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"Prospective visual and hearing clinical trials for patients with Usher syndrome"

Usher syndrome, the most prevalent genetic cause of combined deafness and blindness, manifests in three clinical sub-types categorized by the severity and age of onset of sensory impairments, often accompanied by vestibular areflexia. Genetic research has identified 12 distinct genes linked to Usher syndrome, distributed among its sub-types: 6 for Usher type 1, 3 for Usher type 2, and 2 for Usher type 3. At LSUHSC-NO, our primary focus lies on Usher syndrome type 1C, notably affecting Acadian populations in southern Louisiana and Canada. Approximately 2.5% of Usher cases stem from mutations in the USH1C gene, which encodes the protein harmonin. The USH1C.216G>A (216A) mutation, prevalent among Acadian populations dealing with Usher 1, disrupts the normal splice site of exon 3, leading to a frameshift that prematurely terminates the harmonin protein synthesis in exon 4. Dr. Jennifer Lentz and her lab team are focused on developing therapies to prevent or cure deafness and blindness by studying the sensory loss process in Usher mouse models.

Dr. Jennifer Lentz designed a prospective vision study, a prospective vestibular study, and a retrospective Usher study to observe the development of Usher Syndrome in patients over time. The prospective studies focus on participants with USH1C, tracking their clinical outcomes across multiple visits. The vision prospective study participants undergo four clinical assessments every six months to perform visual tests to monitor the progression of their visual impairment. The vestibular prospective study has participants involved in two clinical visits every six months to evaluate vestibular and hearing function. Since hearing loss is congenital in Usher Syndrome, advancements in newborn hearing screening have facilitated early cochlear implantation for many patients, prompting examinations that emphasize vestibular function and balance. Due to the range of sensory impairments and loss, Dr. Lentz along with Dr. Maria Reinoso are using the studies and clinical trials to identify what the most reliable and efficient assessments are to measure sensory performance and loss progression. Establishing standardized tests is crucial for advancing therapeutic strategies and drug development tailored to patients with Usher Syndrome.

Given the intricate array of sensory symptoms in individuals with Usher Syndrome, the development of precise clinical tests is paramount for advancing drug therapies for future patients. Dr. Lentz, Dr. Reinoso, and a team of researchers at the NIH are dedicated to identifying accurate measurement tools tailored to individuals with Usher Syndrome, systematically analyzing data to uncover significant trends across all three studies. This approach aims to enhance understanding of each test's specific metrics, ensuring therapeutic development focuses on quantifiable statistics that are able to be reproduced.