The International Usher Syndrome Registry

Mark Dunning
Chairman, Usher Syndrome Coalition
President, Decibels Foundation
Father, Bella and Jack
From the Petri Dish to You

• Basic research
  • Understanding the fundamental aspects of a disease
  • Finding genes, genotype/phenotype, disease mechanism

• Translational research
  • Develop effective treatment in an animal model

• Clinical Research
  • Clinical Trials

• Treatment and Follow-up
Research Continuum

- The line between basic and translational research is blurry
- Research and trials take years
- Four phases
  - Phase I: Safety in small group
  - Phase II: Efficacy in small group
  - Phase III: Efficacy in large group
  - Phase IV: Monitor long term clinical usage for side effects
- Only 1 in 12 human trials results in a clinical treatment
## State of Usher Syndrome Research

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<th>Basic</th>
<th>Translational</th>
<th>Trials</th>
<th>Treatments</th>
<th>Notes</th>
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Identify potential candidates
We Are Not in Touch with Enough Usher Families

• Research community agrees

• International Symposium on Usher Syndrome
  • Harvard Medical School, July 2014

• Session to discuss roadmap to treatments

• Greatest barrier to treatment development: We are not in touch with enough families

• Clinical trials will be suspended, abandoned, or simply not begun without enough candidates.
Families are Critical to Research Success

- Source of natural history information
- Source of genetic information
- Pool of candidates for clinical trials
- Source of funding and advocacy
- Source of awareness which impacts funding and motivates researchers
400,000 Miracles

- Conservatively 1/20,000 have Usher syndrome (350,000-400,000)
- Researchers are actively in touch with ~3,000
- Why don’t families join registries or participate in research?
Families are busy living with Usher

• Parents of young children
  • Baby has hearing loss, genetic testing indicates Usher syndrome
  • Found through otolaryngology, not ophthalmology
  • 90% of children born with hearing loss are born to parents with no experience with hearing loss
  • Parents focus on living with hearing loss and communication
    • Language develops in first 5 years of life
  • Fearful of future with vision loss
    • Often in denial
    • Don’t want to meet adults with Usher
  • Vision loss is dealt with later
Families are busy living with Usher

- Teenagers and College age
  - Invincible
    - They’ll never be old, they’ll never be blind
  - Comfortable with hearing loss
  - Driving, college, relationships take priority
Families are busy living with Usher

• Young Adults
  • Focused on starting careers and families
  • Learning to cope with progressing vision loss
    • Mobility, guide dogs
  • Resistance to and acceptance of vision loss
  • Racing against time
    • “Want to do X while I can still see”
Families are busy living with Usher

• Older Adults
  • Already coping with vision loss
  • Skeptical
    • Been told for 40 years that treatments are 10 years away
• Suspicious of large institutions
  • Diagnosis with no hope: Deaf, going blind, no cure
  • Grew up away from family at deaf schools
  • Not allowed to use sign language despite poor technology
• Resigned
  • Believe it is too late for them
Why Aren’t We In Touch with More Families?

• Social Issues
  • Disease is socially isolating
  • Stigma of Deafblindness
  • Vocational impact/Loss of identity
  • Depression, anxiety
  • Transportation, mobility, health issues

• We need to address these issues if we want families to participate in research
Hope Continuum

• Families need time to deal with emotion of diagnosis
• Need to understand disease before following research
• Eventually they want to talk with others about Usher
  • Phase I: Talk to experts
  • Phase II: Meet others of similar age, Usher type
  • Phase III: Participate in larger Usher community
• Full participation in research takes time
The Keys to Engaging Families

• Hope for the future
  • Information about research advancements
  • Establishing relationships with researchers
  • Role models with Usher

• Feel valued
  • Don’t want to be lab rats

• Combat isolation

• Identity, self-esteem
  • People with Usher and strong sense of self stay involved in Usher activities
Hope Continuum

• Web site
  • Dozens of presentations from leading researchers
  • Blog with family experiences and success stories
  • Information on disease
  • Initial sense of hope

• Monthly Researcher Conference Call
  • Notes shared with nearly 1,500 people world-wide

• Quarterly newsletters
### Hope Continuum

- **International Usher Syndrome Family Network**
  - 250 families/20 countries

- **Social Media**
  - Facebook: 1,000 likes/5 stars
  - Twitter: @UsherCoalition

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Hope Continuum

- Annual Family Conferences
  - One day
  - Families and researchers
  - Child programs
  - Accessible
  - Opportunity for all in the Usher syndrome community to meet
  - Long breaks for networking
  - New Orleans, July 11th, 2015
Hope Continuum

- International Symposia
  - Every 2-3 years
  - July 2014, Harvard Medical School
  - Next symposium in Europe
  - Two day science conference combined with one day family conference
  - Collaboration between researchers
  - Opportunity to connect with families

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Participation

• Join the International Usher Syndrome Registry
• Mentors in International Usher Syndrome Family Network
• Volunteering
• Fundraising
• Advocacy
• Participating in psychosocial studies
• Candidate for clinical trials
International Usher Syndrome Registry Facts

• Largest Usher syndrome registry in the world
• More than 700 families
• 40 countries
• Developed by Mani Iyer
  • Has Usher
  • Volunteered his time
  • Created character by character
International Usher Syndrome Registry

- Available online
  - www.usher-registry.org
  - Usher Syndrome Coalition web site
- Voluntary
  - Opt out at any time
- Simple
  - Requires only name and e-mail
- Accessible
  - Works with JAWS and other readers
  - Available in English, Spanish, Hebrew, and Dutch
  - In development: German, Portuguese, French, Swedish
- Secure and confidential: HIPAA compliant
  - Health Insurance Portability and Accountability Act (HIPAA)
The Value of the International Usher Syndrome Registry

• Keeps families informed about clinical trials
• Allows families the opportunity to participate in research
• Gives researchers a means of contacting people with Usher syndrome
• Creates a large pool of potential candidates for clinical trials
  • Registering does NOT mean you have volunteered to participate
• May provides data vital for advocacy efforts
  • Number of people with Usher by country, by state
  • Psychosocial data on health and employment can be monetized
International Usher Syndrome Registry Research Support Process

• Usher Syndrome Coalition acts as firewall
  • Researchers have no direct access to registrants
  • The Coalition shares researcher contact information with appropriate registrants
  • Registrants decide whether to contact or not

• Registry Review Board
  • Review every request for data access

• Researchers must have Institutional Review Board (IRB)

• Only de-identified data is shared with researchers
  • How many Usher 1b are in the registry?
Extending the Community

• 40 countries in registry
• Establishing International Partners
  • Stichting MUS
  • Austria, France, Australia, UK, Sweden, Ireland
• Developing local networks to reach families
  • 5-6% of Dutch families in registry
  • Equivalent of 2,200 in US/20,000 Globally
• Working with Community Providers
  • Helen Keller National Center
  • International support organizations
• 2016 by 2016
Registries Everywhere

• Different registries serve different purposes
• What do you get in return?
• Working to integrate with other registries
• No data will be shared without individual consent
• Value in joining multiple registries
Other Registries

• Track other diseases in addition to Usher syndrome
• My Retina Tracker – Foundation Fighting Blindness
  • Open to all researchers
  • The prevalence, variability, and progression of different diseases
  • Natural history and diagnostic information
• Helen Keller and Sense
  • Support services for deafblind
• Genetic registries
  • Track different disease variants
• Institutional and national registries
Join the International Usher Syndrome Registry

- Better quality of life for people with Usher syndrome
- Strong social connections
- Informed families actively involved in research efforts
  - Participation in research/natural history
  - Raising awareness
  - Funding
- Engaged families
  - Higher self-esteem
  - Reduced incidence of anxiety and depression
Annual Appeal

• Chairman’s Challenge: $7500 match
• Consider a recurring donation
  • $20/month
• Ensures that the Usher Syndrome Coalition continues to provide services
• Visit www.usher-syndrome.org to learn how you can help