



The Coalition for
Usher Syndrome
Research

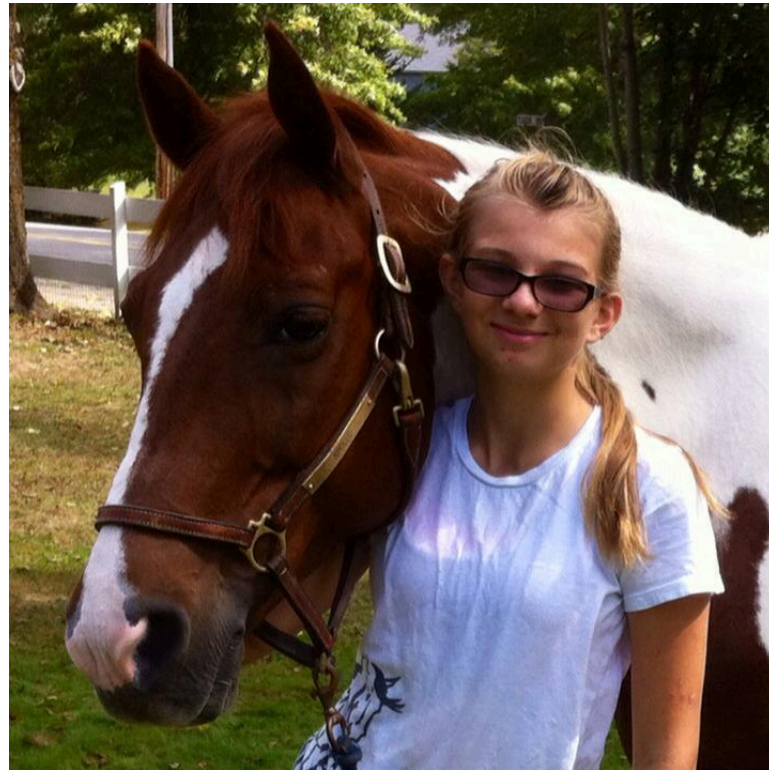
Usher Syndrome Coalition

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

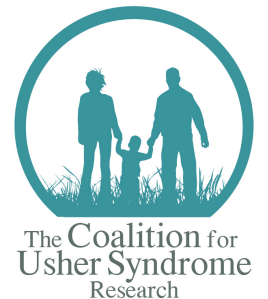
- Best solution: Interventions for Usher syndrome
- Hearing loss
 - Digital hearing aids/Cochlear implants
 - ASL/Deaf community
- Vestibular issues
 - Physical therapy/Hippotherapy
 - Vestibular prosthesis
- Vision loss
 - Vitamin A/DHA/Gene therapy
 - Nothing definitively stops the progression

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.



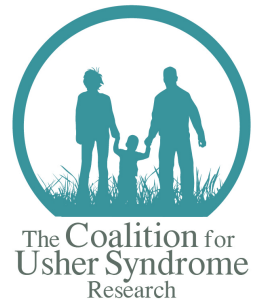
Usher Syndrome Coalition

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



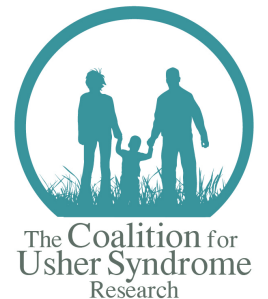
Usher Syndrome Coalition

- 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 in to 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/>
- 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

