Otoferlin Gene-mediated Hearing Loss Natural History Study

Status: RECRUITING

Eligibility Criteria

Sex: ALL
Age: Up to 44 years old
Healthy Volunteers: This study is NOT accepting healthy volunteers
Inclusion Criteria:

1. Clinical presentation of bilateral sensorineural hearing loss (SNHL), including auditory neuropathy (AN) / auditory neuropathy spectrum disorder (ANSD) phenotype or medical history of AN / ANSD phenotype earlier in life
2. Mutation(s) in the otoferlin gene
3. Able and willing to comply with all study requirements, as evidenced by successful completion of the informed consent (and assent, if applicable) process

Additional Criteria for Inclusion in the Prospective Phase:
4. Presence of OAE / CM and absent / abnormal ABRs in at least one ear (that does not have a cochlear implant) within 12 months prior to or at the Month 0 visit

Exclusion Criteria:

1. Unwillingness or inability of the potential participant and/or legally authorized representative to comply with all protocol requirements
2. Presence of cochlear nerve deficiency and/or cochlear nerve dysplasia

Additional Criteria for Exclusion from the Prospective Phase:
3. Presence of bilateral cochlear implants at the time of record review or planned within the next 6 months
4. Presence of middle ear or auditory brainstem implant(s) at the time of record review or planned within the next 6 months
5. Any condition that would not allow the potential participant to complete follow-up assessments during the course of the study and/or, in the opinion of the Investigator, makes the potential participant unsuitable for the study

Note: Potential participants will not be excluded based on their sex, gender, race, or ethnicity

Conditions & Interventions

Interventions:
OTHER: Natural History Study
Conditions:
Sensorineural Hearing Loss, Bilateral
Keywords:
Otoferlin Gene, sensorineural hearing loss (SNHL), auditory neuropathy, auditory neuropathy spectrum disorder

More Information

Description:
This is a retrospective and prospective longitudinal study in participants with Otoferlin Gene-Mediated Hearing Loss.

Contact(s): Elizabeth Dolgas - Elizabeth.Dolgas@cchmc.org
Principal Investigator:
Principal Investigator ID:
Phase:
IRB Number:
System ID: NCT05572073