ICD-10-CM Code Request for
Usher Syndrome

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Where the world comes for answers

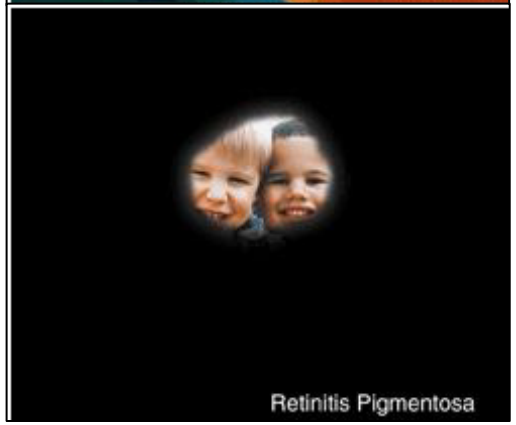
USHER 1F COLLABORATIVE

usher syndrome society

USHER SYNDROME COALITION

Usher Syndrome (USH)

- **Recessive genetic disorder responsible for 50% of hereditary deaf-blindness**
  - Congenital sensorineural hearing loss
  - Retinitis pigmentosa
  - Vestibular dysfunction (balance issues) in some patients
- **Three clinically identifiable sub-types encompass most patients**
- **Genetic causation is well-defined:**
  - 9 causative genes identified to date that are reliably associated with specific clinical sub-types
  - Combined estimated incidence of 4-17:100,000 resulting in approximately 400,000 affected worldwide[1]
- **Diagnosis based upon:**
  - Hearing screening
  - Ophthalmic evaluation including electroretinography (ERG), visual field assessment, and retinal imaging
  - Genetic testing identifying pathogenic variants in one of 12 genes



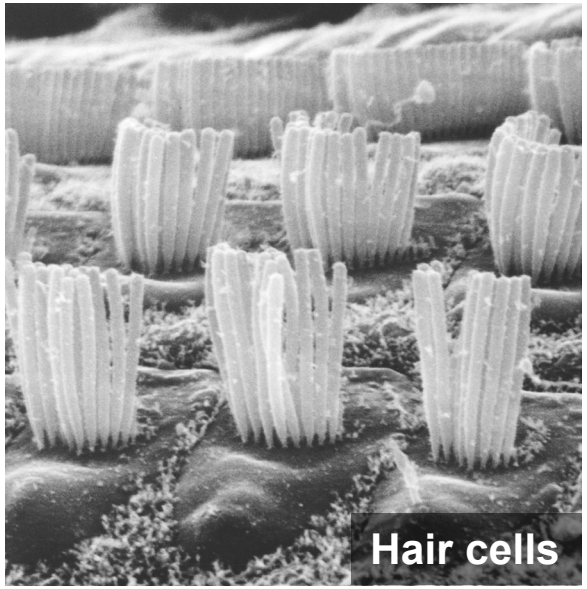
[1]https://www.nidcd.nih.gov/health/usher-syndrome

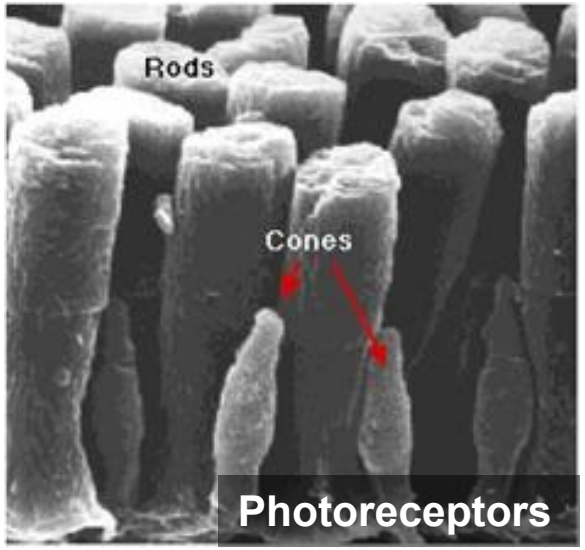
USH Pathophysiology

Causative genes encode proteins critical for function of primary sensory cells needed for normal hearing, vision, and balance

- **Inner ear**
  - USH genes needed for the development and function of hair cells
  - Hair cells are primary sensory receptors for hearing: Encode mechanical stimulation into electrical signals
  - Abnormalities of hair cells can also impact balance and vestibular function
- **Retina**
  - USH genes needed for the development and function of rod and cone photoreceptors
  - Rods and cones are primary sensory receptors for vision: Encode photic stimulation into electrical signals

Dysfunction and eventual degeneration of these cells results in clinical manifestations of USH





https://medlineplus.gov/genetics/condition/usher-syndrome/#causes; https://lab.research.sickkids.ca/harrison/electron-microscopy/#; https://www.chm.bris.ac.uk/webprojects2003/white/functional_anatomy_of.htm

USH sub-types: High clinical & genetic correspondence

Phenotype	Type 1	Type 2	Type 3
Hearing loss	Profound hearing loss or deafness at birth.	Moderate to severe hearing loss at birth.	Progressive hearing loss in childhood or early teens.
Vision	Decreased night vision in childhood with severe vision loss by midlife	Decreased night vision by adolescence with severe vision loss by midlife.	Varied in severity and age of onset; often begin in teens with severe vision loss by midlife.
Balance (vestibular function)	Balance problems from birth.	Normal balance.	Normal to near-normal balance in childhood; chance of later problems.

USH subtype	Gene
1b	*MYO7A*
1c	*USH1C*
1d	*CDH23*
1f	*PCDH15*
1g	*USH1G*
2a	*USH2A*
2c	*VLGR1*
2d	*WHRN*
3a	*USH3A*

*** USH1 and USH2 account for 95% of Usher cases in the United States**

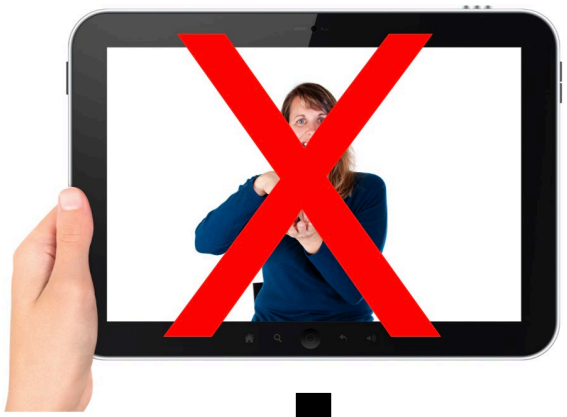
https://www.nidcd.nih.gov/health/usher-syndrome; https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4511090/; Delmaghani and El-Amraoui, Hum Genet, 2022; 141(3-4): 709-735

Why dual and multisensory impairment is different

- **Deaf and hearing-impaired individuals use vision to aid communication**
  - Lip reading
  - Sign language
- **Blind and visually-impaired use their hearing to localize sound:**
  - Orientation in space
  - To listen to changes in surface when using a white cane
  - To listen for changes in traffic patterns when crossing streets
- **Individuals with vestibular dysfunction use vision to aid balance**

In the medical environment, patient management of those with dual or multisensory impairment is more complex than managing the individual symptoms.

Example of communication challenges in clinical setting



Deaf USH patient unable to see ASL interpreter; tactile ASL interpreter required

ICD-10 codes currently used for USH

Codes cover vision, hearing, and balance issues associated with USH but are not specific for USH or useful to identify USH patients to aid in proper management of dual/multisensory impairments and research.

VISION-RELATED CODES

Code	Description
H35.5	Hereditary retinal dystrophy
H35.53	Other dystrophies primarily involving the sensory retina
H35.52	Pigmentary retinal dystrophy

HEARING-RELATED CODES

Code	Description
H91.93	Unspecified hearing loss, bilateral

OTHER

Code	Description
Q87.89	Other specified congenital malformation syndromes

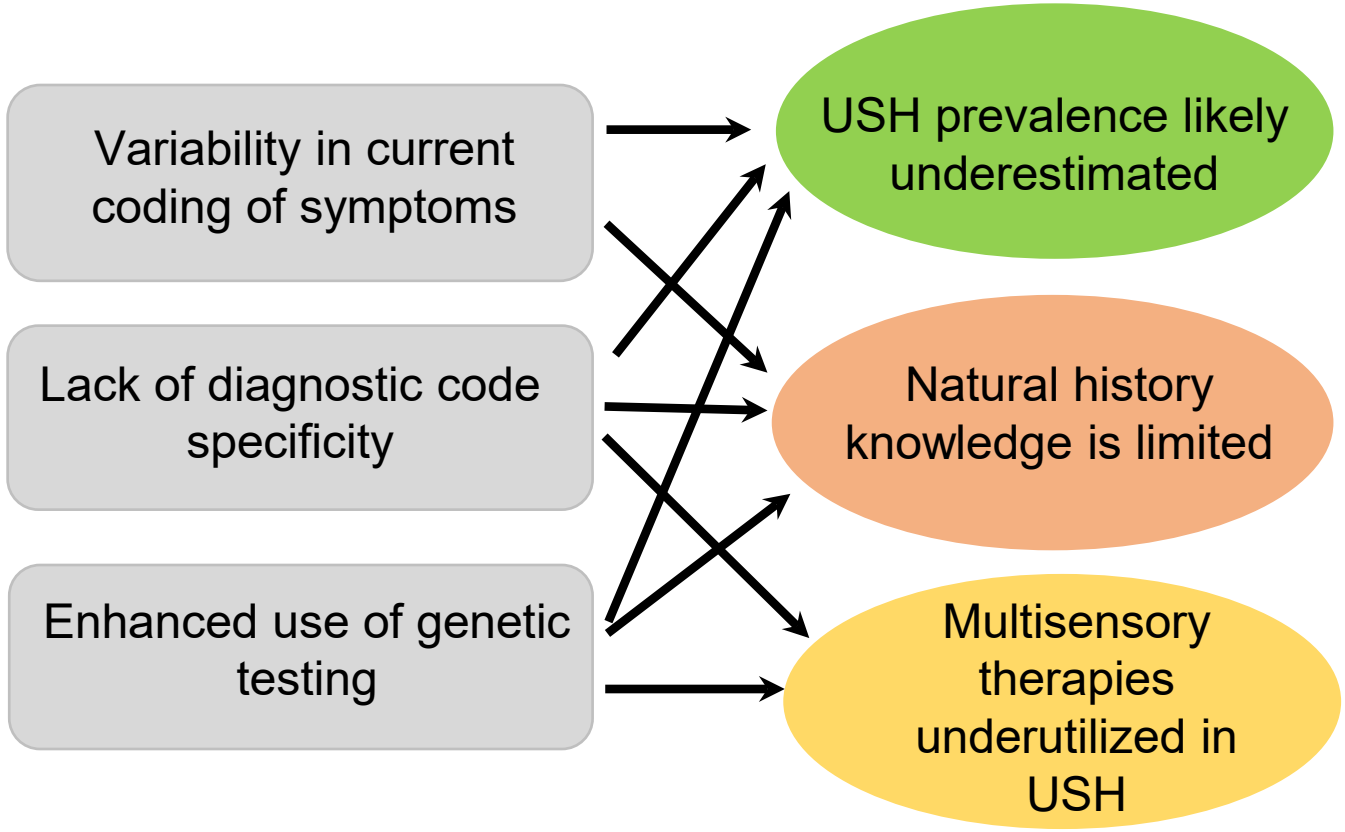
Selection of "Usher syndrome" and even genetically-specific forms of USH in the diagnosis search tool of the most-commonly used electronic medical record pulls individual codes for vision and hearing without specific unifier

Rationale for a New ICD-10-CM Code for USH

Absence of specific code

- **Prevents** precise documentation of subtype
- **Constrains** the path to effective treatments and, thus, patient welfare
- **Limits** meaningful clinical and policy research efforts

GOALS

- Enable more **accurate quantification of the prevalence** of USH
- Facilitate **better definition of the natural history** of phenotypes of USH to provide better anticipatory guidance and monitoring for complications of dual/multisensory impairment
- **Promote identification** of USH patients most likely to benefit from emerging therapeutics and/or modifications of symptom management

Rationale for a New ICD-10-CM Code for USH (Cont'd)