Usher Syndrome

often abbreviated as USH

Main Features

- Retinitis Pigmentosa
- Sensorineural hearing loss
- Vestibular disturbances

Eye Findings

- Retinitis Pigmentosa
    - Pigmentary retinopathy with degeneration of the RPE
    - Optic disc pallor
    - Retinal arteriolar attenuation
    - Symptoms
        - Decreased visual acuity
        - Nyctalopia
        - Peripheral visual field deterioration

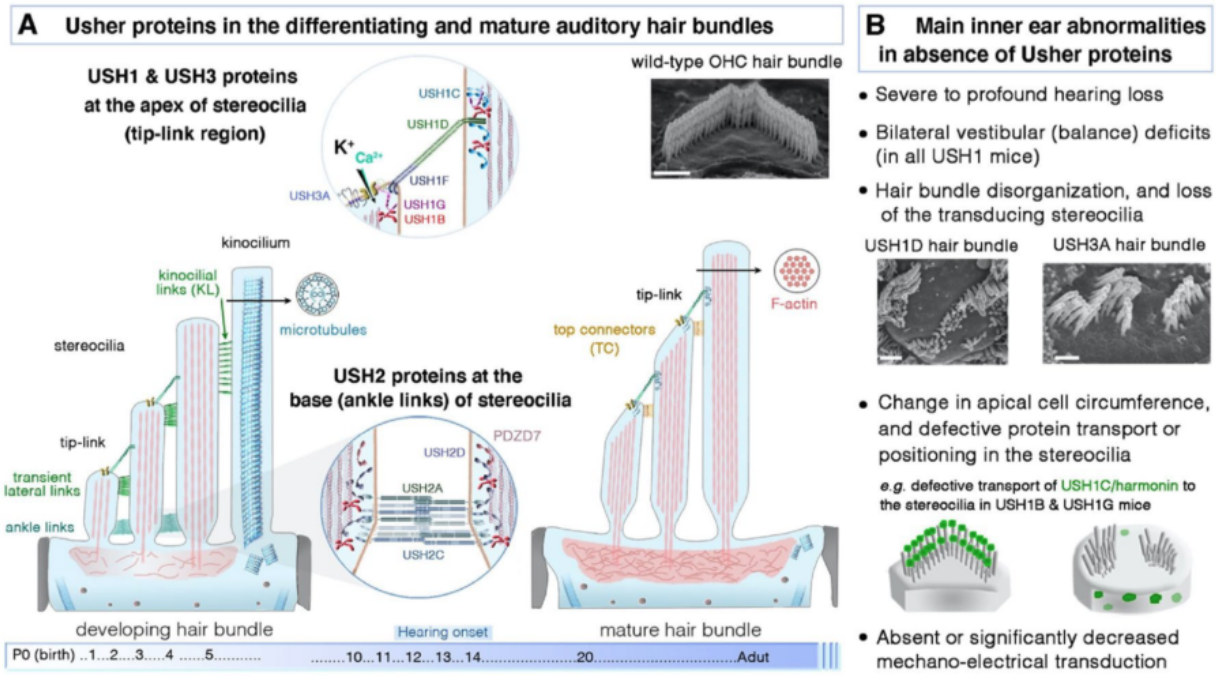
Other Findings

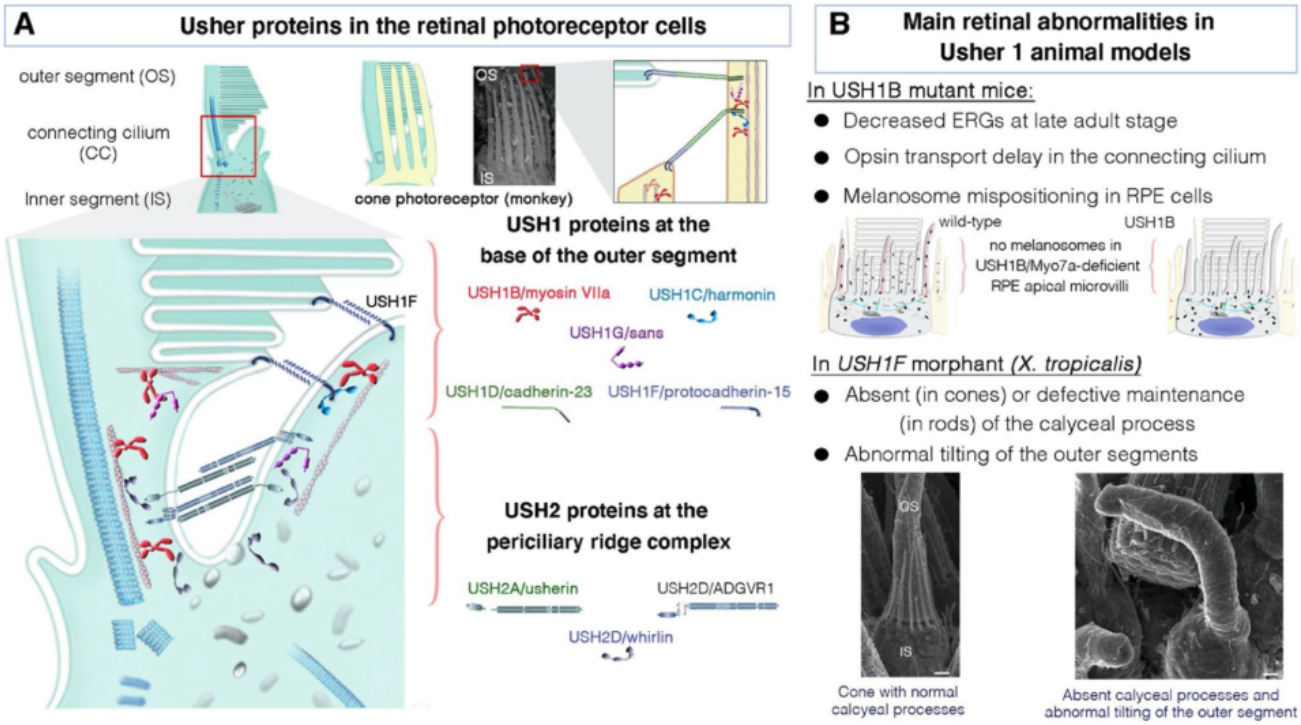
- Progressive retinal degeneration, course depends on the gene affected
    - USH type 1 = profound congenital hearing loss and early vision loss (most severe)
    - USH type 2 = RP by the second decade, moderate to severe congenital hearing loss and no vestibular abnormalities (most severe)
    - USH type 3 = Progressive and variable hearing loss, RP and vestibular abnormalities
    - Significant overlap among subtypes

Etiology

- Autosomal Recessive inheritance
- Included in the category of ciliopathy
    - abnormal formation of cilia
- Major genes affected
    - USH1 genes
        - MYO7A, USH1C, PDCH15, CDH23, USH1G
    - USH2 genes
        - USH2A, ADGRV1, WHRN

    - USH3 gene- CLRN1
- Other genes implicated but need classification
- Usher genes encode for proteins expressed in the inner ear and retina where they provide essential functions for the development of sensory hair cells and photoreceptor maintenance





From Resource (1)

Epidemiology

- Prevalence
  - Worldwide 4-17 per 100,000
  - USA 4.4 per 100,000
- Most common cause for hereditary deafness and blindness
  - 5% of all congenital deafness
  - 18% of retinitis pigmentosa cases

Resources

1. Delmanghani S et al. The genetic and phenotypic landscapes of Usher syndrome: from disease mechanisms to a new classification. Human Genetics 2022;141:709-735

syndrome