Fabio Arrojo, J Proteomics Bioinform 2014, 7:8
http://dx.doi.org/10.4172/0974-276X.S1.074

OMICS Group Conferences
Accelerating Scientific Discovery

4th International Conference on
Proteomics & Bioinformatics
August 04-06, 2014 Hilton-Chicago/Northbrook, Chicago, USA

Deep sequencing to identification and characterization of deafness genes

Fabio Arrojo
University Camp, Brazil

Hearing loss is the most common sensory impairment in humans, affecting approximately 10% of the entire world population. The restriction of communication by oral expression results in changes in cognitive and psychological development of the affected individual. In developed countries, one in every 500 individuals has profound/severe bilateral sensorineural hearing loss. Among the causes of hearing loss, more than 60% of cases of congenital hearing loss is genetic. So far already aware of 150 loci and 103 genes involved in hearing loss, and most of them have at least 20 changes (point mutations, deletions, insertions, etc.) that may to cause deafness.

It is known that 2/3 of the deaf population still has no diagnosis for disability. This is partly because of the large number of genes related to hearing loss, it is estimated that it can reach 300 genes (1% of human genes) and by conventional molecular techniques that do not have great power sweep or affordable to use in the field of diagnostics.

With the emergence of technologies 'high-throughput' (high performance), new molecular platforms have emerged, allowing the simultaneous detection of multiple genes and/or changes. One of the most powerful platforms available today are of Next Generation Sequencing - NGS. With these, you can scan in over 30 Gb of DNA in just one day. Among the variations of existing techniques, there is the Whole exome Sequencing - WES. This variation allows complete sequencing of several exons of an individual, thereby studying alterations in the coding regions of genes. The advantage of this technique is the generation of a few residues, such as introns, the speed and the cost to perform the experiment, they end up being much lower when compared to conventional techniques. Thus, using the WES will be held scan the exons of 284 genes that are related to hearing loss and/or are candidate genes to identify and characterize novel genes that cause deafness in our population. In silico assays and animal model will be performed to validate the changes identified during the sequencing, so as to separate and study frequency in the population.

fabio.arrojo@cbmeg.unicamp.br