

Natural History of Hearing and Vestibular Function in Pediatric USH1C: Timing of Diagnosis, Cochlear Implantation, and Clinical Outcomes

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Background: Usher syndrome type 1C (USH1C) causes congenital severe-to-profound sensorineural hearing loss (SNHL), vestibular dysfunction, and progressive retinal degeneration. Although universal newborn hearing screening (UNHS) facilitates early identification of congenital hearing loss, the timing of SNHL diagnosis and intervention and subsequent auditory and vestibular outcomes, are unknown in pediatric USH1C. We aimed to define the earliest clinical trajectory to inform early definitive diagnosis, intervention, multidisciplinary management, and family counseling.

Methods: A retrospective, longitudinal chart review was performed in pediatric patients with genetically confirmed USH1C from January 1997 to July 2026. Demographics, UNHS results, diagnostic audiologic testing, cochlear implantation (CI) history, audiometric outcomes, vestibular findings, and communication status were extracted. Pre-implantation testing included auditory brainstem response (ABR), otoacoustic emissions (OAE), and tympanometry. For patients with available data, time-to-diagnosis and -intervention were calculated. Post-CI outcomes were assessed using speech recognition thresholds (SRTs) and pure-tone averages (PTAs).

Results: Twenty-four of 25 pediatric patients (4–29 years old) with genetically confirmed USH1C identified from an ongoing retrospective natural history study had sufficient data. Mean age at genetic diagnosis was 7.5 ± 6.8 years ($n=24$). Participants were predominantly from Louisiana, USA (72%, $n=18/25$), and 56% were female (14/25). Among 17 patients with available UNHS data, all failed screening. Diagnostic testing data demonstrated profound SNHL in all patients, with significantly reduced or absent ABRs ($n=18$) and OAEs ($n=13$), with normal tympanometry ($n=12$). Mean time from detection (UNHS) to diagnosis (SNHL) to intervention (CI) was 0.47 ± 0.48 years (range, 0.03–1.72) and 0.92 ± 0.90 years (range, 0.30–4.19), respectively. CI was confirmed in 24 patients, with mean age at first implantation of 1.67 ± 1.11 years (range, 0.55–4.73 years). Fifty-four percent ($n=13/24$) of patients underwent simultaneous bilateral CI, whereas 46% ($n=11/24$) initially received unilateral implantation. Following implantation, mean SRTs were 19.68 ± 6.04 , 22.42 ± 5.92 , and 24.25 ± 9.48 dB HL for right, left, and bilateral listening conditions, respectively. Corresponding PTAs were 26.15 ± 7.14 , 26.79 ± 9.84 , and 25.66 ± 8.23 dB HL for right, left, and bilateral listening conditions. Most patients primarily used spoken language. Vestibular documentation was limited, although gross motor delays and abnormal vestibular evoked myogenic potential testing supported vestibular dysfunction in seven patients (29%).

Conclusions: Pediatric USH1C patients demonstrated congenital, profound SNHL detected through UNHS, however definitive diagnoses (SNHL, then USH1C) and CI occurred on variable timelines. Implanted patients achieved favorable audiometric outcomes and predominantly developed spoken-language communication. The intervals between newborn screening, SNHL diagnosis, and CI highlight opportunities to optimize early diagnostic and rehabilitative pathways, while limited vestibular documentation underscores the need for longitudinal vestibular assessment. These data provide benchmarks for counseling families, coordinating multidisciplinary care, and designing future USH1C therapeutic trials.