the decibels foundation

The Coalition For Usher Syndrome Research

THE CAROLINE BASS FUND

The Connection Between Families and Research

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Father, Jack age 9, Bella (Usher syndrome) age 12

The Problem

- Diagnosis is frightening
- Often poorly delivered “Deaf, blind, no cure.”
- Families struggle to find good information about the disease.
- Families have a hard time finding other Usher families.
- Even informed families can feel helpless.
  - “I’m not a doctor, I’m not rich, I don’t have wealthy friends. Do I just wait and hope someone else finds a treatment?”
- Result: Lose touch with families.

The Impact on Research

- Engaged families are critical to finding a cure.
- Source of natural history information. Only way to tell if a treatment is successful.
- Source of genetic information.
- Pool of candidates for clinical trials.
- Source of funding and lobbying.
- Source of awareness which impacts funding and motivates researchers.
- Educational resource

What is the Coalition for Usher Syndrome Research

- Collection of concerned researchers, families, and organizations dedicated to building an Usher syndrome community to help support Usher research.
- Not a fund raising or funding organization.
- Our members:
  - Foundation Fighting Blindness
  - Harvard Medical School Center for Hereditary Deafness
  - The Decibels Foundation
  - Hear See Hope Foundation
  - Children’s Hospital Boston
  - The Audiological Research Center in Orebro, Sweden
  - Oregon Health and Sciences University Casey Eye Institute
  - Seattle Children’s Hospital
  - Boys Town Hospital
  - University of Iowa Carver Laboratory
  - Unidad de Genetica, Hospital Universitario La Fe, Valencia, Spain
  - The Megan Foundation

Families Struggle to Find Good Information

- Solution: Create a web site with the best Usher information available.
  - Why early diagnosis is important?
  - Connections to families and researchers
  - Explanation of Usher syndrome symptoms/causes.
  - Latest research news.
  - Presentations and abstracts from Usher researcher and family conferences.
  - Links to blogs maintained by Usher families.
- Overall sense of hope.
- www.usher-syndrome.org

Families have a hard time finding other Usher families

- Solution: Usher Syndrome Family Network
- 1 in 6500 people have some form of Usher. It's rare.
- May be very hard to find others with the same type of Usher.
- Connect families with Usher syndrome.
- Members include people with Usher, parents, siblings, and grandparents.
- International connections.

The Diagnosis is Frightening

- Solution: Usher Syndrome Blog
- In depth look at issues facing families with Usher.
- Science behind Usher and Usher research by Jennifer Phillips, Ph.D.
- Family decisions, common problems, questions, and experiences by Mark Dunning and other Usher families.
- *“The most thoughtful exploration of Usher Syndrome that I've ever seen on the internet.”* – *Reader Comment*

The delivery of the diagnosis is often under-informed

- Solution: Education and collaboration
- Usher Syndrome Family Conferences.
- Monthly Researcher/Family Conference Calls.
- Usher Researcher Conferences
- Newsletter
- Goal: Connect families and researchers and build relationships.

Need to identify families for clinical trials and treatments

- Problem II: Need natural history information on families
- Solution: Usher syndrome Registry
- In development – expected to launch in September, 2011
- Web accessible by families and researchers
- Voluntary
- Secure and controllable by individual families
- No unsolicited contact
- Includes:
  - Contact information for Usher families
  - Basic diagnosis information (what type of Usher do you have?)
  - Interests (connecting with families, learning about clinical trials)
  - Research questionnaires
    - Natural history
    - Dietary
    - Psycho-social

What’s missing?

- An education program for physicians
- Promote registry
- Organizers and funding for yearly family conferences
- Organizers and funding for bi-yearly researcher conferences
- Lobbying effort for Usher syndrome line item in NIH budget

What can you do right now?

- Join the family network and stay engaged
- Volunteer to help organize a conference
- Volunteer to help test the registry
- Contact your representatives in Congress about NIH funding
- Once the registry is online, register and be open with your information
- Promote the registry

www.usher-syndrome.org