Texas School for the Blind and Visually Impaired



Outreach Programs

[www.tsbvi.edu](http://www.tsbvi.edu) | 512.454.8631 | 1100 W. 45th St. | Austin, TX 78756

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# **Genetic Testing and Emerging Gene Therapies for People**

# **with Inherited Retinal Diseases**

# **Foundation Fighting Blindness**

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# **Slide 2 About the Foundation**

# Largest private funder of retinal degenerative disease research in the world – raised $816 million.

# Research funded: lab, translational, clinical trials, career development awards, natural history, registry, genetic testing.

# Entire spectrum of RDDs including: retinitis pigmentosa, Stargardt disease, Usher syndrome, Leber congenital amaurosis, dry AMD.

# **Slide 3 Resources**

# Website: www.fightingblindness.org

# Patient Support: 1-888-394-3937 or Email: info@fightingblindness.org

# Chapters: 40+ volunteer-led chapters across the U.S.

# Research Updates: Chapter Webinars, Clinical Trials May

# Events: VisionWalk, Hope from Home, Dining in the Dark

# Continuing Education Courses: April 5th

# My Retina Tracker Genetic Testing Program

# **Slide 4 Why Genetic Testing for IRD Patients?**

# Can confirm or change clinical diagnosis.

# InformedDNA study showed that clinical diagnoses for IRD patients changed in 13% of cases after genetic testing.

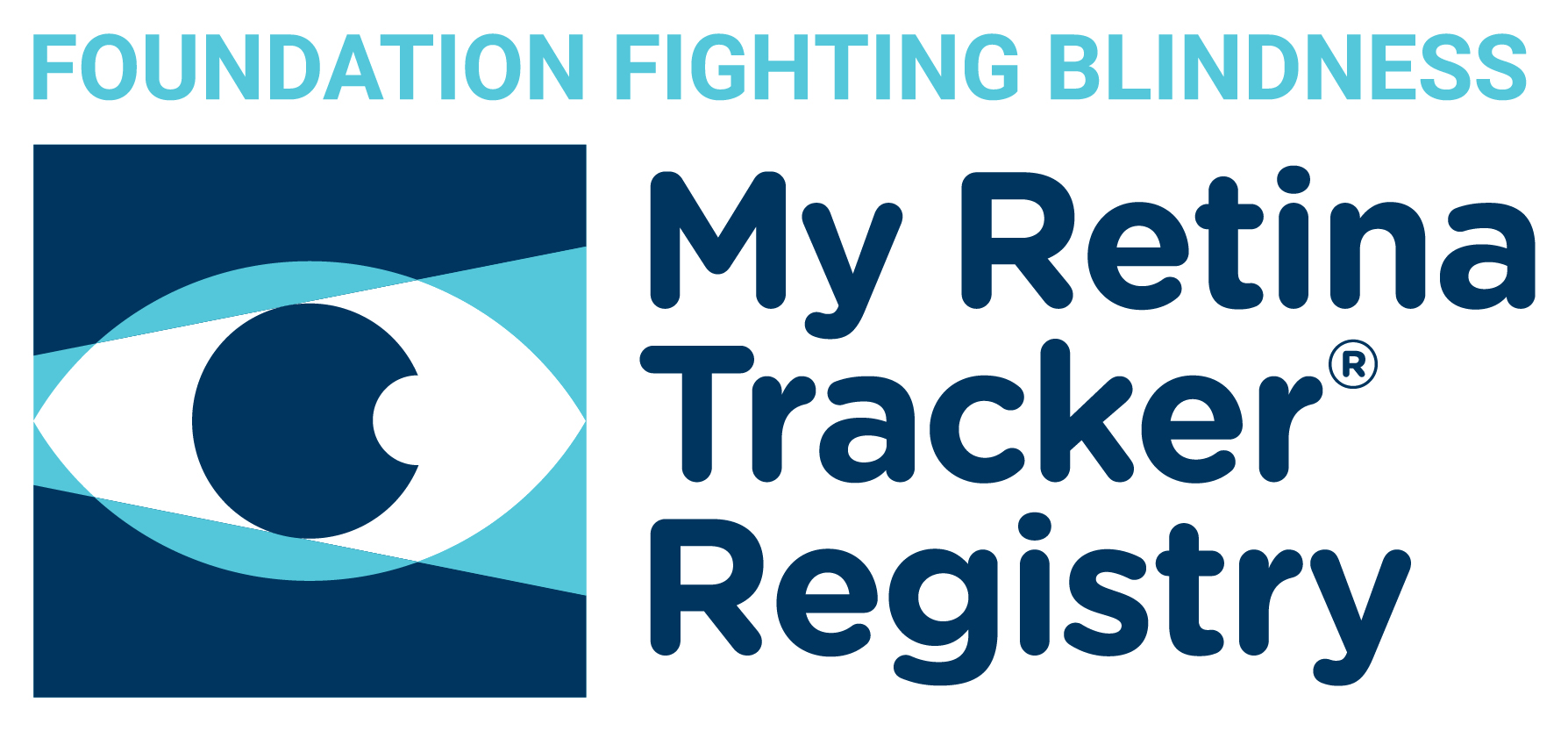
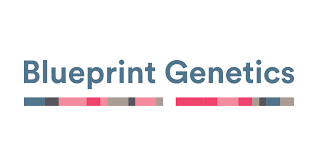
# Provides more accurate prognosis for patient.

# Confirms inheritance pattern, risk for other family members.

# Potential qualification for clinical trials or gene therapy (RPE65, LUXTURNA®).

# Currently ~40 IRD clinical trials underway, most require genetic diagnosis.

# **Slide 5 Best-in-Class Partnership for No-Cost Genetic Testing and Counseling**



# Images 1-3 Logos for Blueprint Genetics, InformedDNA, and the My Retina Tracker Registry

* Helping patients and families manage IRDs.
* Drive clinical research for patients with inherited retinal diseases.
* Getting patients on path to clinical trials and treatments.

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# **Slide 6 Open Access Genetic Testing**

* Open to all patients in the US clinically diagnosed with an IRD. (Not patients who have received genetic testing during the past four years using a panel of >32 genes, WES, WGS.)
* More than 8,000 people have ordered test (since 2017).
* Doctor orders genetic test online through Blueprint. The process is straightforward.
* ~4 weeks turnaround time for results.
* Patients are encouraged to join My Retina Tracker Registry.
* Genetic counseling through InformedDNA or doctor/clinic.

# Image 4 Hand holding a test tube, genetic code (the letters A, C, T, and G) in the background



# **Slide 7 Patient’s Personal Information Always Protected**

# Patient Privacy

# Patient’s personal information is never released or disseminated through the genetic testing program or the registry.

# Only de-identified data provided.

# **Slide 8 Best-in-Class Testing Panel**

* 322 gene panel
* Includes RPGR (XLRP), leading cause of retinitis pigmentosa:
  + 5.7% (90/1582) of IRD patients had RPGR mutations
  + 24% of RPGR patients were females with milder disease
  + ORF-15 region well covered
* Identifies copy number variants (insertions, deletions)
* Identifies non-coding variants (intronic mutations)

Image 5 Logo of Blueprint Genetics

# **Slide 9 Best-in-Class Counseling**

# More than 3,000 patients with IRDs counseled in last 3 years

# More than 90% of doctors using My Retina Tracker Program used InformedDNA

# Counselors have an average of 7 years of clinical genetic counseling experience

# Licensed counselor for every state

# Telephone-based, flexible hours

# Sessions: 60-75 minutes

# Genetic counseling required

# **Slide 10 My Retina Tracker Registry**

# Global, Free, Secure, Easy-to-Use

# Patient-Controlled

# Researchers use for clinical trial recruitment

# Only de-identified data is shared with researchers and companies

# ~16,000 active/useful registrants

# ~150 requests for access (researchers/companies)

# www.MyRetinaTracker.org

Images 5-7 HIPAA compliance logo to ensure data privacy in the US, GDPR logo to ensure data privacy and protection in the EU, Section508 logo to meet US government compliance standards.



# **Slide 11 Clinical Trials**

# 40+ Clinical Trials Underway

# Foundation-funded research attracting commercial investments

# Foundation’s portfolio: 90 projects (global)

# **Slide 12 LUXTURNA: Spark’s RPE65 (LCA & RP) Gene Therapy**

# First FDA-approved gene therapy for the eye or an inherited disease. Patients receiving commercial therapy.

Image 8-9 Yanick Duwe (gene therapy recipient and his father: Spark Therapeutics logo



# **Slide 13 Gene Replacement Therapies in Development**

# AGTC: XLRP (RPGR), achromatopsia (CNGA3, CNGB3)

# MeiraGTx/Janssen: XLRP (RPGR), achromatopsia (CNGA3, CNGB3)

# Biogen: Choroideremia (CHM), XLRP (RPGR), RP (PRPF31)

# 4DMT: Choroideremia (CHM), XLRP (RPGR)

# Horama: RP (PDE6B, CRB1)

# Novartis: RP (RLBP1)

# Iveric bio: RP (RHO), LCA (CEP290), Best disease (BEST1), USH2A

# Atsena: LCA (GUCY2D, MYO7A-USH1B)

# Odylia-MEE: LCA (RPGRIP1)

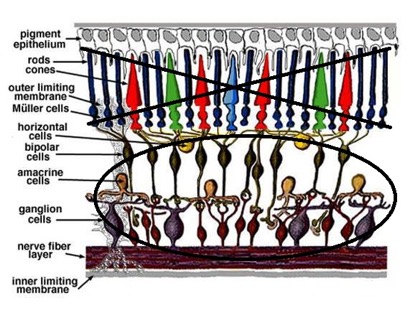
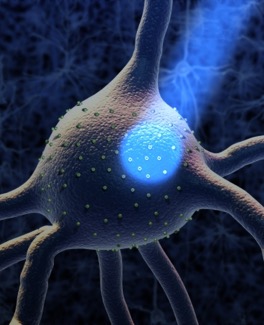
# Numerous other genes targeted: ABCA4

# **Slide 14 Optogenetics**

Restores light sensitivity to retina affected by advanced disease.

* Allergan (US)
* GenSight (UK)
* Bionic Sight (US)
* Vedere

Image 10 -11 Light hitting neuron to signify activation of optogenetic therapy; Side view of retina. The “X” through photoreceptors signifies someone who is completely blind. The oval around ganglion cells that can be targets for optogenetic therapy.



# **Slide 15 Three Major Foundation Investments**

# Nacuity (Dallas) – up to $7.5 million

# NACA – strong antioxidant to slow vision loss (RP, others)

# Phase 1/2 clinical trial launching in Australia for Usher syndrome

# Similar to NAC, N-acetylcysteine (FDA-approved)

# SparingVision (France) – up to €7 million

# RdCVF – rod-derived cone viability factor (protein)

# Saves cones (RP, others)

# Developed at Institut de la Vision

# ProQR (Netherlands) – up to $7.5 million

# Antisense oligonucleotides (like genetic tape/mask)

# USH2A exon 13 (FFB-funded) – Vision improvements, Phase 1/2

# LCA (CEP290) – Vision improvements, Phase 2/3

# RP (RHO-P23H) – Phase 1/2

# **Slide 16 Stem Cells**

ReNeuron (Mass Eye and Ear)

* Transplant partially developed photoreceptors
* Functionally replace lost photoreceptors
* Improved visual acuity for patients in Phase 2 –  
  ~3 lines on an eye chart at 12 months
* Jason Comander leading trial
* Significant funding from FFB

Image 12 Dr. Jason Comander, Mass Eye and Ear, lead investigator on ReNeuron trial



# **Slide 17 Thank You**

# FightingBlindness.org

# ClinicalTrials.gov

# MyRetinaTracker.org