Usher Syndrome Coalition

Nancy O’Donnell
Director of USH Trust Registry
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West Virginia
My Background

• Many years working at HKNC with adults who are deaf-blind
• Volunteer, intern, teacher, administrator
• Usher Syndrome Coalition since 2015
Usher Syndrome Coalition

Who we Are

Identification of Individuals with Usher Syndrome
Usher Syndrome Coalition

• Exclusively focus on Usher syndrome, worldwide
• All ages, all types of Usher syndrome
• Provide information, resources and support
• Bridge between research community and Usher community
Goals for Webinar

- Overview of Usher syndrome
- A look at the numbers
- Individuals you serve
- Service challenges
- Resources through the Usher Syndrome Coalition
Overview of Usher syndrome
Usher Identified

• 1858 - First described by German ophthalmologist
• 1914 - Charles Usher, British ophthalmologist studied 69 individuals
Usher syndrome

- Congenital bilateral sensorineural hearing loss (SNL)
- Retinitis pigmentosa (RP)
- Vestibular issues
FAQs

• Most common genetic cause of combined vision and hearing loss
• 3 major types - About 10 subtypes
• Affects males and females
Usher syndrome Type 1

• USH 1B, 1C, 1D, 1E, 1F, 1G;
• Profound hearing loss (deaf);
• Early onset RP (first decade of life);
• Vestibular (balance) problems.
Usher syndrome Type 2

- USH 2A (most common), 2B (rare), 2C, 2D
- Moderate to severe non progressive high freq hearing loss
- RP evident in teen years, but kids adapt well
- No balance problems
Usher syndrome Type 3

- Type 3a
- Progressive hearing and vision loss;
- Progressive balance loss in 50% of people with USH3;
- Rare;
- More common in Finland and Ashkenazi Jews.
Atypical Usher syndrome

The Usher gene is present, but

• symptoms do not fully express OR
• symptoms are absent
• Cases of nonsyndromic deafness have been linked to mutations in the Usher 1B, 1C, 1D, 1F and 2D genes (Ahmed et al., 2002, Bork et al., 2001, Liu et al., 1997b, Liu et al., 1997c, Mburu et al., 2003 and Weil et al., 1997).

• Conversely, mutations in the Usher 2A and 3 genes can cause autosomal recessive RP without reported hearing loss (Rivolta et al., 2000, Seyedahmadi et al., 2004a and Seyedahmadi et al., 2004b).
A Look at the Numbers
Usher Estimates by Prevalence

• Range of estimates:
  • Conservative: 3%-6% of children with congenital bilateral SNHL
  • Inclusive: 8-10% of children with congenital bilateral SNHL
Congenitally Deaf/HOH Children

• CDC – 4,000,000 babies born annually (2015)
• 3/1000 born with congenital bilateral SNHL=12,000/year
• 21 years (school age) x 12,000 = 252,000 congenital D/HOH youth
# Children with Usher, 0-21

• Based on 252,000 congenital D/HOH youth, 0-21.
• Estimates range 3-10% of D/HOH kids have Usher:
  • 3% = 7560
  • 6% = 15,120
  • 8% = 20,160
  • 10% - 25,200
Usher estimates by births/year

- Researchers estimate 20-50,000 with Usher in USA
- Usher is not due to cycles of virus or environmental factors
- Should be consistent % of births/year
- Life expectancy = 80
- 20,000 divided by 80 = 250 Usher births/year
- 50,000 divided by 80 = 625 Usher births/year
Employment age adults with Usher

- Employment age – 18-62
- $250 \times 44 = 11,000$ employment age adults with Usher
- $625 \times 44 = 27,500$ employment age adults with Usher
What We Know - NCDB

• Mark Shalock – NCDB demographics
• https://nationaldb.org/library/page/2199
2015 Deafblind Child Count

• NCDB 2015 – 10671 all deafblind children served by states
• 295 with Usher reported nationally
  • Actual # eligible to receive services in 2015 = 352
  • Slight increase over past 6 years
  • Largest increase Usher 1
• 2 in WV
What We Know from USH Trust

• 1252 registered worldwide
• 828 in the USA
• 5 adults with Usher registered in WV
• 0 children with Usher registered in WV
Where is everyone??

• Most medical professionals don’t know about Usher syndrome
• Adults with Usher don’t know anyone else with Usher
• Many are unaware of resources:
  • Usher Coalition: www.usher-syndrome.org; www.usher-registry.org
  • NCDB: www.nationaldb.org
  • State deafblind project: WV http://wvde.state.wv.us/osp/deafblindresources.html
  • HKNC – regional rep Molly Sinanan – molly.sinanan@hknc.org
Usher Syndrome Coalition
Resources
What is the USH Trust Registry?

• USH Trust registry - www.usher-registry.org
  • Researchers can’t find enough people with Usher syndrome to participate in clinical trials
  • Our goal – be the bridge between research community and Usher community.
  • Identify everyone in the world with Usher syndrome
  • All ages/types of Usher
USH Trust Registry’s potential

• Inform community of research/treatments and cure
• Conduct secure surveys – employment; social; communication; technology;
• Identify (or debunk) co-morbidities;
• Help to define Usher syndrome;
Current USH Research

- Usher 1B: UshStat® treatment is designed to halt vision loss in people affected with Usher syndrome type 1B, which is caused by defects in the MY07A gene. Retinitis Pigmentosa - entering a Phase II clinical trial
- Drug therapies – which could potentially help restore hearing in certain types of Usher syndrome.
Unraveling USH

• Collaboration with Project Usher/Stephen A. Wynn Institute
  • Genetic testing
  • Definitive diagnosis needed for participation in studies, treatment
  • Financial assistance available
• Website - Step by step process, letters and forms at: https://www.usher-syndrome.org/take-action/join-the-family/family-network.html
Other Resources

• USH Talks - short webcasts about Usher and related topics


• USH Blue Book online forum

Global Awareness

• Social Media
  • Facebook: https://www.facebook.com/UsherSyndromeCoalition
  • Twitter: @UsherCoalition

• Own the Equinox Campaign

• Young children and families stand to benefit the most
• Can’t benefit if they don’t know
• Cure RP within 10 years?
• Need genetic verification to participate
Usher Syndrome Coalition

USH Trust Registry: www.usher-registry.org
Web site: www.usher-syndrome.org
Krista Vasi, Executive Director: k.vasi@usher-syndrome.org
Mark Dunning, Board Chair, Founder and parent of Bella
Nancy O’Donnell: n.odonnell@usher-syndrome.org