SW, OTX2, P3H2, PCARE, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PDZD7, PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26, PHYH, PITPNM3, PLA2G5, PLK4, PNPLA6, POC1B, POMGNT1, PRCD, PRDM13, PROM1, PRPF3, PRPF4, PRPF6, PRPF8, PRPH2, RAB28, RAX2, RBP3, RBP4, RCBTB1, RD3, RDH5, RDH11, RDH12, REEP6, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROM1, RP1, RP2, RPE65, RPGRIP1, RPGRIP1L, RS1, RTN4IP1, SAG, SAMD11, SDCCAG8, SEMA4A, SLC24A1, SLC7A14, SNRNP200, SPATA7, SPP2, TEAD1, TIMP3, TMEM126A, TMEM216, TMEM237, TOPORS, TPP1 (CLN2), TRAF3IP1, TRIM32, TRNT1, TRPM1, TSPAN12, TTC8, TTLL5, TUB, TUBGCP4, TUBGCP6, TULP1, UNC119, USH1C, USH1G, USH2A, VCAN, VPS13B, WDPCP, WDR19, WHRN, ZNF408, ZNF423, ZNF513

◯ 2. GENE-SPECIFIC FAMILY FOLLOW-UP TESTING *For relatives of a program participant ('proband') who received a Pathogenic/Likely Pathogenic result or approved VUS.*

Proband's Invitae Order ID:	This patient's relationship to proband:	Gene(s) to be tested in this patient:
RQ# _____	◯ Parent ◯ Sibling ◯ Grandchild ◯ Child ◯ Other: _____	

***NOTE**: The presence or absence of all variants identified in the proband for the gene(s) ordered for gene-specific family follow-up will be commented on in this patient's report unless a limited selection is specified in the **Requested Variants** section above. Invitae will report any Pathogenic/Likely Pathogenic variants found in this patient for the gene(s) ordered.*

Invitae continually updates its panels based on the most recent evidence. If an order is placed using an outdated test requisition form, Invitae reserves the right to upgrade ordered tests to their current versions. Test IDs containing add-on codes will include the original panel as well as the add-on.

By signing this form, the medical professional acknowledges that the individual/family member authorized to make decisions for the individual (collectively, the "Patient") has been supplied information regarding and consented to undergo genetic testing, substantially as set forth in Invitae's Informed Consent for Genetic Testing (www.invitae.com/forms). In connection with the Program the Patient has been informed that Invitae may notify them of clinical updates related to genetic test results (in consultation with the ordering medical professional as indicated) and has been informed that deidentified Patient data may be used and shared with third parties, for research and commercial purposes and, in the U.S., to contact their medical professional. The medical professional warrants that he/she will not seek reimbursement for this no-charge test from any third party, including but not limited to federal healthcare programs. The medical professional also hereby acknowledges that organization and clinician contact information provided in the order may be shared with third parties, including commercial organizations, that may contact the medical professional directly in connection with the Program, and that they have made the Patient aware that de-identified Patient data may be used and shared with such third parties, for purposes which include contacting their medical professional directly in connection with the Program. A list of third party partners may be provided upon request. I attest that I am authorized under applicable state law to order this test.

Medical professional signature (required)	Date (MM/DD/YYYY)

AUTHORIZATION TO USE AND DISCLOSE HEALTH INFORMATION

I authorize the laboratory that has conducted or will conduct my genetic testing under the ID YOUR IRD 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 help Spark support the broad Inherited Retinal Disease (IRD) community by developing commercial programs and services that may be of interest to me or others with IRDs
- to contact me via mail, telephone, in electronic format or otherwise, to provide or offer information or services, including genetic counseling, that Spark believes to be of interest to me
- to provide me with educational or marketing information about IRDs and disease management
- 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 **ID YOUR IRD 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 from the date I sign below. I may obtain a copy of this Authorization to keep for my records.

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

Date

Print name of patient or parent/legal guardian

Relationship to patient (if patient/legal guardian)

TERMS AND CONDITIONS

THE ID YOUR IRD PROGRAM TERMS AND CONDITIONS

The ID YOUR IRD program is a genetic testing program ("the Program") that tests for mutations in approximately 250 genes suspected to be associated with inherited retinal diseases. 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) and Privacy Policy (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) suspected of having an Inherited retinal disease (IRD); (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.

Testing results performed in connection with the Program are not intended to be comprehensive. The Program tests only for gene mutations related to IRD. 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) uncertain (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 TESTING RESULTS. SPARK THERAPEUTICS DOES NOT PROVIDE MEDICAL

TERMS AND CONDITIONS

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 will be offered an optional opportunity to discuss your genetic test results by telephone with a genetic counselor. 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 RIGHT OR INTERESTS IN ANY INVESTIGATIONAL OR COMMERCIAL PRODUCTS THAT MAY BE DEVELOPED BY SPARK THERAPEUTICS AND/OR ITS COLLABORATING PARTNERS. No purchase is necessary to participate in the ID YOUR IRD program.

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.