

Through his analysis of the temporal bones and medical records donated by cochlear implant users for study in the temporal bone bank collection at the House Ear Institute, Jose Fayad, M.D., made an exciting discovery.

The Bones Tell the Story

Contrary to popular belief, a deaf person can achieve an excellent result, regaining significant hearing ability using a cochlear implant, even with as little as 10% remaining auditory nerve function. This finding was made possible by the presence of a complete clinical history, including cochlear performance data that accompanies each set of temporal bones in the Institute's

collection, and is but one example of the research value to be found in this unique resource.

The Source:

From their inception over 60 years ago, House Ear Institute and House Clinic have encouraged patients with hearing problems to pledge the donation of their temporal bones for scientific study after they pass. In 1960, the Eccles Temporal

Bone Laboratory was established within the Institute's Department of Histopathology. Today, this extensive and uniquely documented collection (1,172 temporal bones from 586 subjects) is one of the Institute's most valuable research resources. As a resource for scientific studies, it is unparalleled at any other hearing center and attracts scientists and scholars from every corner of the world.



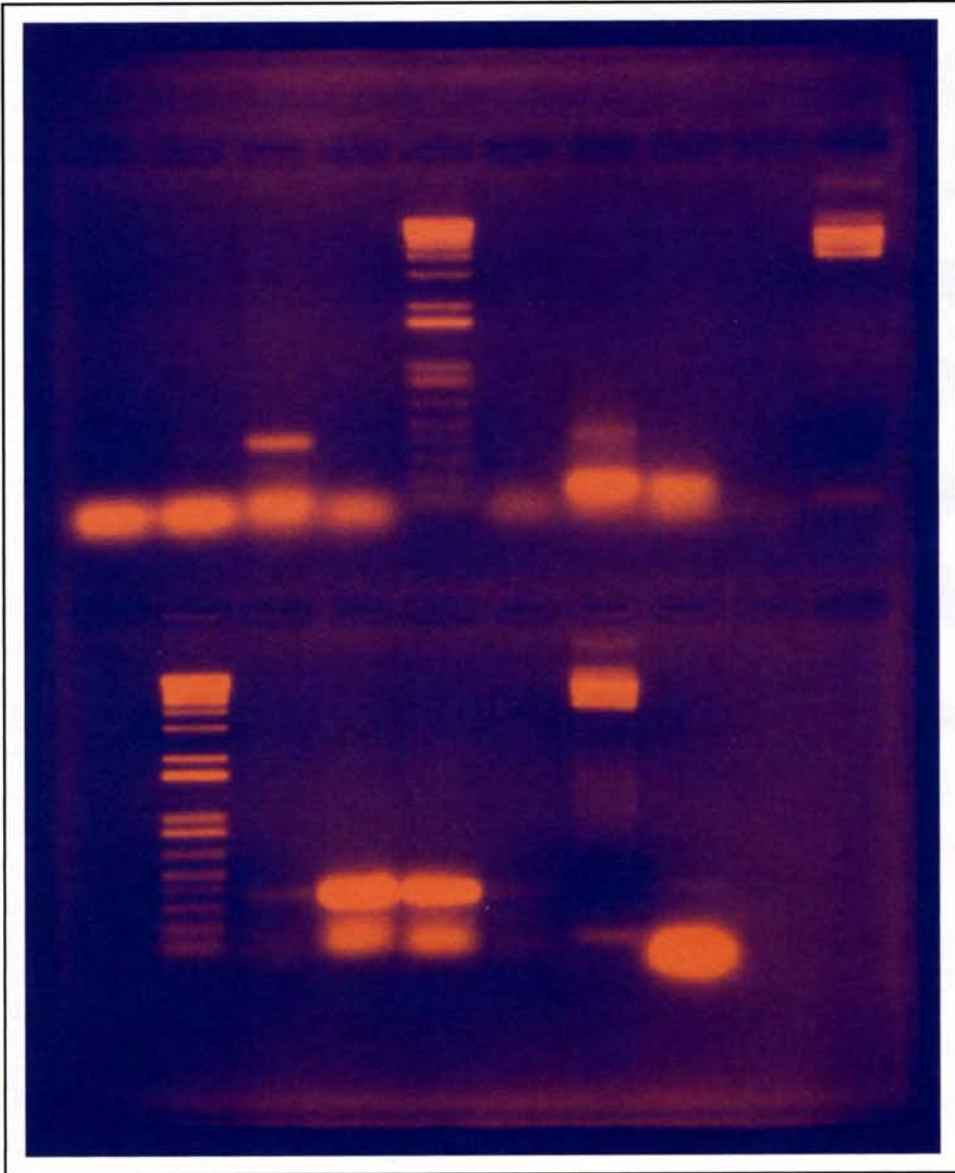
*Clockwise from left:
Fred H. Linthicum, M.D.,
Jose N. Fayad, M.D.,
Robert Gellibolian, Ph.D.*



The Scientists:

Four years ago, Dr. Fayad joined Fred Linthicum, M.D., and his dedicated team of researchers, as co-director of the Institute's Histopathology Department/Temporal Bone Lab. In all, seven researchers and technicians currently are focused on laboratory projects. Dr. Linthicum's extensive experience in the pathology of hearing disorders is complemented by Dr. Fayad, who divides his time between the lab in the Institute

Gel chromatogram of amplified fragments of DNA isolated from human temporal bones.



The DNA available from archival temporal bones is a rich source for understanding the molecular origins of various forms of hearing loss and other related diseases.

and his medical practice in the House Clinic, applying clinical insights to research studies. The lab recently was bolstered by the addition of Robert Gellibolian, Ph.D., who specializes in molecular biology and biotechnology.

The Impact of DNA Recovery and New Technologies:

The DNA available from

archival temporal bones is a rich source for understanding the molecular origins of various forms of hearing loss and other related diseases. Until recently, research was hampered by the quality and yield of DNA in archival bones, which often is diminished by the age of certain specimens and the way bones were prepared and preserved. Refinement of the method for

extracting DNA from these bones has yielded improvements in this process and has been one of the most significant Institute achievements in recent years. HEI scientists using these new methodologies have demonstrated that viral nucleic acids can be readily detected in archival temporal bones. This has paved the way for molecular analysis of temporal bones and has enabled researchers to study the molecular basis of otologic disorders directly within bone specimens, allowing progress in many areas of research.

PROFILES IN SCIENCE

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Jose Fayad, M.D.

Dr. Fayad and daughter Nathalie



“This was my dearest dream and I couldn’t imagine ever being lucky enough to work at the House Clinic. I’m now living my dream. And, this is extra special after all I have been through and all that my family has been through to get here.”

JOSE N. FAYAD, M.D.

Dr. Jose Fayad is known for his expertise in otology, and for his gentle approach to patients. Fluent in French, Spanish, English and Arabic, Dr. Fayad is able to communicate easily with patients from many backgrounds. Between surgeries and patient consultations, he prefers to relax by reading French literature – classics by Hugo, Beaudelaire, Flaubert, and Mauriac. French is also Dr. Fayad’s language of choice at home in Southern California., where he lives with wife Cathy and their two teenagers, Daniel and Nathalie. A horticulture enthusiast, on weekends Dr. Fayad is outside planting fruit trees and grape vines, and cultivating a garden of camellias and gardenias, the exotic flowers favored by his family.

Dr. Fayad has a real passion for ear medicine and science. This passion has its foundation in an early experience he had during medical school in Toulouse, France. Dr. Fayad had decided to specialize in otology and was completing his residency in ear, nose and throat (ENT) medicine at the University Paul Sabatier Hospital when he helped treat an otosclerosis patient with a stapes procedure. Her hearing loss was significant enough that most of Dr. Fayad’s pre-surgery interview with her had to be conducted in writing. Then, immediately after completing her stapes surgery, he tested her surgical outcome by asking her a few questions in the operating room. She heard every word, even at a whisper, and Dr. Fayad felt a rush of excitement that he’d

been able to help a patient in such a profound and immediate way. His early enthusiasm for otology brought him to a visiting physicians' course at the House Ear Institute many years ago, where he got inspiration from the House physicians who mentored him. He is thrilled to be working alongside those same physicians now at House, and, feels the same surge of excitement at the conclusion of every stapes surgery, a procedure he's now performed for hundreds of patients at the House Clinic. He feels an equal sense of reward at the completion of each cochlear implant surgery, and with every milestone he reaches in his collaborative research investigations with Dr. Fred Linthicum in the Histopathology Laboratory. Dr. Fayad feels he is where he should be, at the "Mecca" of hearing science and medicine, where the most exciting things in neurotology are happening.

Dr. Fayad's calm and soft-spoken nature belies the many traumatic experiences he endured as a youth in war-torn Lebanon. As a 16-year-old he literally dodged a bullet in Beirut, and his father realized it was time to move the family out of the country. After many narrow escapes and family separations, the family safely relocated to Venezuela, where Dr. Fayad completed high school at the Lycée Francais in Caracas before heading to France for college and medical school. This summer, Dr. Fayad was forced to relive some of this early trauma when his 16-year-old daughter visited Beirut to attend the American University there for a summer program. She arrived just days before Beirut's most recent upheaval during which Lebanon was bombed. The Fayad family struggled to get her onto the first ship out of the country. Now his daughter is safely back home in California, and Dr. Fayad has new reasons to be thankful that he pursued his dream to practice otology at House and brought his family to the U.S. ❖

Study Examples:

- HEI scientists are currently studying the association between viral diseases and deafness. CMV (cytomegalovirus), one of the most common human viruses, infects between 50% and 85% of adults in the United States by 40 years of age and is the virus most frequently transmitted to a developing child before birth. Donated bones are assayed for viral DNA to determine if CMV may be playing a role in unexplained deafness.
- The Institute's DNA extraction technique is also used to detect bacterial DNA in temporal bone specimens and has allowed the identification of pathogens that underlie infectious and inflammatory diseases of the middle and inner ear, including otitis media, and labyrinthitis, a devastating inflammation of the inner ear causing profound hearing loss, vertigo and balance disorders.
- HEI scientists are now able to assay genetic material from temporal bones for the detection of mutations associated with hearing and balance disorders, and correlate this information with the bone's histopathology. Institute scientists are currently in the process of investigating the correlation between various mutations in three genes to pathologies in the cochlea. These genes are: Connexin-26 (Cx26), coagulation factor C homolog (COCH), and Pendrin.

The Cx26 protein is essential to intercellular communication and mutations of this gene have now been traced to 50% of the cases of childhood non-syndromic recessive hearing loss. To date, seven known COCH mutations have been found. The possibility that these play an important role in balance disorders and age-related hearing loss

(Continued on page 43)

(Continued from page 37)

Laboratory goals are continued collaboration among internal and external researchers, and documentation and analysis of temporal bones.

is being considered. Pendrin mutations can cause non-syndromic hearing loss, as well as Pendred Syndrome, a condition characterized by hearing loss and an enlarged thyroid gland.

Recently, the Temporal Bone Laboratory received a 5-year contract from the National Institutes of Health (NIH) to develop and apply new techniques to study human temporal bones and to utilize the highly valuable resources at HEI. Collaborative studies will incorporate immunohistochemistry, molecular biology (RNA and DNA recovery), morphometry, and advanced imaging techniques. Laboratory goals are continued collaboration among internal and external researchers, and documentation and analysis of temporal bones received as a result of the pledge program and in conjunction with the National Temporal Bone, Hearing and Balance Pathology Resource Registry. In the near future, HEI's online database will be made accessible to other hearing institutes and centers worldwide, expanding and promoting the network of collaborations globally. ❖

Family CAMP

Building Bridges

Since 1985, House Ear Institute's annual Family Camp program has reached out to parents and children who are coping with a pediatric hearing loss in their family. The issues and choices they face can be overwhelming and there are strong needs to share and connect with others in similar situations.

A focus on self discovery, communications and building relationships are at the core of the Family Camp experience. Each year Family Camp presents a weekend of fun and unique opportunities to improve bonds between siblings and parents. It also fosters valuable links to other families facing similar issues. Dates for the 2007 camp are June 1-3. For more information on registration, volunteer counselor opportunities or other ways you can help, contact Marilee Potthoff at (213) 353-7078; online information is available at www.hei.org.



Actor Steven Anthony Lawrence, who has appeared in many films including "Kicking & Screaming" with Will Ferrell and "Cheaper by the Dozen" with Steve Martin, visited the Institute's Family Camp. He put his knowledge of sign language to use as he greeted campers and signed autographs.

