# Diagnosing hearing loss at a fraction of the time and cost

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Over 28 million Americans are hearing impaired, and 50 percent of these cases can be traced to genetic causes. The condition can be especially challenging for children born hearing impaired because spoken language, reading, and cognitive development are tied to hearing. That makes early diagnosis essential for identifying appropriate therapy and treatment.

Unfortunately, diagnosis for genetically caused hearing loss has traditionally been a slow and costly pursuit. A comprehensive genetic assessment for a deaf patient costs about $20,000 and takes years to complete. Now Prof. Karen Avraham of Tel Aviv University's Sackler Faculty of Medicine, in a unique collaboration with Prof. Moein Kanaan from Bethlehem University, has successfully developed a fast-track genetic diagnosis for hearing loss through "exome deep sequencing" - a method that sequences hundreds of thousands of genes at a time.

Profs. Avraham and Kanaan studied a group of 11 Jewish Israelis and Palestinian Arabs, none of whom were related to each other but all of whom had deafness in their families. Using a technique called exome sequencing, which collects relevant DNA from specific sites of the body, they scanned 246 genes, ultimately identifying five mutations that lead to genetic hearing loss within the specified population. Reported in the journal *Genome Biology*, their method provides doctors with better diagnosis capabilities and can improve the quality of care for patients with hearing loss. For less than $500, researchers can scan all the known genes for deafness and provide results in a matter of weeks.

**Tracing hereditary deafness**

Though it is common to lose hearing as we age, three out of every 1,000 infants born in the U.S. are hearing impaired, and a third of these cases have hereditary causes. Although scientists have long searched for the genetic mechanisms underlying hearing loss, Prof. Avraham says it is a challenging task, noting that the critical genetic mutations that cause hearing impairment differ among populations, countries, and ethnicities.

Some of the mutations, says Prof. Avraham, have never before been identified in the Middle Eastern deaf population. This is the first time this technology has been applied to these populations, she says.

**Better care on the horizon**

The earlier hearing loss can be diagnosed, Prof. Avraham notes, the better care a patient will receive. More information about the genetic causes of a patient's hearing impairment enables doctors to better determine how it might progress, and to formulate treatment plans and rehabilitation options based on the challenges a patient might face.

In the past, doctors have been confined by methods of genetic analysis that are laborious, time consuming, and expensive. The number of mutations that scientists had the capacity to look for in any one patient was limited. Prof. Avraham's method allows doctors to identify the genetic cause of a patient's hearing loss in just a few weeks. And the more information researchers have about the genetic mutations that can cause hearing loss, says Prof. Avraham, the more likely that targeted and more effective treatments can be developed.

"It is a remarkable step forward in helping us to find treatments - and even cures - for patients. This new technology is changing the way we practice genomic medicine, and revolutionizing genetic diagnostics," she explains, noting that the technology can be applied to search for genetic mutations that characterize any disease or condition.

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