Cochlear implantation in a profoundly deaf patient with MELAS syndrome

Cochlear implantation is now an established technology for restoring hearing in profoundly deaf patients. Adults who have lost all useful hearing in both ears are suitable for cochlear implantation if they are profoundly deaf (gives implications hearing thresholds of 100 dB nHL or worse, across the frequency range 125 to 8000 Hz), with aided hearing thresholds worse than 60 dB for the frequencies 250 to 4000 Hz and scoring less than 30% in a test of sentence discrimination, using their hearing aids and without lip reading. We describe a patient with MELAS syndrome (mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes) who became profoundly deaf and who has successfully undergone cochlear implantation and rehabilitation.

A right-handed secretary with MELAS syndrome confirmed at age 10 to 12 mutation at nucleotide 3243 in the mitochondrial genome, was referred to the cochlear implant programme of The Royal National Throat, Nose, and Ear Hospital. She had insulin dependent diabetes, congenital cataracts, short stature, weakness, fatigue, and hearing loss. She had never had encephalopathy or strokes. Her mother is also diabetic, has glaucoma, and has a lesser degree of deafness, and her sister has been profoundly deaf from adolescence in addition to having severe mental retardation. The patient had begun to experience bilateral hearing loss at the age of 22, with slow deterioration up to the age of 29, by which time she was profoundly deaf in the right ear. By the age of 30 she was also profoundly deaf in the left ear and had developed tinnitus. She had no spontaneous vertigo, but sudden movements could leave her temporarily unsteady. At the age of 34 she was referred for assessment for cochlear implantation. Her ability to communicate with her family was severely restricted due to deafness. She had begun to read her husband to a limited extent, but communication with her family has improved. As she had normal bilateral vestibular function, as she had virtually no hearing in either ear. Middle ear impedence was normal, and she had no peaks in response to wide band clicks prior to implantation. Electrical stimulation of the cochlea, responses and electrocochleography showed that the patient has resumed full time work in an office. The tinnitus has remained stable, and there have been no vestibular problems. At the 2 year assessment she scored 97% correct on CUNY/UCL British adaptation of a sentence discrimination test developed at City University, New York), using her implant and lip reading and 92% correct on BKB (Bamford, Kowal and Bench) sentences (another speech discrimination test) using her implant but without lip reading. Speech production was within normal limits, although the narrow pitch range reflected her slightly flat pattern of intonation.

MELAS syndrome was first described in 1984 and is one of a group of mitochondrial cytopathies, associated with point genetic mutations. In the brain the characteristic abnormalities are basal ganglia calcification and focal lesions with surrounding cerebral atrophy, resulting from cellular rather than vascular dysfunction. Although it does not feature in the acronym, hearing loss is a common finding in MELAS. Reports of large kindreds and patient series have shown that at least 50% of patients have a moderate or severe sensorineural hearing loss: 21 of 28 patients with MELAS in an Australian series were deaf, as were eight of 14 patients in a British series. The phenotypic expression of the mutation is subject to at least three constraints; the percentage of mutant mitochondrial DNA in the target tissue (which has at most a loose correlation with clinical lesions), the oxidative stress to which different organs or cell populations are exposed, and as yet unidentified collaborating somatic mutations which enhance selective aspects of the syndrome.

The cochlea is an organ extremely vulnerable to oxidative stress. The outer hair cells have a precarious, indirect metabolic support from Deiter cells, and the stria vascularis is both metabolically very active and non-mitotic, hence further subject to mutation accumulation. Recently detailed audiological findings have been reported in 18 patients with MELAS, and the authors argued that the hearing loss in their patients was entirely due to cochlear lesions. There were excellent speech discrimination scores in six of 12 patients with MELAS, and the authors argued that the hearing loss in their patients was entirely due to cochlear lesions. There were excellent speech discrimination scores in six of 12 patients with MELAS, and the authors argued that the hearing loss in their patients was entirely due to cochlear lesions.

A central auditory lesion was hypothesized due to hyperfusion of the occipital and parietal lobes, with a significant deficit in perfusion reserve. A case report of a patient who died after having had severe seizures and stroke-like events, and who had had multiple imaging studies, showed mild temporal lobe atrophy at necropsy with associated spongy degeneration of the cortex. Her selection as a candidate for cochlear implantation was straightforward, and she has been successful in adapting to the device and has gained a significant benefit from it. The performance of the patient in the BKB word test places her in the top 5% of adult performers in our patient series. Another patient with profound deafness and MELAS, who had had seizures and strokes, has recently been reported incidentally in a large series to have been implanted with a successful outcome, but unfortunately details were not provided.

The fact that this patient has gained considerable benefit from her cochlear implant raises the possibility that profound deafness with MELAS syndrome and profound sensorineural deafness could benefit from this procedure.

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CORRESPONDENCE

Lead poisoning from complementary and alternative medicine in multiple sclerosis

In response to the article Lead poisoning from complementary and alternative medicine in multiple sclerosis, we are very concerned that this