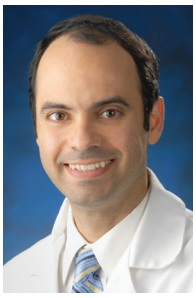


Symptoms: Ear Plugging and Hearing Loss

By Hamid R. Djalilian, MD

A 58-year-old patient presents with a history of decreased hearing and ear plugging on the left side. He typically uses an earplug when he sleeps and has had problems placing the earplug in that ear, the patient says.

He also experiences occasional drainage from the ear, which he likens to a cheesy material, and he has never had ear surgery, the patient reports.

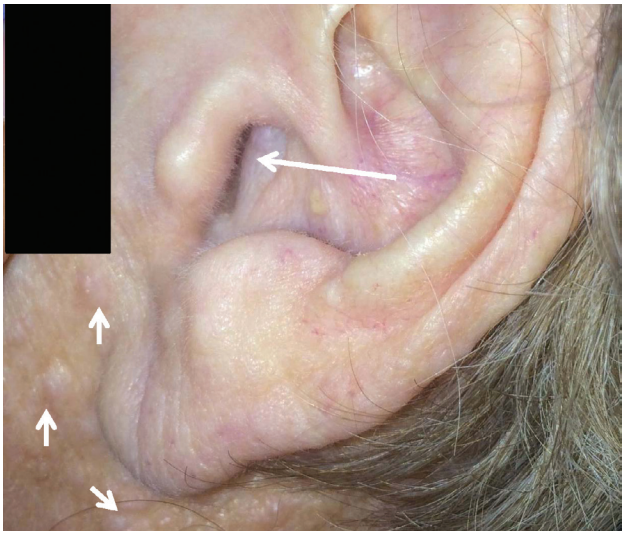
An examination of the ear reveals a mass completely blocking the external auditory canal. The mass is soft and somewhat compressible.

Once the mass is moved aside, the rest of the ear canal and the tympanic membrane appear normal except for some moist, dead skin in the medial canal.

The figure on the right shows the patient's examination.

What is your diagnosis? See p. 12.

Dr. Djalilian is director of neurotology and skull base surgery and associate professor of otolaryngology and biomedical engineering at the University of California, Irvine.



In this photo of our patient's ear, the large arrow shows the mass blocking the ear canal, and the small arrows point to skin nodules.

Diagnosis: Neurofibromatosis Type 1

By Hamid R. Djalilian, MD

Continued from p. 10

A close examination of the patient's ear, depicted in the photo on page 10, shows multiple skin nodules on the same side as the audiological symptoms (short arrows). These masses represent small neurofibromas of the facial skin. The mass in the ear canal is a neurofibroma of a nerve that supplies sensation to the external auditory canal.

Cranial nerves V, VII, IX, and X innervate the ear canal and auricle. Upper cervical nerve rootlets also provide sensation to the ear, primarily via the greater auricular nerve and other distal nerves. Patients can develop neurofibromas from any of the small distal tributaries of these nerves, which may then grow and block the ear canal.

PROGRESSIVE BUT DIVERSE CONDITION

Neurofibromatosis type 1 (NF1) is a rare genetic disorder seen in one of every 3,000 to 4,000 people worldwide. The autosomal dominant condition, which affects half of a patient's children, is characterized by changes in skin coloring, or pigmentation, and the growth of tumors along nerves. These tumors are most commonly seen on the skin and can present anywhere there is a peripheral nerve.

The *NF1* gene provides instructions for making a protein termed neurofibromin. The protein is produced in many cells, including nerve cells and the specialized cells surrounding nerves, which are called oligodendrocytes in the brain and spinal cord, and Schwann cells in peripheral nerves.

Neurofibromin is a tumor suppressor, preventing cells from growing and dividing too rapidly or in an uncontrolled manner. Mutations in this gene lead to the production of a nonfunctional version of neurofibromin that cannot regulate cell growth and division. Therefore, tumors such as neurofibromas can form along nerves throughout the body.

It is unclear how mutations in the *NF1* gene lead to other features of neurofibromatosis type 1, such as café-au-lait spots and benign growths called Lisch nodules, as well as the learning disabilities often seen in these patients.

Neurofibromatosis type 1 is most commonly recognized in early childhood, when almost all people with the condition have multiple café-au-lait spots. These flat patches on the skin are darker than the surrounding area, and they can grow in size and number as the patient gets older. Patients also may later develop freckles in the axilla and groin.

During childhood, Lisch nodules often appear in the iris of the eye. Some patients with NF1 develop tumors that grow along the optic nerve, called optic gliomas. These tumors may interfere with vision.

Most adults with neurofibromatosis type 1 have benign tumors generally located on the skin or immediately subcutaneously. Uncommonly, patients with this condition develop malignant peripheral nerve sheath tumors. NF1 patients also have an increased risk for brain tumors and leukemia.

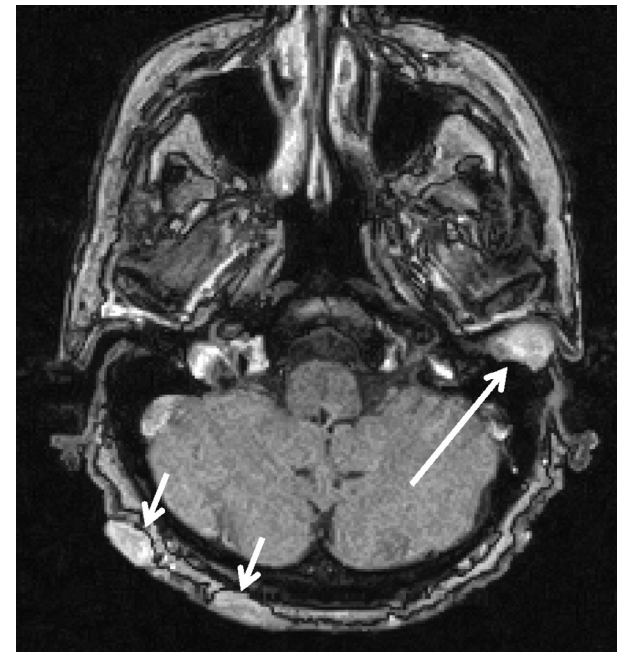


Figure 1. The T1-weighted post-enhancement axial MRI shows the mass (long arrow) blocking the ear canal. The short arrows indicate subcutaneous neurofibromas of the scalp.

Additional signs and symptoms of neurofibromatosis type 1 include high blood pressure, short stature, an unusually large head (macrocephaly), and skeletal abnormalities such as scoliosis.

NF1 is a generally progressive but diverse condition. Some patients with the genetic mutation have no symptoms, while other members of the same family with the identical genetic mutation may have severe manifestations. This differing level of symptoms is called variable expressivity of the gene.

SPECIAL CONSIDERATIONS

A significant concern of patients with NF1 is disfigurement caused by the cutaneous neurofibromas and pigmented lesions. The majority of NF1 patients live completely normal and uninterrupted lives.

When evaluating a neurofibroma of the ear canal, the primary considerations are the nerve of origin and the proximal spread of the tumor. In other words, if the tumor derives from the facial nerve's branch to the ear canal, we want to ensure that the tumor has not spread along the small nerve in the ear canal to the main trunk of the facial nerve.

Figure 2. The T1-weighted post-enhancement coronal MRI also shows the mass (arrow) blocking the ear canal.

Such spread potentially makes the surgery more dangerous, as pursuit of the tumor may lead to the main trunk of the facial nerve and cause facial paralysis upon tumor removal. Other considerations include spread of the tumor near the jugular bulb or even intracranially.

Our patient's MRIs showed that the mass was confined to the ear canal and had not spread along the nerve fibers to the main trunk of any of the major cranial nerves (see figures 1 and 2). CT imaging indicated no destruction of the bony canal.

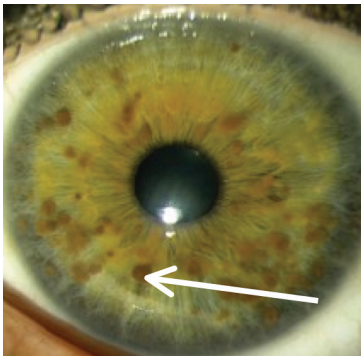


Figure 3. In patients with neurofibromatosis type 1, benign growths called Lisch nodules (arrow) often appear in the iris during childhood. (Reprinted with permission from the *Oman Journal of Ophthalmology* [2013;6(3):159-164].)

Given the blockage that was present, the patient had the mass excised. When a mass involves a significant portion of the ear canal, the excision generally is accompanied by split-thickness skin grafting to allow for replacement of the excised skin. If the ear canal is left open without replacement of the excised skin, the canal likely will contract and narrow. tjh