Usher Syndrome in Medicine

Nathaniel Glbson, Nikki Gillies, Michael Norman
Human Development Center

Introduction

Usher Syndrome is a clinically and genetically heterogeneous disorder characterized by congenital sensorineural hearing loss and progressive vision impairment due to retinitis pigmentosa. It is the **leading genetic cause of combined deaf-blindness worldwide**, with an estimated prevalence of 3 to 6 per 100,000 individuals [Kimberling et al., 2010]. Despite its significant impact on communication, mobility, and quality of life, **awareness of Usher Syndrome among healthcare providers remains limited**, particularly in general medical education and training programs [Pennings et al., 2004; Bonnet & El-Amraoui, 2012]. Early identification is critical for appropriate interventions such as cochlear implantation, low vision services, and genetic counseling, which can significantly improve patient outcomes. However, delays in diagnosis are common due to fragmented care between audiology, ophthalmology, and primary care, compounded by a lack of routine screening or syndromic suspicion in hearing-impaired children. Based on these patterns in the literature, we hypothesized that **medical students would demonstrate limited knowledge and recognition of Usher Syndrome**, while physicians may be familiar with the name but **lack formal training or direct clinical exposure** due to the rarity of the condition and its minimal inclusion in standard curricula.

Methods

A comprehensive mixed-methods questionnaire is currently being developed by the **LSU Health Sciences Center Human Development Center** research team, including **medical student Nathaniel Gibson** under the mentorship of **Dr. Nicky Gillies** and **Dr. Michael C. Norman**.
Instrument design and construct selection were guided by previously validated frameworks assessing clinician and parent awareness of sensory disorders, including Ayton et al. (2023) — which surveyed allied health professionals' knowledge of Usher Syndrome — and Day & Brice (2012) — which developed the Hearing Parents' Perceptions of Health Professionals' Advice Questionnaire.
The LSU instrument adapts these evidence-based domains to evaluate medical students' and physicians' awareness of Usher Syndrome, focusing on inheritance patterns, symptom recognition, multidisciplinary coordination, and referral behavior. The questionnaire is **under active development** and pilot testing is

Survey Highlights

Previous research shows that both healthcare professionals and caregivers often report limited familiarity with Usher Syndrome.

- **Ayton et al. (2023)** found that although most audiologists, optometrists, and orthoptists recognized Usher Syndrome as genetic, many lacked understanding of vestibular dysfunction and the roles of genetic counseling, speech pathology, and coordinated multidisciplinary care
- **Day & Brice (2012)** demonstrated that hearing parents of deaf or hard-of-hearing children frequently encounter inconsistent or incomplete professional guidance during the diagnostic process, underscoring the need for improved interprofessional communication and informed-choice education

These findings informed the LSU team's questionnaire domains and highlight the ongoing educational gap our developing instrument seeks to quantify within the medical community.

Data Heading 1

While LSU-specific results remain under review and are not yet public, the **findings from prior studies** offer a benchmark for understanding educational and awareness needs surrounding Usher Syndrome.

Ayton et al. (2023) emphasized that many clinicians lacked familiarity with **vestibular symptoms**, **vision loss progression**, and **the psychosocial impact** of Usher Syndrome. The authors concluded that **training in multidisciplinary recognition and referral** could significantly improve patient outcomes.
Similarly, **Day & Brice (2012)** demonstrated that parents valued clear, accessible, and empathetic guidance, but noted that **medical professionals often failed to provide comprehensive explanations of genetic testing or dual sensory loss**.

Together, these studies suggest that both providers and caregivers face **critical knowledge and communication gaps**, which the developing **LSU questionnaire** aims to measure and address through structured assessment of awareness, preparedness, and interprofessional coordination in medical education settings.

Data Heading 2

The LSU instrument builds upon the structure of the Ayton et al. (2023) clinician survey and the validated Hearing Parents' Perceptions of Health Professionals' Advice Questionnaire developed by Day & Brice (2012). Data collection for the LSU survey is still ongoing, and **new results are not yet available for release**. However, the referenced literature provides important context for expected outcomes.

In **Ayton et al. (2023)**, a cross-sectional online survey of 167 clinicians revealed that while most respondents (over 80%) recognized *Usher Syndrome* as a genetic condition, only **38% correctly identified vestibular dysfunction** as a key component, and fewer than half understood the **multisystem sensory implications**.
Although awareness was relatively strong among audiologists and optometrists, there were major **knowledge gaps in interdisciplinary collaboration**, such as when and how to refer patients for **genetic counseling, speech therapy, and mobility support**.

In contrast, **Day & Brice (2012)**, which assessed hearing parents' experiences with professional advice after diagnosis of hearing loss, found that **many parents received inconsistent or incomplete medical information**. The study highlighted **limited professional confidence** in discussing hereditary deafness and **few referrals to genetic specialists**, underscoring systemic communication challenges between medical professionals and families.

Conclusion

This project builds directly upon validated surveys assessing clinician and caregiver knowledge to create a new, LSU-based instrument tailored for medical learners and practicing physicians.

Current literature demonstrates persistent gaps in understanding of Usher Syndrome's genetic basis, sensory impacts, and multidisciplinary management.
Our questionnaire—still in development—aims to measure and ultimately bridge those gaps through targeted educational initiatives and interprofessional awareness efforts inspired by the frameworks of Ayton et al. (2023) and Day & Brice (2012).