

# UNIVERSAL RARE GENE STUDY

## UNI-RARE

A Registry and Natural History  
Study of Retinal Dystrophies  
Associated with Rare Disease-  
Causing Genetic Variants

## ENROLLMENT IS OPEN!

IDENTIFY MORE  
PEOPLE FOR CLINICAL  
TRIALS

**INFORM DESIGN OF  
CLINICAL TRIALS**

IDENTIFY  
THERAPEUTIC TARGETS  
FOR MORE PEOPLE

*Sponsored by  
the Foundation Fighting Blindness,  
Uni-Rare is a new study of ~1,500 people  
with one of 300+ rare genes associated with  
inherited retinal diseases (IRDs).*

## INTERESTED IN ENROLLING?



participating clinical sites  
can be found on  
[clinicaltrials.gov](https://clinicaltrials.gov) | UNI-RARE



contact the site nearest to  
you and a site member will  
review the availability and  
details of this study with  
you

## WHAT IS INVOLVED?

Uni-Rare has two phases:

### 1- The Registry Phase

- open to rare IRD genes
- one single visit (regular checkup) to evaluate genetic data and collect routine clinical measures

### 2- The Natural History Study (NHS) Phase

- opens up gene-by-gene
- ask site nearest to you which genes are currently open
- annual visits over 4 years

## WHO CAN ENROLL?

Study participants must:

- be at least 4 years old
- have a clinical diagnosis of retinal disease
- have a rare IRD gene with at least 1 mutation that is the cause of the retinal disease



# Usher syndrome type 1B (MYO7A)

UNI-RARE

**Uni-Rare** is a natural history study of vision loss progression. It is a critical and necessary step in supporting the approval of upcoming clinical trials and future therapies.

## ENROLLING USH1B PATIENTS NOW!

### COMMON QUESTIONS

- How long does the entire natural history study last? ***It's over a 4 year period***
- How many visits will I need to attend annually? ***Only one visit annually (4 total, plus one registry visit)***
- How long is a single visit? ***Plan for a full day, about 4-5 hours***
- Does it cost anything? ***The initial registry visit will be billed to your health insurance as a "standard care visit". If you do not have health insurance, you will need to pay out-of-pocket. However, the 4 annual visits per year are paid for by the study (no cost to you or your health insurance).***
- Do I need health insurance? ***No, health insurance is not required.***
- What is the closest location to me? ***You can check clinic locations on [clinicaltrials.gov](https://clinicaltrials.gov) - search for UNI-RARE. The first two locations currently enrolling are Retina Foundation of the Southwest (Dallas, TX) and Mass Eye and Ear (Boston, MA). Other locations around the US will begin enrolling soon.***
- Will costs associated with travel, if traveling from afar be covered? ***Yes, costs associated with travel from afar will be covered.***
- What type of tests will they be performing during visits? ***All non-invasive tests, including: imaging, tests of retinal sensitivity and visual acuity.***
- Should someone come with me? ***It's not necessary, but you will need someone to drive you home.***

### HAVE MORE QUESTIONS?

Ask your retinal specialist, or contact:

**Uni-Rare Coordinating Center**

**813-975-8690**

**Email: [ffb@jaeb.org](mailto:ffb@jaeb.org)**

# Uni-Rare Participating Locations and Contacts

## US LOCATIONS

### Phoenix, Arizona United States, 85020

Associated Retina Consultants  
Contact: Benjamin Bakall, MD, PhD  
Contact: Gabrielle Santor, 602-242-4928  
gabrielle.santor@doctrials.com

### Little Rock, Arkansas, United States 72205

University of Arkansas, Jones Eye Institute  
Contact: Sami Uwaydat, MD  
Contact: Sajida Chauhan

### La Jolla, California, United States 92093

Univ. of California San Diego, Jacobs Retina Center  
Contact: Shyamanga Borooah, MD  
sborooah@healthucsd.edu  
858-822-3179

### San Francisco, California, United States 94158

(UCSF) University of California San Francisco  
Contact: Jacque Duncan, MD  
Contact: Stephanie Duret - 415-353-2800  
stephanie.duret@ucsf.edu

### Denver, Colorado United States 80207

Colorado Retina Associates  
Contact: Alan Kimura, MD, MPH  
Contact Sara Shaw - sshaw@retinacolorado.com  
303-261-1600

### Jacksonville, Florida, United States, 32209

University of Florida Health Jacksonville  
Contact: Sandeep Grover, MD  
Contact: Ghulam Hamdani - 904-244-9305  
ghulam.hamdani@jax.ufl.edu

### Miami, Florida, United States 33136

University of Miami, Bascom Palmer Eye Inst.  
Contact: Byron Lam, MD  
Contact: Miguel Correa - 305-326-6021  
mac681@med.miami.edu

### Atlanta, Georgia, United States, 30322

Emory University, Emory Eye Center  
Contact: Nieraj Jain, MD  
Contact: Olga Ostretsova  
olga.ostretsova@emory.edu - 404-778-2020

### Baltimore, Maryland, United States 21236

Johns Hopkins University, Wilmer Eye Intst.  
Contact: Mandeep S Singh, MD, PhD  
Contact: Sierra Thomas - sthom142@jhmi.edu  
443-287-8343

### Boston, Mass. United States, 02114

Harvard Univ. Mass Eye and Ear Infirmary  
Contact: Rachel Huckfeldt, MD, PhD  
Contact: Emma Nguyen - 617-573-3621  
my\_nguyen@meei.harvard.edu

### Ann Arbor, Michigan, United States 48105

University of Michigan, Kellogg Eye Center  
Contact: Abigail Fahim MD, PhD  
Contact: Callie Gordon - 734-232-8080  
callieg@med.umich.edu

### Rochester, Minnesota, United States 27705

Mayo Clinic  
Contact: Brittni Scruggs, MD, PhD  
Contact: Suzanne Wernimont - 507-538-8119  
werimont.suzanne@mayo.edu

# Uni-Rare Participating Locations and Contacts (cont)

## US LOCATIONS (CONT)

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### **Durham, North Carolina, United States, 27705**

Duke University, Duke Eye Center

Contact: Ramiro Maldonado, MD

Contact: Rebecca Schmidt - 901-737-1915

rebecca.schmidt@duke.edu

### **Portland, Oregon, United States, 97239**

OHSU, Casey Eye Institute

Contact: Mark Pennesi, MD, PhD

Contact: Lillian Wheary - wheary@ohsu.edu

503-494-8386

### **Pittsburgh, Pennsylvania, United States, 15213**

UPMC Eye Center

Contact: Boris Rosin, MD, PhD

Contact: Michelle Alabek - alabekm@upmc.edu

412-647-3343

### **Dallas, Texas, United States, 75231**

Retina Foundation of the Southwest

Contact: David Birch, PhD

Contact: Kaylie Jones - 214-363-3911

kwebb@retinafoundation.org

### **Houston, Texas, United States, 77030**

Baylor College of Medicine, Alkek Eye Center

Contact: John T. Stout, MD, PhD

Contact: Moises Sanchez Guzman - 713-798-8419

moises.sanchezguzman@bcm.edu

### **Salt Lake City, Utah, United States, 84132**

University of Utah, John Moran Eye Center

Contact: Paul Bernstein, MD, PhD

Contact: Susan Allman - Susan.Allman@hsc.utah.edu

801-581-4069

### **Madison Wisconsin, United States 53711**

University of Wisconsin Madison

Contact: Kimberly Stepien, MD

Contact: Nickie Stangel - nstangel@wisc.edu

608-263-8783

### **Milwaukee, Wisconsin, United States, 53226**

Medical College of Wisconsin Eye Inst.

Contact: Thomas Connor, MD

Contact: Katie McKenney - kmckenney@mcw.edu

414-955-7840

## INTERNATIONAL

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### **Belo Horizonte, Rio De Janeiro, Brazil**

Neuro e Retina LTDA

Contact: Fernanda Belga Ottoni Porto, MD, PhD

Contact: Mariana Amoroso - 55-31-987-932809

marianaAmorosoINRET@gmail.com

### **Sao Paulo, Sao Paulo Province, Brazil**

Instituto de Genetica Ocular

Contact: Juliana Sallum, MD, PhD

Contact: Caio Marques - 55-11-557-64848

pesquisa@instituodegeneticaocular.com.br

### **Toronto, Ontario, Canada, M5G 2L3**

University of Toronto, Hospital for Sick Children

Contact: Elise Heon, MD

Contact: Sabahat Javaid - 1-416-813-7654

sabahat.javaid@sickkids.ca

### **Helsinki, Finland, 00100**

Helsinki University Hospital

Contact: Eeva-Marja Sankila, MD

Contact: Sofia Paleologos - 358405703681

sofia.paleologos@hus.fi

# *Uni-Rare Participating Locations and Contacts (cont)*

## **INTERNATIONAL (CONT)**

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### **Paris, France, 75012**

CHNO des Quinze-Vingts

Contact: Isabell Audo, MD - [isabelle.audo@inserm.fr](mailto:isabelle.audo@inserm.fr)  
331-400-21430

### **Jerusalem, Israel, 9112002**

Hadassah-Hebrew University Medical Center

Contact: Eyal Banin, MD

Contact: Devora Marks Ohana - 972-2-6776585  
[devoramarks@gmail.com](mailto:devoramarks@gmail.com)

### **Yucatan, Merida, Mexico**

Retina and Genomics Institute

Contact: Adda Villanueva, MD  
52-999-223-3623

### **Nijmegen, Netherlands, 6525 GA**

Radboud University Medical Center

Contact: Suzanna Yzer, MD

Contact: Jack Weeda - 31-(0)24-3613212  
[jack.weeda@radboudumc.nl](mailto:jack.weeda@radboudumc.nl)

### **Basel, Basel-Stadt, Switzerland**

University Hospital Basel

Contact: Henrik Scholl, MD, PhD

Contact: Daniela Hauenstein - 41-61-265-8686  
[daniela.hauenstein@usb.ch](mailto:daniela.hauenstein@usb.ch)

### **London, United Kingdom, EC1V 2PD**

Moorfields Eye Hospital

Contact: Michel Michaelides, MD

Contact: Matilda Fenn - [m.fenn1@nhs.net](mailto:m.fenn1@nhs.net)  
02072533411

## **HAVE QUESTIONS?**

Ask your retinal specialist, or contact:

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