

USHER SYNDROME TYPE 1

Defining Characteristics & Research Significance

Usher syndrome is a rare genetic condition characterized by partial or total hearing loss and vision loss that worsens over time due to Retinitis Pigmentosa. Usher syndrome is the most common form of combined deaf-blindness. There are 3 types – type 1, 2 and 3 – categorized by severity of hearing loss and speed of vision loss. Type 1 is the most severe; nonfunctioning vestibular system, profound hearing loss at birth and significant and rapid progression of vision loss in childhood.

PROFOUND CONGENITAL DEAFNESS



Ush type 1 is associated with profound hearing loss at birth. Hearing aids will provide no benefit; the only option for "hearing" are cochlear implants.

Why is this important to understand: Future genetic treatments for restoring hearing are not an option for those with cochlear implants because of the structural damage done to the cochlea during implantation.

RAPID ONSET OF VISION LOSS



The speed at which vision loss progresses is faster than other types. Individuals with USH1 mutations reach legal blindness on average 15 years earlier than other types. Significant vision loss often happens prepubescent, starting with a complete loss of night vision between the ages of 3-5 and accelerated peripheral vision loss due to Retinitis Pigmentosa.

Why is this important to understand: The therapeutic window for finding treatments to preserve functioning vision is significantly smaller than other types, meaning there is less time to find treatments for those with Ush1.

NONFUNCTIONING VESTIBULAR SYSTEM



Ush type1 is also characterized and associated with severe balance issues due to a nonfunctioning vestibular system. Those with Ush type1 are late to walk, run and jump. Balance issues persist throughout life affecting individuals overall mobility.

Why is this important to understand: Epidemiology studies and natural history studies look at the entire impact of the disorder on the individual and society. In order to receive more funding and design treatments that account for overall accessibility it is critical to understand the entire impact on the individual's quality of life.