

# Usher Syndrome Coalition

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USHER SYNDROME  
COALITION

# 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

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# 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](http://www.usher-syndrome.org); [www.usher-registry.org](http://www.usher-registry.org)
  - NCDB: [www.nationaldb.org](http://www.nationaldb.org)
  - State deafblind project: WV <http://wvde.state.wv.us/osp/deafblindresources.html>
  - HKNC – regional rep Molly Sinanan – [molly.sinanan@hknc.org](mailto:molly.sinanan@hknc.org)

# Usher Syndrome Coalition Resources

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# What is the USH Trust Registry?

- USH Trust registry - [www.usher-registry.org](http://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<sup>®</sup> treatment is designed to halt vision loss in people affected with Usher syndrome type 1B, which is caused by defects in the MYO7A 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 <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 Blue Book online forum
- USH Blog – individual reflections <https://www.usher-syndrome.org/our-story/ush-blog.html>

# Global Awareness

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

# 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

# Usher Syndrome Coalition

USH Trust Registry: [www.usher-registry.org](http://www.usher-registry.org)

Web site: [www.usher-syndrome.org](http://www.usher-syndrome.org)

Krista Vasi, Executive Director: [k.vasi@usher-syndrome.org](mailto:k.vasi@usher-syndrome.org)

Mark Dunning, Board Chair, Founder and parent of Bella

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