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“Usher Syndrome in Medicine”

Background: Usher Syndrome is a rare genetic disorder characterized by combined hearing loss and progressive vision loss. While early recognition is critical for intervention and patient support, existing literature suggests that many medical students and physicians have limited awareness or clinical exposure to this condition. This study hypothesizes that, consistent with prior research, most medical students and even practicing physicians may be unfamiliar with the nuances of Usher Syndrome diagnosis and management.

Methods: We designed a comprehensive survey targeting physicians, incorporating validated and newly developed questions to assess recognition of Usher Syndrome inheritance patterns, clinical features, and management strategies. The survey also included items on demographics, prior training, and self-reported confidence in addressing genetic conditions. Distributed across diverse practice settings, it simultaneously served as an educational tool by offering respondents resources to enhance their awareness of Usher Syndrome.

Results: 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.

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).