INVITAE

ORDER ID
For Invitae internal use only

Requisition Form
ID YOUR IRD TRF948-1

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. To submit orders for genetic testing outside of this program, please order through Invitae's online portal or use a standard requisition form, accessible at www.invitae.com/order-forms.

PROGRAM ELIGIBILITY:

Patients suspected of having an inherited retinal disease AND who have experienced one or more of the following: peripheral field loss, nyctalopia, deterioration in color vision, central vision loss, or photophobia. ID YOUR IRD is not appropriate for patients with age-related macular degeneration or ocular/oculocutaneous albinism.

PATIENT INFORMATION

First name	MI	Last name
Date of birth (MM/DD/YYYY)	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

Address	City

State	ZIP code	Country

SPECIMEN INFORMATION

Label each tube with the patient's full name, date of birth, and specimen collection date.
A requisition form MUST accompany each specimen. www.invitae.com/specimen-requirements

Specimen type : ◯ Blood ◯ Saliva ◯ Assisted saliva

We are unable to accept blood/saliva from patients with:

- *Allogeneic bone marrow transplants*
- *Blood transfusion <2 weeks prior to specimen collection*

Collection date (MM/DD/YYYY) *If not provided, date will be 1 day prior to our receipt of specimen.*

Special cases : ◯ History of/current hematologic malignancy ◯ Resubmission

REASON FOR TESTING

Previous results *(if applicable and not included in clinical criteria below)*

ORGANIZATION INFORMATION

Organization name and address

Organization name	
Phone	Fax

Address	City

State	ZIP code	Country

Primary clinical contact

Name	Role/title
Phone	NPI

Email address (for report access)

Ordering physician

◯ **Same as primary clinical contact**

Name	NPI

Email address (for report access)

Additional clinical or laboratory contact (optional)

Name	Email address (for report access)

INVITAE PARTNER CODE	SPARK

FAMILY VARIANT TESTING

Invitae's family variant testing programs involves full analysis of the gene in which the original family member's variant was identified. For more information, visit www.invitae.com/family-testing.

Please attach the proband's clinical report or provide Invitae RQ#

INVITAE PROBAND RQ#	RELATIONSHIP TO PROBAND	GENE(S)	VARIANT(S)

ID YOUR IRD PROGRAM ELIGIBILITY/CLINICAL INFORMATION

Note: This test is not appropriate for patients with age-related macular degeneration or ocular/oculocutaneous albinism.

Required patient information

Eligibility criteria
Select all that apply. Patient must have at least one to qualify.

- ☐ Nyctalopia
- ☐ Peripheral field loss
- ☐ Central vision loss Age of onset: _____
- ☐ Deterioration of color vision
- ☐ Photophobia

Clinical diagnosis, if known:

_____

_____

To request a complimentary specimen collection kit visit
www.invitae.com/request-a-kit

SHIPPING INSTRUCTIONS
Please ship specimen to Invitae:

Attn: Invitae Client Services
1400 16th Street
San Francisco, CA 94103

Additional information (for variant classification)

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

Additional 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?	☐	☐	☐
Is there family history of retinal disease?	☐	☐	☐

If yes, please specify relationship(s) and diagnosis:

_____

_____

_____

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:

_____

_____

ASSAY

Invitae is a CAP-accredited and CLIA-certified clinical diagnostic laboratory performing full-gene sequencing and deletion/duplication analysis using next-generation sequencing technology (NGS). Search for details on the analysis of any gene in our test catalog at **www.invitae.com/physician/search**.

Invitae continually updates its panels based on the most recent evidence. Please note that if an order is placed using an older version of this form, Invitae reserves the right to upgrade any ordered panel(s) to the current version(s).

TESTS INCLUDED IN THE PROGRAM

INVITAE INHERITED RETINAL DISEASE PANEL

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, ARL2BP, ARL13B, ARL2BP, ARL3, ARL6, ARMC9, ARSG, ASRGL1, ATF6, ATOH7, BBIP1, BBS1, BBS2, BBS4, BBS5, BBS7, BBS9, BBS10, BBS12, BEST1, C1QTNF5, C2orf71, C8orf37, C21orf2, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CCT2, CDH3, CDH23, CDHR1, CEP19, CEP41, CEP78, CEP83, CEP164, CEP250, CEP290, CERKL, 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, 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*

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/patient-consent) and in connection with the ID YOUR IRD® program, and 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). The medical professional warrants that he/she will not seek reimbursement for this no-cost 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 Spark Therapeutics, that may contact the medical professional directly in connection with the ID YOUR IRD program, and that they have made the Patient aware that third parties, including Spark Therapeutics, may contact their medical professional regarding de-identified information gathered through the program. The medical professional acknowledges that Spark Therapeutics sponsors a program through which the Patient may be offered optional genetic counseling through third parties. In addition to the above, I attest that I am the ordering physician, or I am authorized by the ordering physician to order this test, or I am authorized under applicable state law to order this test.

Medical professional signature	Date

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) 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 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.