Hope and Building an Usher Syndrome Community

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Bella’s Story

• 14 years old
• Born deaf
• Diagnosed with Usher at age 8
• Loves horses
• Grudgingly loves her little brother, Jack
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 bilateral 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...
Hearing Loss

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

- Degenerative condition of retinal cells.
- Rod cells: vision in low light.
- Cone cells: color and acuity.
- Nightblindness
- Peripheral vision
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.
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!
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
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• The deafblind person can become more isolated, withdrawn and grief-stricken.
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
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
Now for the good stuff...

• The world is changing.
• There is hope!
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 associated 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 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
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
Thank You

• Registry:  www.usher-registry.org
• Web site:  www.usher-syndrome.org
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