USH Connections Week 2020

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Genetic Testing

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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
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
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
My Retina Tracker Program

Open Access Genetic Testing

- 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
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
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
In Our Genetic Testing

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

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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
Genetic Testing Program

Most Common Genetic Cause of Usher Syndrome

- Usher type 2A USH2A - 59%
- Usher type 1B MYO7A - 13%
- Usher type 2C ADGVR1 - 9%
- Usher type 1C USH1C - 2%
- Cone-rod dystrophy and hearing loss 1 CEP78 - 2%
- Usher type 1F PCDH15 - 2%
- Usher type 3A CLRN1 - 3%
- Usher Type 1D CDH32 - 9%
- 11 other genes - 5%

- 264 genetic test results analyzed
- At least 20 causative genes
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
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
Partners Support Registry and Genetic Testing

My Retina Tracker Registry Partners

Open Access Genetic Testing Partners

- THE GEORGE GUND FOUNDATION
- Eloxx Pharmaceuticals
- agtc
- ProQR
- InformedDNA
- Blueprint Genetics
- Fighting Blindness Foundation
- SOFIA SEES HOPE
- MEIRA GTx
- THE GEORGE GUND FOUNDATION
- Fighting Blindness Foundation
Resources

USH Trust Registry
www.usher-registry.org

My Retina Tracker Registry
www.MyRetinaTracker.org

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