The Connection Between Families and Research

Mark Dunning
President, The Decibels Foundation
Executive Director, The Coalition for Usher Syndrome Research
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