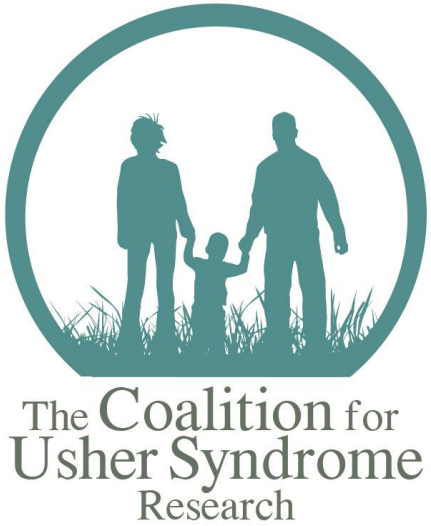


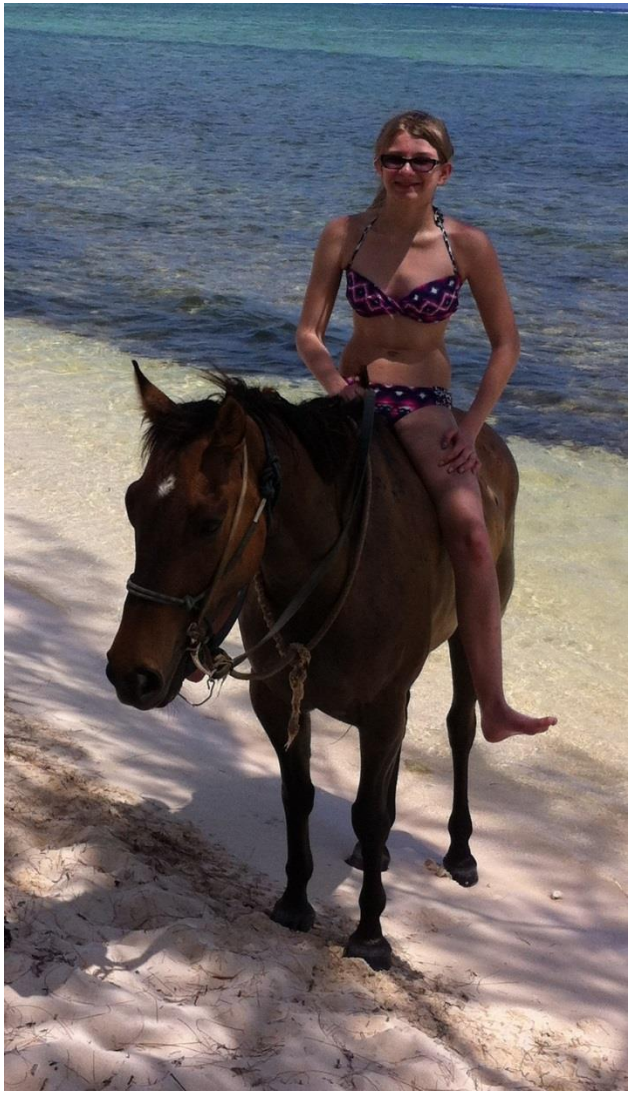
Hope and Building an Usher Syndrome Community

Mark Dunning

Chairman, Coalition for Usher Syndrome Research

Bella's Story

- 14 years old
- Born deaf
- Diagnosed with Usher at age 8
- Loves horses
- Grudgingly loves her little brother, Jack



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What is Usher syndrome?

- Leading cause of deaf-blindness in the United States and Europe.
- Congenital hearing loss and progressive vision loss from Retinitis Pigmentosa.
- Historically 3% of patients with congenital bi-lateral sensorineural hearing loss
- Genetic testing indicates higher: 8-12%
- 30,000 to 50,000 people in the United States.
- Orphan disease, defined as “any disease or condition that affects less than 200,000 persons in the United States"

Scary Stuff to Follow…

- Clinical impact of Usher syndrome…

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Hearing Loss

- Congenital: Hearing loss at birth(?)
- Bi-lateral: Both ears
- Sensorineural: Hair cells, not structural
- Usually moderate to profound

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Loss of Vestibular Function

- Vestibular function low or not present in certain types of Usher
- Late walker, poor balance
- Clumsy, low tone
- Three components of balance: vestibular, vision, and musculature

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Retinitis Pigmentosa

- Degenerative condition of retinal cells.
- Rod cells: vision in low light.
- Cone cells: color and acuity.
- Nightblindness
- Peripheral vision

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Normal night vision

Night vision with Usher syndrome

Normal peripheral vision

Usher syndrome early stages

Usher syndrome middle stages

Usher syndrome late stages

Traditional Communication of Diagnosis

- People with Usher are deaf and go blind.
- There are no treatments for Usher syndrome.
- There is nothing you can do to slow the progression.
- It is rare so you probably won't meet other families with Usher syndrome.

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Really Scary Stuff to Follow…

- It’s not the disease that frightens families…
- It’s not the future that scares families…
- Social isolation is the fear!

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Diagnosis of RP
(Ilene Miner)

- Already having problems
- Diagnosis is devastating
- Gone is the expected life, facing the unknown
- Finding information about living with RP is difficult
- Fears, anxieties, depression, suicidal ideation
- People with Usher are more than 2.5 times as likely to commit suicide

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Community & social relationships (Ilene Miner)

- Deafblind people don't fit anywhere
- Friends withdraw
- Community roles/activities are difficult - attendance stops
- Transportation is difficult
- Asking for help can engender embarrassment
- The deafblind person can become more isolated, withdrawn and grief-stricken.

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Vocational
(Ilene Miner)

- Accommodation only goes so far
- People leave/lose their jobs early
- Further loss of self esteem, identity, roles
- Job retraining– perhaps with less remuneration
- Fewer people work as their vision deteriorates
- 82% unemployment among deafblind adults

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On-going Grief
(Ilene Miner)

- Usher isn't the same disease every day or year
- Progression requires ongoing repeated adaptation and change
- Issues change.. New solutions are always necessary

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Now for the good stuff…

- The world is changing.
- There is hope!

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Truth about diagnosis

- We don’t know the normal progression of vision loss in Usher syndrome
- Deaf is not deaf
  - Cochlear implants/hearing aids
- Blind is not blind
  - 50% of people with Usher can still read a newspaper at age 50
  - What if we can slow the progression?

Treatments Today

- Vitamin A
  - High doses of vitamin A can slow the degeneration associated with retinitis pigmentosa.
- DHA/Lutein
  - Combined with vitamin A can further slow the degeneration associate with retinitis pigmentosa.
- Sunglasses and hats
  - In process study appears to show link between bright light and onset of retinitis pigmentosa in Usher mice.

The Impact on Research

- *"The future influences the present just as much as the past." – Friedrich Nietzsche*
- People who fear the future disengage
- Slows research, ensure the future they fear
- 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.

The Coalition for Usher Syndrome Research Engages Families

- Working to build a global Usher syndrome community
- Registry with families from 23 different countries
  - Spanish, English, Hebrew (soon), Dutch and French (in development)
- Family network that connects hundreds of families globally
- Annual family conferences
- Monthly conference calls
- Daily communication on Usher syndrome issues and research
- Advocacy efforts to increase federal funding for Usher syndrome research

The Coalition for Usher Syndrome Research

The Real Hope

- People with Usher have athletic success
- People with Usher syndrome go to college
- People with Usher syndrome fall in love and get married
- People with Usher syndrome have children
- People with Usher syndrome have successful careers

The Coalition for Usher Syndrome Research

Get Engaged

- Join the family network
- Join the Usher syndrome registry
  - www.usher-registry.org
- Join our monthly conference calls
  - Captioned, notes available afterward
- Follow our blog on our web site
  - www.usher-syndrome.org
  - *"The most thoughtful exploration of Usher Syndrome that I've ever seen on the internet." – Reader Comment*
- Follow us on Facebook and Twitter

Come to the International Symposium on Usher Syndrome

- Harvard Medical School, Boston, Massachusetts
  - July 10-12, 2014
  - Combined family/science conference
  - Meet the leading Usher syndrome experts from around the world

The Coalition for Usher Syndrome Research

Thank You

- Registry: www.usher-registry.org
- Web site: www.usher-syndrome.org
- m.dunning@lek.com

The Coalition for Usher Syndrome Research