**Let’s Chat: About Usher Syndrome**

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**My Background**

* Usher Syndrome Coalition – Director of USH Trust
* Ava’s Voice
* Molloy College
* HKNC

**Usher Syndrome Coalition**

* Usher Syndrome Coalition - Director of the USH Trust Registry since 2015
* Ava’s Voice
* Molloy College
* HKNC

**For the sake of consistency**

* Usher syndrome not Usher’s
* Unless used in a title or beginning of a sentence, “syndrome” is written with a lower case “s”
* Researchers refer to USH1B, USH2A, etc.
* Also described as Usher type 2 or Usher type II

**Presentation Outline**

* What is Usher syndrome
* Population Estimates
* Students with Usher Syndrome
* Ask the Experts
* Resources

**What is Usher Syndrome**

**Discovering Usher syndrome**

* 1858 - German ophthalmologist Albrecht von Graefe first described 3 deaf siblings with RP in a family of 5 children
* 1861 – Richard Liebreich - Berlin researcher identified pattern of deafness with RP in siblings and blood related marriages
* 1914 - Charles Usher, Scottish ophthalmologist put it all together when he studied 69 cases.

**Usher syndrome =**

* Most common genetic cause of combined vision and hearing loss
* A leading genetic cause of deafness
* Congenital bilateral sensorineural hearing loss (SNL)
* Adventitious vison loss - retinitis pigmentosa (RP)
* Vestibular issues
* No associated intellectual disability

**Types and Subtypes**

* 3 major types - 1, 2 and 3
* NO type 4
* 10 subtypes discovered so far
  + <https://hereditaryhearingloss.org/usher>

**Usher syndrome type 1**

* Profound hearing loss at birth (deaf); 1B, 1C, 1D, 1E, 1F
* No type 1A
* @ 40-45% of the Usher community
* Early onset RP (first decade of life);
* Vestibular (balance) problems
* French/Canadian/Acadian – 1C <https://www.youtube.com/watch?v=qCPTNS2d4ao>
* Ashkenazi Jews – 1F <https://www.youtube.com/watch?v=r_HnD1TaEts>

**Type 1 - Impact**

* + Late in sitting up, walking;
  + Balance may improve with athletics that build core muscles – horseback riding, swimming
  + Younger generation – early diagnosis
    - cochlear implants; function orally
    - When devices come off, profoundly deaf
    - In dark environments, functionally deafblind
  + Older generation – Culturally Deaf/ASL/Tactile ASL/Haptics/ProTactile
    - adults may walk like they’re “drunk”
    - giving up driving is a big hurdle

**Usher syndrome type 2**

* + USH 2A (most common), 2C, 2D
  + No type 2B
  + Moderate to severe non-progressive high frequency hearing loss
  + RP evident in teen years
  + No balance problems
  + Meet Brendan HKNC 2017 <https://www.youtube.com/watch?time_continue=82&v=2TaByN6spCE&feature=emb_logo>

**Type 2 Impact**

* + Function oral/aural and visual
  + Hearing aids; may get CI as they get older
  + Mainstream school with supports;
  + Can be “invisible”
  + Typical diagnosis of adults – 20s to 40s
  + Now, dx through **prenatal** genetic testing
  + Impacts careers when vision loss hits critical point

**Usher syndrome type 3**

* + Type 3A, 3B;
  + Rare;
  + Progressive hearing and vision loss;
  + Progressive balance loss in some with USH3;
  + More common in Finland and Ashkenazi Jews;
  + Rebecca Alexander: <https://www.youtube.com/watch?v=BbbivZBXECg>

**Type 3 - Impact**

* + Function oral/aural and visual
  + Hearing aids; may get CI as hearing loss progresses
  + Mainstream school with supports
  + Typical diagnosis of adults – 20s to 30s
  + Impacts careers when vision and hearing loss hits critical point

**Atypical Usher syndrome**

The Usher gene is present, but

* + symptoms do not fully express
    - Deaf with no vision loss
    - RP with no hearing loss
  + OR
  + symptoms are absent

**Deaf + RP may not equal Usher**

* People can be born with RP and acquire deafness from other causes – high fever, congenital rubella, encephalitis, ototoxicity,
* Combined vision and hearing loss can occur from other causes
* Only genetic testing can verify that you have Usher syndrome

**Population Estimates**

**A) Usher Estimates by Prevalence**

* Estimates from researchers vary:
  + Between 1/17000 or 1/6000 of USA population
  + Based on USA 2018 population of 327,200,000
    - 1/17,000 = 19,267
    - 1/6000 = 54,589

**B) Usher Estimates by %**

* Estimates based on deafness statistics (EHDI):
  + 3%-10% of children with congenital bilateral sensorineural hearing loss (SNHL)
    - Between 3200 to 11,000 kids, birth to 21

**C) 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
* Numbers fluctuate with birth rate
* Life expectancy = 80
* 20,000 divided by 80 = 250 Usher births/year
* 50,000 divided by 80 = 625 Usher births/year

**National Center on Deaf-Blindness  
2019 Child Count**

* 10,627 deafblind children on December 1, 2019
  + 383 with Usher, up from 345 in 2018; 29 in TX
  + Usher kids have 58% CIs as compared to the overall 11.7% in non-Usher kids
  + “This is not your father’s Buick” - Dramatic change in USH1 community of learners since CIs became an option

**Coalition’s USH Trust\***

* 1671 registered worldwide from 64 countries
* 1106 in the USA;
* 79 in TX (total should be between 1700 and 4800)
* Type 1 – 9; 1B - 5; 1C – 0; 1D – 2; 1F - 0
  + Type 2 -19; 2A – 19; 2C - 3
  + Type 3 - 5
* 17 – type unknown or not reported
* 39 report genetic confirmation

\*As of 2021.1

**Where is everyone??**

* Most medical professionals don’t know about Usher syndrome
* Children with Usher are mainstreamed or in local school with supports
  + Cochlear implants mitigate hearing loss
  + Their vision issues are not yet serious or dx
  + Children are very clever at adapting to night blindness or think it’s “normal”
  + No one has realized “vision loss + hearing loss may equal Usher syndrome” OR
  + Parents know but haven’t told the child yet
* Adults with Usher think they’re “the only one”
* Some cultures are protective of people with disabilities

**Students with Usher Syndrome**

**Usher is Invisible**

* No intellectual disability
* Historically under-identified due to lack of awareness of Usher and slow progressive nature of vision loss.
* Different from congenitally deafblind due to late onset of vision loss, minimal or no comorbidities
* MANY KIDS ARE THRIVING!

**Usher = Constant Change**

* Vision loss = progressive, unpredictable, fluctuating
* Strategies that worked yesterday may not work tomorrow, i.e.
  + - * Interpreters – distance, tracking, tactile, Haptics
      * Regular print, high contrast, larger font, or braille
      * As vision loss progresses, speech reading may be affected; can be misdiagnosed as additional hearing loss

**Communication is Diverse**

* Outreach and education must include many different methods of communication:
  + Spoken languages
  + Sign languages, tactile sign languages
  + Print, large print, braille
  + Web based - captions, image descriptions, text transcripts

**Educational Considerations (1)**

* Impact on learning, classroom setting, communication
  + Classroom set-up – preferential seating, ALDs, visual accessibility (no glare), adequate lighting, note takers, interpreters, large print, color contrast, loss of color differentiation
  + Situational blindness in dimly lit areas
  + May need “sensory breaks” similar to “hands down” for DB adults
  + Appear to be “typical deaf learners” – teachers think they’re “doing fine!”
* Interveners – what is their role with kids with Usher? Academic interveners?

**Educational Considerations (2)**

* Time for eyes to adjust to different lighting;
* May stop and scan room before entering;
* Braille – should it be introduced and when?
  + Based on English, implications for “English as a new language” learners;
* O&M – additional time to travel to classes, offices, meetings, cafeteria, etc.;
  + Use a cane? When to introduce?;
* The role of [Haptics](https://www.helenkeller.org/hknc/haptics-demonstration-videos) in teaching braille, O&M, etc.;
* Opportunities to balance work, socialization/play, incidental learning.

**Educational Considerations (3)**

* Technology: Helps to provide equity – CIs, ALDs, smartphones, laptops, tablets, equipment with Bluetooth connections;
* Need time to “set themselves up” in classrooms, preferential seating; locker near classes;
* What happens when a device malfunctions? Troubleshooting and Plan B;
* Mental, visual, and auditory fatigue

**Educational Considerations (4)**

* Simple visual or tactile environmental markers - locker, exits, key areas (benefits everyone!)
* How to get student’s attention, visually or tactually
  + Hand waving, foot stomping, light flashing
  + Physical touch on “neutral zone”
  + Never grab hands
  + Don’t play – “Guess who?”
* [Vibrating alerting systems](https://www.diglo.com/serene-innovations-centralalert-ca380-wearable-notification-system;sku=HC-CA380;s=334;p=HC-CA380%20%20Emergency%20evacuation)
* “X” = Exit now, no questions asked;
* Buddy system.

**Educational Considerations (5)**

* Accommodations alter *how* a student learns. They do not change **what** the student is expected to learn. Excellent webinar by parents of 2 proficient communicators with Usher syndrome
  + [**​**Educational Considerations for Students with Usher Syndrome](https://www.nationaldb.org/updates/2017-national-webinar-educational-considerations-for-students-with-usher-syndrome/)

**Educational Considerations (6)**

* Considerations for remote learning during COVID
  + - Compare features of remote platforms, i.e. Zoom, Google Meet, MS Teams
    - Integration of live captions - automatic? CART? Interpreter typing real time summary?
    - All videos captioned
    - Full and accurate text transcript
    - Adequate visual access to signs/interpreter - Internet speed, quality of camera, screen size of device, pinning key communicators
    - Permission to record sessions for review later
    - Adequate visual access to teacher/students for speech reading
    - Chat box visually accessible?
    - Q&A visually accessible?
    - Hand raise visually accessible?
    - Images/graphics clear with high contrast and alt text?
    - Solid contrasting background behind teacher to reduce
      * glare and visual noise?

**Resources from the Experts**

**Learn from Families**

* NFADB Board Member, USH Ambassador and TX Usher Mom, Nilam Agrawal
* [Ava’s Cheat Sheet Template and Sample](https://avasvoice.org/ushresources/)
* [Lane of Inquiry](https://www.laneofinquiry.org/)
* [Usher Syndrome Coalition - Just for Parents](https://www.usher-syndrome.org/our-story/just-for-parents.html)
* [Article - *Denied an IEP or 504 Because Grades are Good*](https://www.specialmomadvocate.com/denied-an-iep/?fbclid=IwAR2rGKcwIP25amfphB5hiM5eCQWzuFBecznWEU9HGe-Wlzpq6hmw1ZMQXJY)
* [Student Led IEPs](https://www.learnwithtworivers.org/student-led-ieps.html)

**Connect Families**

Yearly [USH Connections Conference](https://www.usher-syndrome.org/our-story/ush-connections-conference-summaries.html)

* + [Ava’s Voice: The Diagnosis](https://vimeo.com/435083123/1c2c27d533?fbclid=IwAR3iA3XxfH1RNXd7HP6EW9_1sS7RCgC3XaE_W0Fj4Qfeb-NzJQm86Mj8B1M) – interviews with kids with Usher and their parents
  + [discUSH Family Weekend](https://avasvoice.org/discush/) - for families of kids with Usher up to age 10
  + Facebook Groups
  + [F2FC](mailto:avasvoice@gmail.com) - monthly calls for Usher parents
* [NFADB](https://www.nfadb.org) - FB, groups, activities

**Learn from Youth**

* Young adults tell us: “Ask US what we need!”
* “Not everything is about Usher. We can also have learning disabilities, mental health issues, dyslexia, etc.”
* “We want to learn how to advocate for ourselves.”
* “We are eager to meet others with Usher.”
* Survey or interview graduates of your program - 5, 10 years later

**Connect Youth**

* [USHangouts](https://avasvoice.org/ushangouts/) – camp for all kids attending the USC conference
* Video: [Advocacy in the DeafBlind World](https://www.helenkeller.org/hks/introducing-advocacy-deaf-blind-world)– Ryan Odland, HKNC
* [USHthis Summer Camp for Youth with Usher Syndrome](https://avasvoice.org/ushthis/), ages 11-17
* [USHthis Summer Camp Counselor](https://avasvoice.org/ushthis-staff/) opportunities, ages 18-24

**Connect Youth (2)**

* Just for Young Adults section of the website: <https://www.usher-syndrome.org/our-story/just-for-young-adults/>
* Young Adult Blog – Seeing is Believing, Megan Lengel <https://www.usher-syndrome.org/our-story/just-for-young-adults/megan-lengel.html>
* Young Adults Living with Usher Syndrome (18-28) private FB page: <https://www.facebook.com/groups/2305692923042666>
* Monthly Zoom Calls of FB group members

**Learn from Adults**

* Tim Chambers**,** master painter with USH2A recently painted this series showing how RP impacts vision and color perception. You can learn more on his website in [the USH View](https://timothychambers.com/the-ush-view/). How might this influence your work with students?

**Connect with Adults**

* Task Forces
* Committees
* Consultants
* Staff
* Mentors
* Linguistic Models
* PT and Haptics trainers

**Learn from the Community**

* + [Professionals Serving DeafBlind Consumers](https://groups.google.com/u/1/g/psdbc/about) (PSDBC forum)
    - Information on the evolving culture, values of the community
  + [Blind but Not Blind](https://www.youtube.com/watch?v=6uxZ4u6nses) – JennyLynn Dietrich, DeafBlind adult on autonomy
  + [Protactile ASL – A New Language for the DeafBlind](https://www.youtube.com/watch?v=9GrK3P15TYU)
  + [DeafBlind BIPOC Panel on access to interpreters and impact of COVID on the community,](https://www.eventbrite.com/e/bipoc-deafblind-panel-tickets-135222571351?fbclid=IwAR1DNuKrLFusF2uvO54NhX7ygXgOwJKbINRO1pnJopZ27m6UPmzn_RFxY5Q) January 16, 2021, 1-3 pm EST

**Resources from the Coalition**

**Find the Community**

* + USH Trust registry - [www.usher-registry.org](http://www.usher-registry.org/)
    - Close the gap – 1600 vs 400,000 worldwide
    - Be the bridge between research community and Usher community.
    - Identify everyone in the world with Usher syndrome
    - Educate and connect the community

**Build the Local Community**

* USH Ambassadors - building the community in their state or country <https://www.usher-syndrome.org/our-story/ambassadors.html>
  + Represent the Coalition in their state/country
  + Reach out to organizations, agencies and individuals
  + Must have a connection to Usher syndrome
  + United States plus India, Canada, Dominican Republic, France, Mexico…and growing

**Connect the Community**

* USH Connections – [annual conference/symposium](https://www.usher-syndrome.org/our-story/ush-connections-conference-summaries.html)
* [USH Blue Book online forum](https://www.usher-registry.org/en/network_members/new) – for individuals with Usher syndrome.

**Educate the Community**

* [First ever series about Usher in ASL](https://www.usher-syndrome.org/what-is-usher-syndrome/about-usher-syndrome-in-asl.html)
* [USH Talks -](https://www.usher-syndrome.org/what-is-usher-syndrome/presentations/ush-talks-library.html) short webcasts about Usher and related topics
* [USH Blog](https://www.usher-syndrome.org/our-story/ush-blog.html) – individual reflections
* [Free Genetic testing resources](https://www.usher-syndrome.org/what-is-usher-syndrome/get-genetic-testing.html)
* Facebook posts on science and research

**Partner with the Community**

* [Ava’s Voice](http://www.avasvoice.org/) – [USHthis camp](https://www.facebook.com/177853542264934/videos/744152806016733)
  + [USHangouts](https://avasvoice.org/ushthis/); [discUSH Family Weekends](https://avasvoice.org/discush/); [educational materials](https://avasvoice.org/ushresources/)
  + [Usher Syndrome Society](http://ushersyndromesociety.org) – photo journalism, [Sense Stories](https://www.ushersyndromesociety.org/sense-stories/)
  + [HearSeeHope](http://www.hearseehope.com/) – awareness and funding research
  + [Usher 1F Collaborative](http://usher1f.org/) – natural history study, scientific conference
  + [Usher KidsUK-](http://usherkidsuk.com) bringing families together in UK

**Collaborate with the Community**

* Coordinate with related organizations
  + [NCDB](http://www.nationaldb.org/), [NFADB,](http://www.nfadb.org/) [HKNC](https://www.helenkeller.org/hknc/), [State DB Projects](https://www.nationaldb.org/state-deaf-blind-projects/)
    - Proficient Communicators work group
    - Building Family Partnerships
    - Deaf-Blind Resource Group
    - HKNC Community of Practice

**Raise Global Awareness**

* International Usher Syndrome Awareness Day – 3rd Saturday of September
* Social Media
  + [Facebook](https://www.facebook.com/UsherSyndromeCoalition)
  + Twitter: @UsherCoalition

**Usher Syndrome Coalition**

* USH Trust Registry: [www.usher-registry.org](http://www.usher-registry.org/)
* Website: [www.usher-syndrome.org](http://www.usher-syndrome.org/)
* Krista Vasi, Executive Director
* Lane McKittrick, Board Chair
* Julia Dunning, Program Coordinator
* Nancy O’Donnell, Director of Outreach: [n.odonnell@usher-syndrome.org](mailto:n.odonnell@usher-syndrome.org)
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