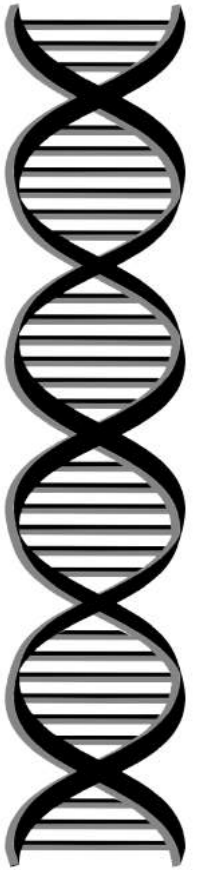
USH Connections Week 2020

July 11, 2020

Genetic Testing



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Brian Mansfield, PhD
Executive Vice President Research, Interim Chief Scientific Officer

Genetic Testing for an Inherited Retinal Disease

- May identify gene causing retinal condition
- May provide more comfort in the certainty about your retinal condition
- May confirm or refine a clinical diagnosis – suggest additional tests
- May guide better informed decisions
- May guide you to potential clinical trials
- When shared in a Registry, can inform prevalence, motivate research

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Foundation Fighting Blindness

No Cost Genetic Testing Program

Two programs available

Your clinician's choice

Both provide:

- ✓ The same high quality genetic test
- ✓ Genetic counseling
- ✓ No cost for the test or counseling
- ✓ Must be ordered by a clinician – may charge for a visit

NO COST

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My Retina Tracker Registry - IRB Genetic Testing Study

- Membership of My Retina Tracker Registry required for testing
  - Foundations inherited retinal disease registry
- Pilot study – still available
- Only approved clinical sites can order
- Demonstrated demand and feasibility
- Identified challenges in administration of the program

MEMBERS ONLY

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My Retina Tracker Program

Open Access Genetic Testing

ALL
WELCOME

- ✓ Initiated October 2019
- ✓ Any qualified clinician can order the test
- ✓ Anyone with an IRD may be eligible
- ✓ DO NOT need to be member of the My Retina Tracker Registry

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Open Access - Eligibility for Testing

Live in the US or US territories

Clinically diagnosed with an inherited retinal disease

NOT to be used for general eye disease screening

NOT tested since 2016 for more than 32 relevant genes

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How to Get Tested

Receive a diagnosis for an inherited retinal disease

Ask your clinician to order the test

www.fightingblindness.org/open-access-genetic-testing-program

Genetic counselor explains result and opportunities and can help you join My Retina Tracker Registry

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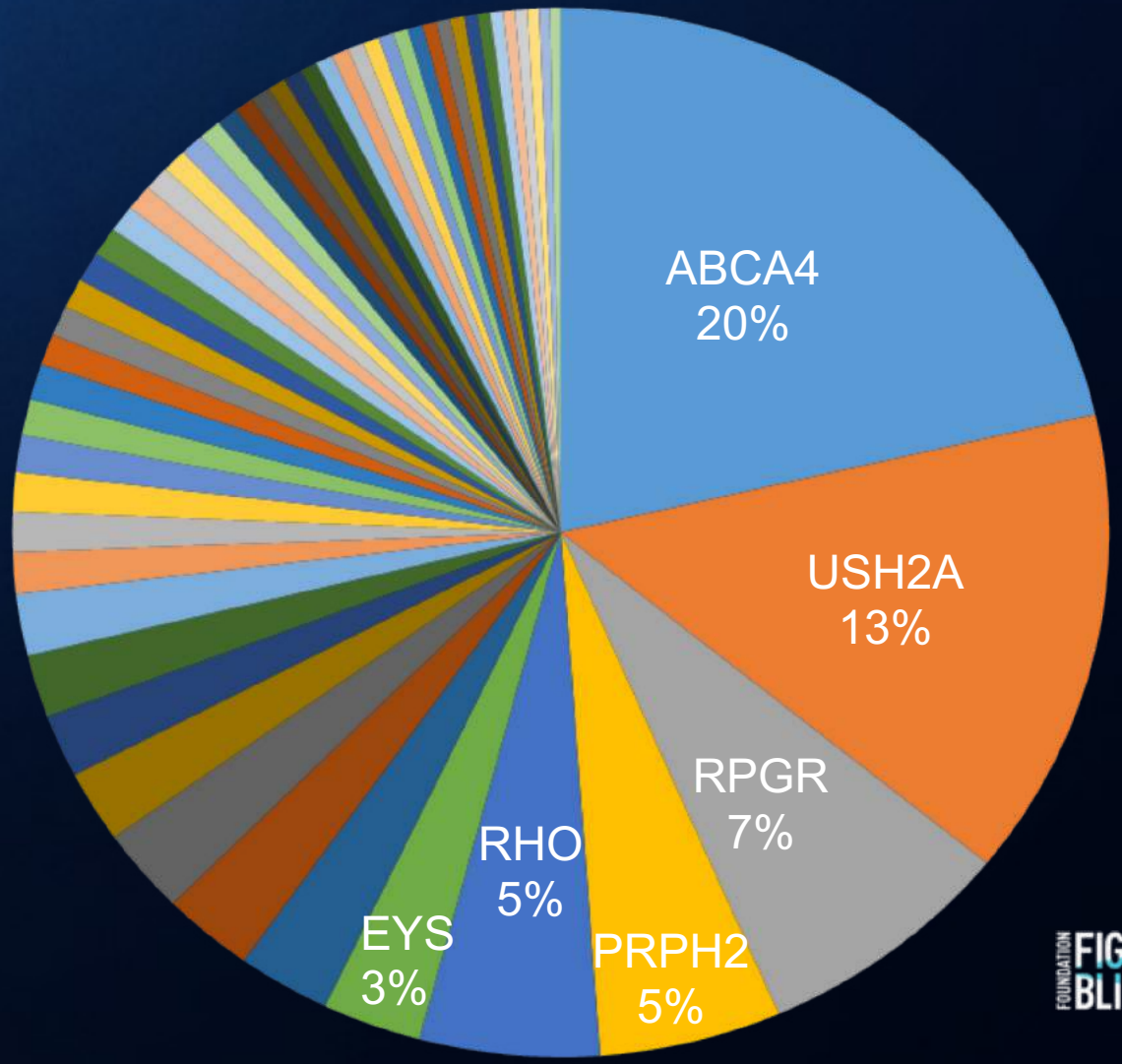
In Our Genetic Testing

- 7,700 test requests
- 148 different disease genes
- 6 genes accounted for 53%

Assistant Research Scientist
Genetic Counselor,
Inherited Retinal Dystrophy Clinic
Kellogg Eye Center, Michigan



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Outcomes of Usher Syndrome Genetic Testing

For people with a clinical diagnosis of Usher syndrome

- Most are referred with an unspecified type
- 91% do have Usher syndrome
  - 3% have atypical genotype phenotype relationship
- 3% have vision and hearing loss, but the gene is consistent with a different syndrome such as Alstrom, Stickler, Zellweger
- 2% have mutations in genes associated with non-syndromic disease such as CHM, PDE6A, SNRNP20, MAK, ABCA4

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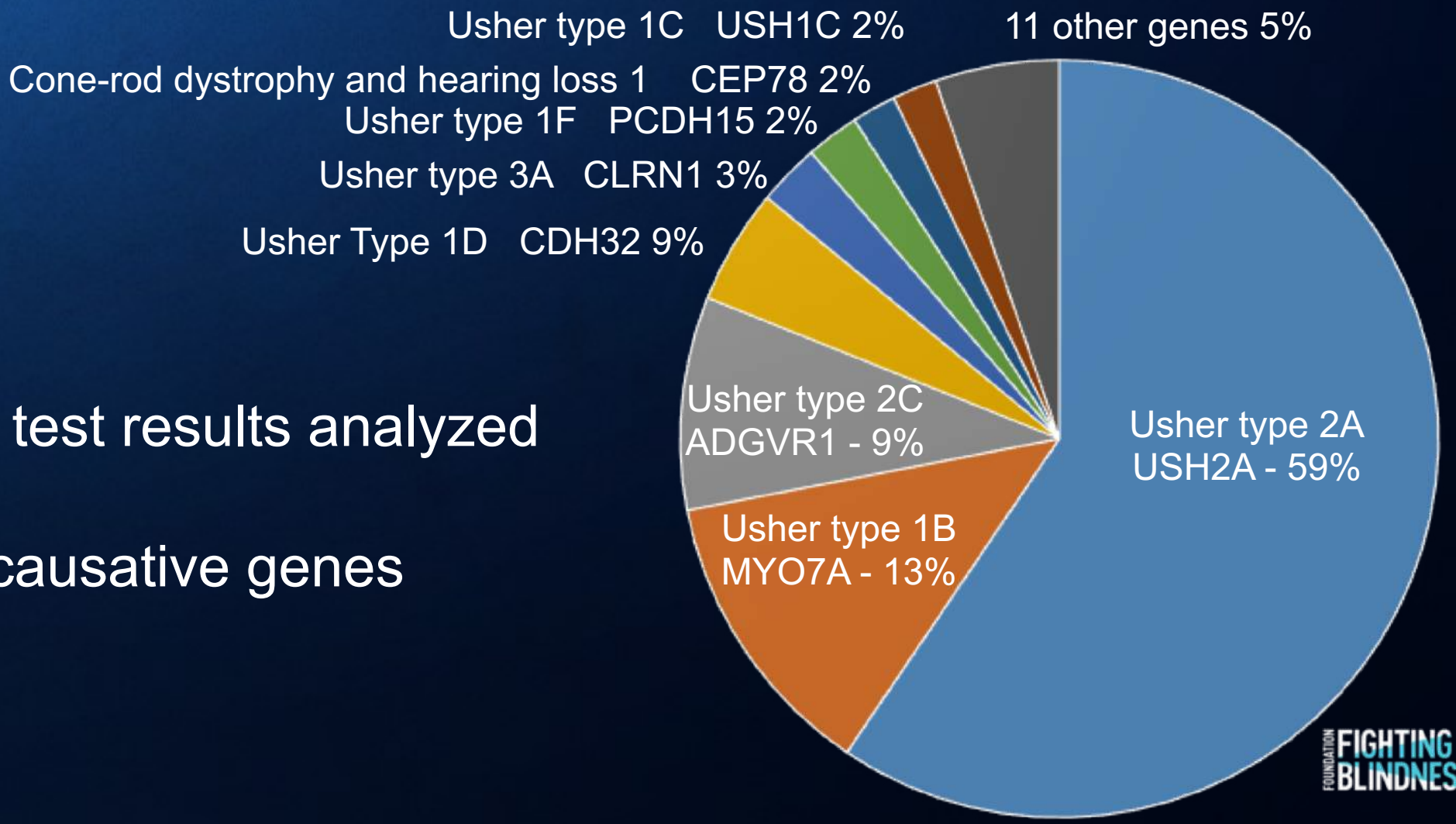
Genetic Testing Program

Most Common Genetic Cause of Usher Syndrome



- 264 genetic test results analyzed
- At least 20 causative genes

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Registries Help Share Data Responsibly

- ✓ Centralized sources of information
- ✓ Connection to researchers, industry, clinical trials
- ✓ Share de-identified data
  - ✓ Member perspective of their disease
  - ✓ Clinical perspective
  - ✓ Genetic data
- ✓ More complete perspective on disease

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Registries Help Share Data Responsibly

- **USH Trust Registry**
  - Focused on all aspects of living with hearing and vision loss
- **My Retina Tracker Registry**
  - Focused on retina only - all inherited retinal diseases
- Membership is not exclusive – complementary
- Joining both helps make no cost genetic testing sustainable

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Partners Support Registry and Genetic Testing

My Retina Tracker Registry Partners

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ELO Eloxx Pharmaceuticals

agtc Visionary science for life changing cures.

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SOFIA SEES HOPE

MEIRAGTx

Open Access Genetic Testing Partners

ProQR®

InformedDNA Genetics, Decoded.

SOFIA SEES HOPE

Blueprint Genetics

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Resources

USH Trust Registry

www.usher-registry.org

My Retina Tracker Registry

www.MyRetinaTracker.org

Coordinator@MyRetinaTracker.org

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