Usher Syndrome Coalition

Mark Dunning
Chairman, Coalition for Usher Syndrome Research
Bella’s Story

• 15 years old
• Born deaf, 2 cochlear implants
• Diagnosed with Usher at age 8
• Loves horses
• Grudgingly loves her little brother, Jack
Psychology of Usher syndrome

• Why do we fear the Usher diagnosis?
  – Um, deaf? Blind? Duh!

• Usher is a threat to:
  – Security of employment, family, property
  – Bond with others, friendships, relationships
  – Self-esteem, confidence, achievement

• Maslow/Attachment Theory
Usher syndrome interventions

• Best solution: Interventions for Usher syndrome

• Hearing loss
  – Digital hearing aids/Cochlear implants
  – ASL/Deaf community

• Vestibular issues
  – Physical therapy/Hippotherapy
  – Vestibular prosthesis

• Vision loss
  – Vitamin A/DHA/Gene therapy
  – Nothing definitively stops the progression
What can a family do?

- How can I help?
- Give money to fund research
- What if I don’t have money to invest in research?
  - 82% unemployment in deaf-blind
  - Raise money to fund research
- What if I don’t have ability to fundraise?
  - Social isolation
  - Participate in clinical trials/research
- What if there are no appropriate clinical trials?
  - Ummm...?
- Need other means to engage families in support of research
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.
Usher Syndrome Coalition

• Goals:
  – To support research
  – Bond with others
  – Improve self-esteem, confidence
  – Participate in ultimate achievement: treatments for Usher syndrome
Usher Syndrome Coalition

• Krista Vasi – Executive Director
• Host of volunteers
• Dedicated researchers/clinicians
• Office in Maynard, MA
Participating with the Coalition

• What are we doing?
• How can you get involved?
Usher Syndrome Registry

- Online registry of people with Usher syndrome
- Currently contact information/mailing list
- Researchers request access to data
- Requires IRB approval, approval by registry review board
- Chinese wall: Coalition contacts registrants on behalf of researchers
- Source of natural history information
- Candidates for clinical trials
  - Families from 25 countries
- Available in English and Spanish
- Hebrew and French coming soon
- German and Dutch in the works
Helping with Usher Syndrome Registry

• Join the registry
  – www.usher-registry.org

• Assist with outreach initiatives to recruit new registrants
  – Assist in educational efforts to the Usher syndrome community around the need for participation in clinical research
  – Physicians, support groups, and other stakeholders in the Usher syndrome community

• Assist with enhancements and developments
  – Testing
  – Translate registry into more languages

• Provide Registry support and work with leading researchers
Usher Syndrome Family Network

• 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.
• More than 200 families from 13 countries.
• Contact information is protected
  – When a new family joins, we pass on their contact information to the others in the network.
  – Choice of the current families to decide if they want to contact the new member or not.
  – The new family will not know to whom we've passed on information, so there is no pressure to contact them.
Helping with the Family Network

• Join the family network
• Contact new families that join
• Share information
Government Relations

• Working to increase federal funding for Usher syndrome research.
• Government Relations committee meets every other week
• Process
  – Identify champions
  – Language submitted to sub-committees in both chambers
  – Language accepted by both chambers
  – Language in final budget bill
Helping with Government Relations

• Contact members of Congress
  – [http://ushersyndrome.net/site/advocacy/](http://ushersyndrome.net/site/advocacy/)
• Help develop work plan
• Track and research legislation and other Congressional Initiative
• Research Federal agencies, programs and grants which could accelerate Usher syndrome research, such as the President’s BRAIN Initiative
• Assist with the writing and compilation of written materials for internal and external audiences
International Symposium on Usher Syndrome

• Harvard Medical School, July 10-12, 2014
  – Combined family/science conference
  – All the leading Usher syndrome researcher in the world
  – Largest gathering of Usher families and researchers

• 2015 Family Conference, New Orleans, LA
Confirmed Speakers

- Zubair Ahmed, PhD (Cincinnati Children’s Hospital, Ohio, USA)
- Kumar Alagramam, PhD (Case Western Reserve University, Ohio, USA)
- Maria Bitner-Glindzicz, MD (University College London, United Kingdom)
- Constance Cepko, PhD (Harvard Medical School, Boston, USA)
- Dominic Cosgrove, PhD (Boys Town National Research Hospital, Nebraska, USA)
- Mark Dunning (Coalition for Usher Syndrome Research, Boston, Massachusetts, USA)
- Anne Fulton, MD (Boston Children’s Hospital, Boston, Massachusetts, USA)
- Margaret Kenna, MD, MPH (Boston Children’s Hospital, Boston, Massachusetts, USA)
- William Kimberling, PhD (University of Iowa, Iowa, USA)
- Hannie Kremer, PhD (Radboud University Nijmegen Medical Centre, Netherlands)
- Jennifer Lentz, PhD (Louisiana State University, Louisiana, USA)
- José M. Millán, PhD (Unidad de Genética, Spain)
- Ilene Miner, LSCW (Gallaudet Research Institute, Washington, DC, USA)
- Kerstin Nagel-Wolfrum, PhD (Johannes Gutenberg Universität Mainz, Germany)
- René Pellerin (The Unstoppable, Vermont, USA)
- Christine Petit, MD, PhD (Institut Pasteur, Paris, France)
- Eric Pierce, MD, PhD (Massachusetts Eye and Ear, Boston, USA)
- Heidi Rehm, PhD (Harvard University, Boston, USA)
- Stephen Rose, PhD, (Foundation Fighting Blindness, USA)
- Anne-Françoise Roux, PhD (Hôpital Universitaire, Montpellier, France)
- José-Alain Sahel, MD, PhD (Institut de la vision, Paris, France)
- Nizar Smaoui, MD (GeneDX, Maryland, USA)
- Edwin Stone, MD, PhD (University of Iowa, Iowa, USA)
- Richard Weleber, MD (Oregon Health and Science University, Oregon, USA)
- Monte Westerfield, PhD (Institute of Neuroscience, Oregon, USA)
- Uwe Wolfrum, PhD (Johannes Gutenberg Universität Mainz, Germany)
- Luk Vandenberghe, PhD (Massachusetts Eye and Ear, Boston, Massachusetts, USA)
Helping with International Symposium and future conferences

- Assist with event coordination
- Assist with childcare coordination
- Coordinate interpreters CART, FM, Braille requests
- Maintain/update website
- Manage social media content/e-mail communication
- Bookkeeping
- Fundraising/Sponsorships
- Funding needed for Symposium: $60,000-$80,000
- 2015 Family Conference: $25,000
Help with Funding

• Fundraisers
• Identifying funding sources
• Writing grants
• Operating budget 2013: $130,000
  – Executive Director, Registry, Family Network, Family
    Conference, Monthly Phone Calls, Web Site, Government
    Relations
• Operating budget 2014: $240,000
  – Above plus International Symposium
• Goal: $400,000 annually
  – Above plus Event Coordinator, Registry Coordinator,
    Government Relations support, Administrative support
Be Happy

• Usher syndrome is a threat, not a sentence
• Stay engaged, learn latest on research
• Opportunities to participate in finding treatments
• Develop and maintain relationships with other Usher families
• Attend Symposium and family conferences
• Help us build an Usher community
• Usher syndrome can bring us together rather than isolate us
Thank You

• Registry:  www.usher-registry.org
• Web site:   www.usher-syndrome.org
• Advocacy site: www.ushersyndrome.net
• m.dunning@lek.com
• K.vasi@usher-syndrome.org