

2021 Research Award: Alport Hearing Loss

The human inner ear is inaccessible in life and therefore an understanding of how the hearing is affected depends upon the study of donated ear specimens that are obtained through enrollment in the National Temporal Bone Registry. With \$26,000 in funding provided by the Alport Syndrome Foundation, Dr. Felipe Santos and his team at the National Bone Registry will implement a year-long study of the inner ear specimens of patients with Alport syndrome and correlate their findings to the individual genetic sequence.

Sensorineural hearing loss in Alport syndrome is progressive. The risk of developing hearing loss in Alport syndrome appears to be dependent on the genetic variants of this condition. For example, among patients with X-linked Alport syndrome, hearing loss can be more severe with some genetic sequence variants when compared to others. It is not known how these genetic variants affect the microstructures and function of the inner ear.

Characterization of the genotype-phenotype of sensorineural hearing loss has important clinical implications for timing of surveillance with audiological testing and auditory rehabilitation with hearing aids or cochlear implants when indicated. Understanding the human phenotype of heritable hearing loss is fundamentally necessary to understand the pathogenesis of hearing loss, to validate animal models of disease, to propose rational treatment algorithms for a cure, and to ultimately assess outcomes for these treatments.



Dr. Felipe Santos

