Usher Syndrome Coalition 2017

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National Center on Deaf-Blindness

USHER SYNDROME COALITION

Where is Everybody?

Starting a National Dialogue on Finding Children with Usher Syndrome

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My Background

- Many years working with adults who are deaf-blind
- Part of the original Technical Assistance Center staff in 1984
- Worked for DB-LINK as Information Specialist
- Usher Syndrome Coalition since 2015

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Usher Syndrome Coalition

- Exclusively focus on Usher syndrome
- International, all ages, all types of Usher syndrome
- Provide information, resources and support
- Bridge between research community and Usher community

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Goals for Webinar

- Overview of Usher syndrome
- A look at the numbers
- Resources through the Usher Syndrome Coalition

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Overview of Usher syndrome

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Usher Identified

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

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Usher syndrome

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

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FAQs

- Most common genetic cause of combined vision and hearing loss
- Recessive; affects males and females
- 3 major subtypes

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Usher syndrome Type 1

- Profound hearing loss (deaf)
- Early onset RP (first decade of life)
- Vestibular (balance) problems
- USH 1B, 1C, 1D, 1E, 1F, 1G

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Usher syndrome Type 2

- Moderate to severe hearing loss (hard of hearing)
- RP evident in teen years
- No balance problems
- USH 2A, 2C, 2D

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Usher syndrome Type 3

- Progressive hearing and vision loss.
- Progressive balance loss.
- Rare - more common in Finland and Ashkenazi Jews
- USH 3A

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Atypical Usher syndrome

- Doesn’t fit any of the first three categories:
  - symptoms do not fully express OR
  - symptoms are absent

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A Look at the Numbers

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Prevalence of Usher

- Conservative: 3%-6% of children with congenital bilateral SNHL
- More likely: 8-10% of children with congenital bilateral SNHL

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Congenitally Deaf/HOH Children

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

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Children with Usher, 0-21

- 252,000 congenital D/HOH youth, 0-21
  - 3% = 7560
  - 6% = 15,120
  - 8% = 20,160
  - 10% - 25,200

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Another Way to Look at it

- Researchers estimate 50,000 with Usher in USA
- Life expectancy – 80
- 50,000 divided by 80 = 625 Usher births/year
- 625 x 21 = 13,125 children with Usher

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What We Have

- USH Trust – 180 children 0-21
- NCDB 2015 - 295 with Usher
  - Actual # eligible to receive services in 2015 = 352
  - Slight increase over past 6 years
  - Largest increase Usher 1

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The BIG question

Where are the

THOUSANDS

of children with Usher syndrome in the US?

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Perhaps…

- Not yet identified with Usher
  - Medical community unaware of Usher
  - Hearing loss identified; retinitis pigmentosa (RP) not on radar
- Identified and living well
  - Cochlear implant or hearing aid; vision still very good
  - Don’t see a reason to identify formally through program or registry

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Or…

- Don’t know that anyone else with Usher is out there
- Unaware of resources:
  - Usher Coalition
  - NCDB/state projects
  - HKNC

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Better Identification Could:

- Change the profile of deaf-blind child count (are we ready for that?)
- Provide supports to children and families
- Learn about Usher in its entirety – natural history
- Inform community about resources, treatments, trials, cure

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Identification Efforts

- Newborn hearing screenings great
- Medical profession still unaware of Usher
    - Pediatric ENT “doesn’t matter where hearing loss came from”
- Statewide screenings at schools for the deaf
    - What happened to that data
    - Are any states continuing that practice?

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Usher Syndrome Coalition Resources

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USH Trust Registry

- USH Trust registry
  - Only Usher syndrome
  - Worldwide
  - Inclusive of all ages/types
  - Multiple languages/one database
  - www.usher-registry.org

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Unraveling USH

- Collaboration with Project Usher/Stephen A. Wynn Institute
  - Genetic testing
  - Definitive diagnosis needed for participation in studies, treatment, if desired
- 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

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Other Resources

- USH Talks - short webcasts about Usher and related topics https://www.usher-syndrome.org/what-is-usher-syndrome/presentations/ush-talks-library.html
- USH Connections – annual conference/symposium (July) https://www.usher-syndrome.org/about-the-coalition/event-calendar.html/event/2017/07/15/9th-annual-ush-connections-conference/130502
- USH Blog – individual reflections https://www.usher-syndrome.org/our-story/ush-blog.html

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Global Awareness

- Social Media
  - Facebook: https://www.facebook.com/UsherSyndromeCoalition
  - Twitter: @UsherCoalition
- Own the Equinox Campaign

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Support. Treatments. Cure.

- 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

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We Agree!

"The opportunity for early identification, intervention and the provision of instructional services aimed at the unique needs of young children and students who are deaf-blind is a critical component that cannot be lost." ***NCDB 2015 Report***

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Thank You NCDB!

Linda, Sam, Gail and Robbin for the opportunity to present this webinar and continue to work together.

Mark Schalock providing awesome demographic details

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Stay Tuned!

NCDB will be presenting an USH Talk with an overview of services available for children with Usher syndrome and their families.

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Usher Syndrome Coalition Contacts

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