CALIFORNIA STATE SCIENCE FAIR
2010 PROJECT SUMMARY

Name(s)

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Project Number

S0404

Project Title

Gene Discovery for Mendelian Forms of Hearing Loss

Abstract

Objectives/Goals

This study identifies genes within the remaining 80 plus genetic loci that have not been fully associated with hearing loss. If several hearing loss studies have found the origin of the hearing loss to be in the inner ear, then the genes responsible for deafness are expressed in the cochlea. Therefore, most forms of the disease are due to a defective form of some protein that is critical for normal cochlear function. If there are genes involved with functions that are not directly associated with hearing, then they are not expressed in the cochlea.

Methods/Materials

A list of genetic loci linked to hearing loss, which was compiled from the Hereditary Hearing Loss Homepage, had previously been written. These Mendelian forms of hearing loss were arranged depending upon whether they are dominant, recessive, X-linked, or Y-linked. Then, the genetic loci were identified and defined by the markers that were used to conduct familial linkage experiments. Once all of the genetic markers were entered and the borders delineated, the region of each loci was queried for genes. This acquired list of genes was compared to data of known mouse cochlear expressed genes. Loci with a limited number of genes that are present in both databases was acquired and evaluated by DNA sequencing.

Results

Out of 2,690 hearing loss related intervals and 7,086 mouse genes expressed in the cochlea, 42 loci overlapped. Of the 42 loci that were found to overlap in both lists, 7 intervals containing 1 to 3 overlapping genes were particularly noted due to their high likeliness of involvement in Mendelian forms of hearing loss.

Conclusions/Discussion

The low number of genes, known as positional candidates, within these 7 intervals makes the interval easier for future mapping. The findings of overlap between the list compiled from the Hereditary Hearing Loss Homepage and the list of known mouse cochlear expressed genes shows that forms of hearing loss can result from a defective protein that is critical for normal cochlear function and that genes involved with the function of hearing are expressed in the cochlea.

Summary Statement

By using bioinformatics to compare two previous studies involving cochlear mice genes and Familial Linkage Studies, this project identifies genes found in the cochlea and related to Mendelian forms of hearing loss.

Help Received

Dr. Jeff Ohmen, from House Ear Institute in Los Angeles, was my mentor for this research.