

Development of Bead-Based Suspension Array Technology for the Diagnosis of Hereditary Hearing Loss

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Objective

Hearing loss is one of the most common birth defects and one of the most prevalent sensorineural disorders. Given that hereditary hearing impairment exhibits incredible genetic heterogeneity, effective diagnostic methods are highly demanded by the clinical management.

Methods

We developed a new diagnostic assay using the bead-based suspension array technology (BBSAT), which can detect 20 point mutations in four genes of Asian prevalent hearing impairment in parallel within 5 hours. More than 200 previously genotyped clinical samples were used to determine the cut-off values and to validate the accuracy of new assay.

Results

In the BBSAT assay, suspended beads increase surface area to 3-D and more probes concentrated on beads. Besides, probes and targets mixed in the same mobile phase also generate better hybridization dynamics. With the multiplex PCR primer and probe design strategy, 20 point mutations in four genes of Asian prevalent hearing impairment can be simultaneously detected in a single assay. The results of BBSAT were completely in line with known genotypes, indicating that the new diagnostic assay possesses excellent clinical sensitivity (100%) and specificity (100%) to identify genetic mutations of hearing loss.

Conclusion

The BBSAT is a rapid, accurate and cost-effective technology, suitable for molecular diagnosis of the hereditary hearing loss in large populations.