# The International Usher Syndrome Registry

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USHER SYNDROME COALITION

# 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

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# 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

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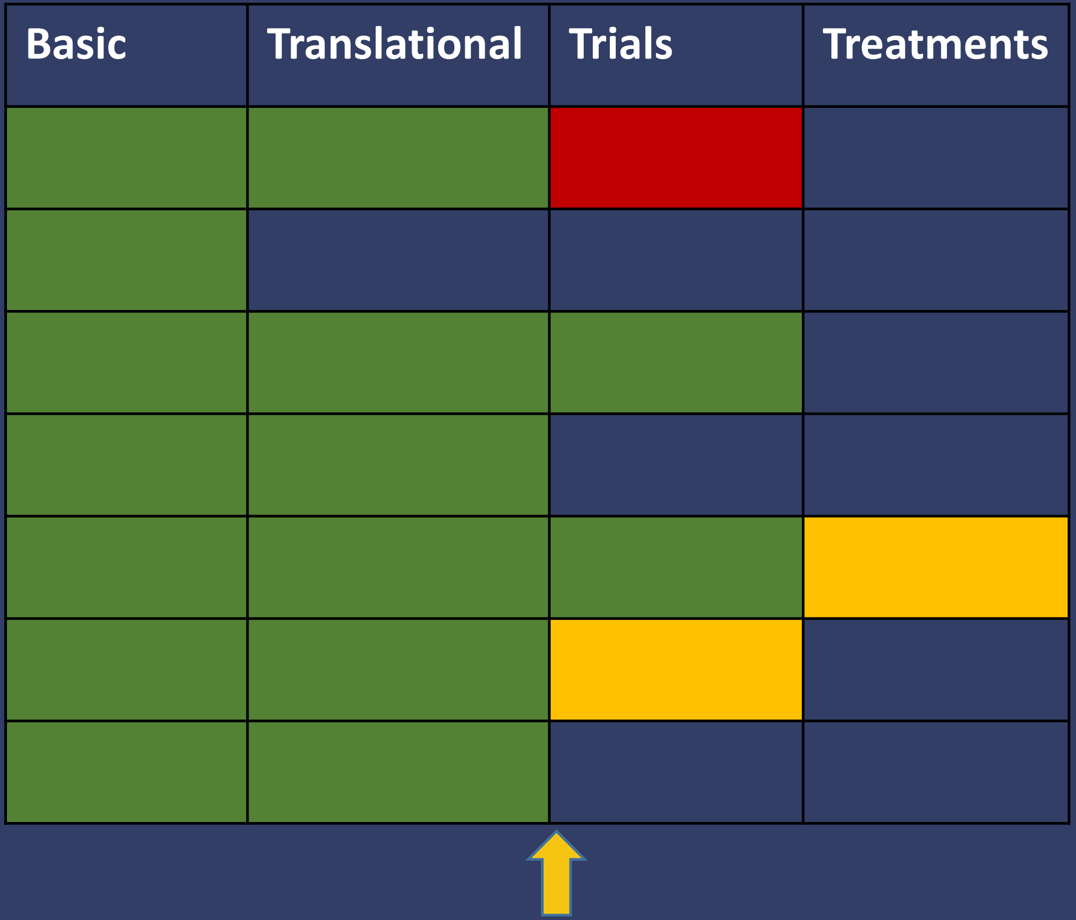
# State of Usher Syndrome Research



| | Basic | Translational | Trials | Treatments | Notes |
|---|---|---|---|---|---|
| **Stem Cells (Vision)** | 🟩 | 🟩 | 🟥 | | Stargardts and AMD |
| **Stem Cells (Hearing)** | 🟩 | | | | |
| **Gene Therapies** | 🟩 | 🟩 | 🟩 | | Usher 1b |
| **Drug Therapies** | 🟩 | 🟩 | | | |
| **Ocular Implants** | 🟩 | 🟩 | 🟩 | 🟨 | Usher? |
| **Vestibular Implants** | 🟩 | 🟩 | 🟨 | | Usher? |
| **Optogenetics** | 🟩 | 🟩 | | | |

Identify potential candidates

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# 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.**

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# 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

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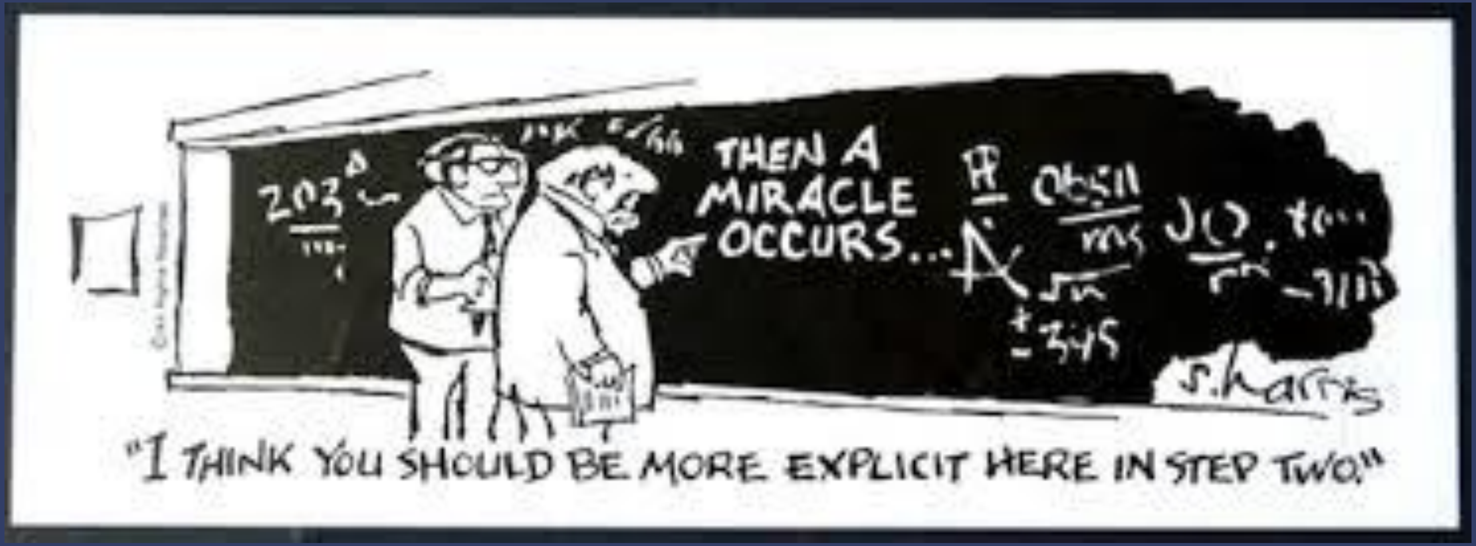
# 400,000 Miracles

- 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?



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# 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

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# 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

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# 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

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# 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

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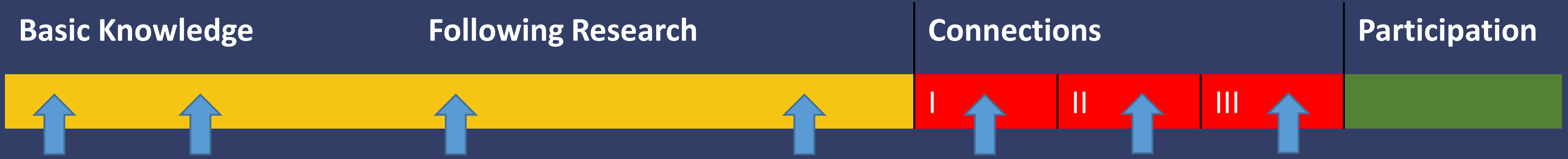
# Hope Continuum

- 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



**Basic Knowledge** **Following Research** **Connections** **Participation**

I II III

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# 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

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# 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

**Basic Knowledge** **Following Research** **Connections** **Participation**

I II III

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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

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# Hope Continuum

- International Symposiums
  - 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

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# 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

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# 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)

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# 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

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# 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?

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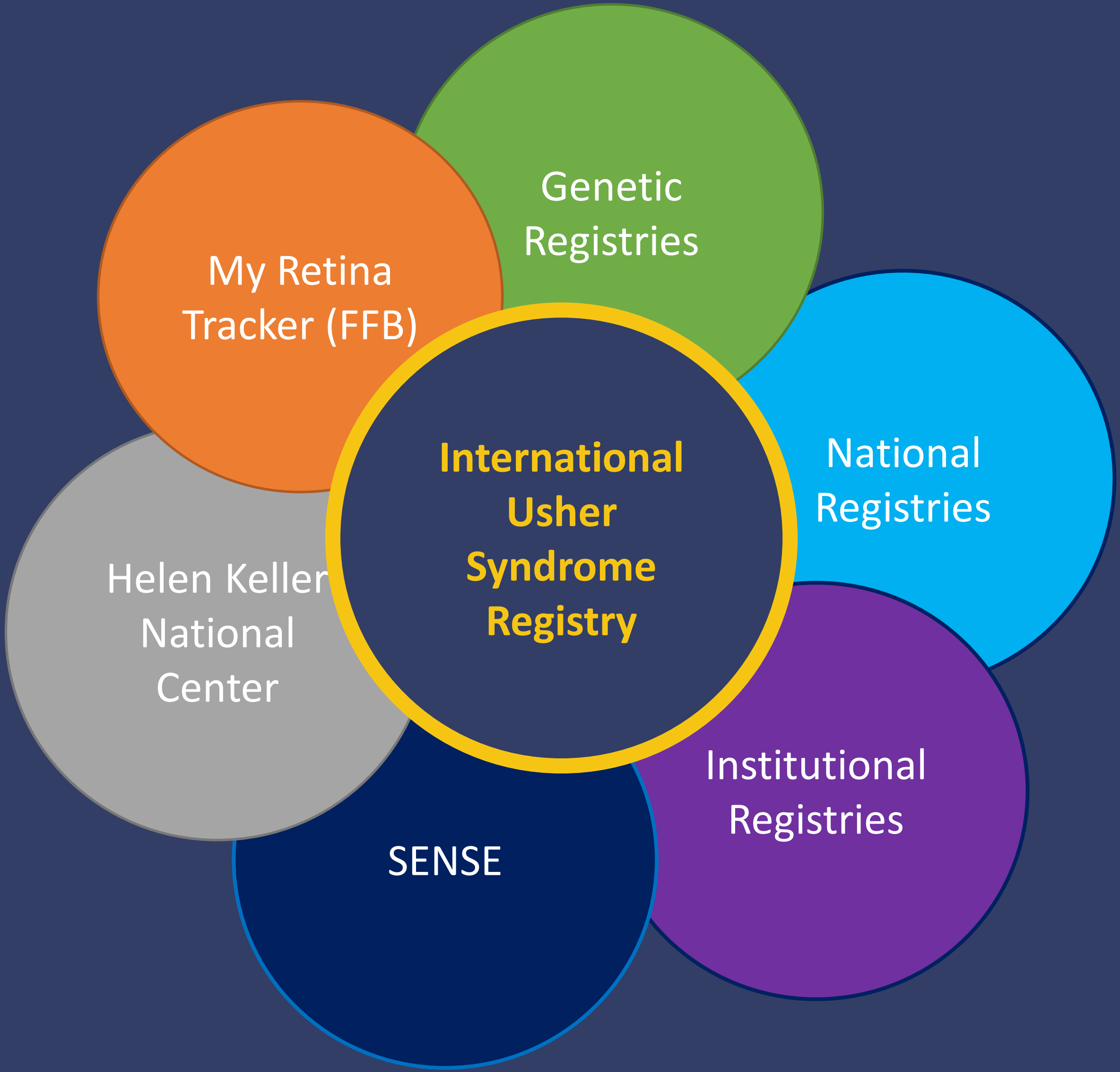
# Extending the Community

- 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

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# 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

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# 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

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# 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

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