gesting that the deletion is responsible for
the myopathy. There was no associated
duplication, in accordance with Poult et
al,\(^1\) who showed that duplications are carac-
teristic of Kearns-Sayre syndrome. The
deletion was flanked by a 14 bp imperfect
tandem repeat at positions 8407–8420 and
15 658–15 671. It encompassed nine genes
encoding subunits of the complexes I, II,
IV, and V of the respiratory chain, as well as
ten RNA genes. So far no deletion has been
mapped with these boundaries.\(^2\) There is a
correlation between the percentage of
deleted mtDNA and the severity of the
myopathy: a recent study\(^3\) showed 31 (26\%) of
mutant DNA in unaffected muscles and up to
95\% of deleted muscles in affected
muscles. With 51\% of mutant versus total
mtDNA in a very mildly affected muscle our
results are in accordance with the above sur-
vey. The assay for enzymatic activity of the
respiratory chain was normal showing that a
large deletion, with a proportion of 51\% of
mutant mtDNA in the biopsied muscle is
compatible with normal respiratory chain
activity.

Histologically signs of a mild myopathy
were apparent, with few cytochrome oxidase
negative fibres. There were no ragged red
fibres, consistent with the fact that these
may be absent in established mitochondrial
encephalomyopathies.\(^1\)

The mitochondrial DNA deletion was not
present in leucocytes, showing the need for a
muscle biopsy to prove the genetic defect.

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ultrastructural analysis.

\(^{1}\) Poult et al

\(^{2}\) Poult et al

\(^{3}\) Poult et al

**Figure 1** Southern blot analysis of muscle DNA of the patient (P) and of a healthy control (H), digested with PvuII and BamHI, and hybridised to a mitochondrial tRNA\(^{\text{wcu}}\) probe (A), which detects all mtDNA. The PvuII digest of the patient DNA shows a 16.5 kb band (LWT) and an additional smaller band corresponding to the deleted molecules (LD). BamHI digestion shows two additional slower migrating bands (CDM, CDD) that are not seen when the same filter is hybridised with a probe lying within the deletion (B) and that only detects wild type and duplicated molecules, therefore ruling out the coexistence of a duplication. LWT = linear wild type; LD = linear deleted; CDM = circular deletion monomer; CDD = circular deletion dimer. In the wild type sequence there is an imperfect tandem repeat of 14 base pairs in length (C), flanking the deletion. In the mutant DNA (clone J5U) only one copy of the repeat is present and the sequence is identical to the "lower" sequence. Mismatches are underlined. Numbers refer to nucleotide positions of mtDNA.

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**Pure word deafness after resection of a
tectal plate glioma with preservation of
wave V of brain stem auditory evoked potentials**

Pure word deafness from brainstem lesions
is uncommon because of the bilateral repre-
sentation of the auditory system within the
brainstem.\\(^2\) The unique involvement of the
inferior colliculi in cases of word deafness in
which other anatomically close structures of
the auditory system had been spared has only
been reported twice.

It is generally accepted that the inferior
colliculus is the generator of wave V of
brainstem auditory evoked potentials (BAEPs)
in which its bilateral destruction will inevita-
ably lead to abnormalities in their recording.\\(^1\) Bilateral destruction of the infe-
rior colliculi with preserved wave V of BAEP
is rare and has not been described to our
knowledge.

Here we report a patient in whom a cir-
cumscriptive lesion of both inferior colliculi
has led to the isolated neurological deficit of
pure word deafness with repeatedly docu-
tmented preservation of wave V of BAEPs.

This 36 year old Turkish patient developed
progressive signs of raised intracranial pres-
sure at the age of 28, was diagnosed as hav-
ing obstructive hydrocephalus, and was
shunted. On CT one year before admission a
pinal region mass was described for the first
time. This showed pronounced enlargement on MRI and occupied the dor-
sal midbrain with exophytic growth. His
neurological examination was normal apart
from a reduced visual acuity. Speech audiog-
ram, pure tone audiogram, and BAEPs were
normal.

The lesion was attacked via a infratentor-
il-supracerebellar approach. The inferior
colliculi could not be identified during
surgery. Complete removal was documented
on postoperative MRI as well as the destruc-
tion of both inferior colliculi (figure). Histology
disclosed a pilocytic astrocytoma grade I.
Axial T1 weighted MRI 48 hours after surgery. Complete tumour removal was documented in all planes. The section through the caudal midbrain, just above the posterior mesencephalic junction, clearly shows the destruction of both inferior colliculi.

After surgery the patient was unable to understand verbal communication, but non-verbal hearing, reading, writing, and speaking abilities were unimpaired. He could still identify and localise all sources of non-verbal sounds. Furthermore, he also identified correctly pieces of music that he had known before. Pure tone audiogram and BAEPs were normal and identical to those before operation. Speech audiogram performance, however, had dramatically deteriorated to discrimination scores of 10 and 20.

Hearing impairment related to brainstem disease is rare and the clinical picture is mostly unimpressive with abnormalities detected only by subtle audiological testing. Sixty two cases of hearing impairment by indirect compression of the brainstem in patients with tumours in the pinal region are described in the medical literature. Due to the proximity of the auditory nuclei and their interconnections within the brainstem it was obvious that auditory centre caused the deficit in these cases. Only three cases had syndromes of pure word deafness with lesions restricted to the inferior colliculi.

It is agreed that only bilateral destruction of the inferior colliculi produces clinical apparent hearing loss. Bogner et al. described a patient with a unilateral inferior collicular lesion with apparent lack of auditory consequences, who exhibited a significant extinction of the contralateral ear during dichotic testing. This extinction might constitute just the mildest degree of failure in verbal comprehension and word deafness, the minimal variant of the same type of auditory disorder. Multiple interconnections at all levels of the brainstem auditory system readily explain the necessity for bilateral destruction to produce apparent clinical consequences. 1, 4

It is, however, not easy to explain why the bilateral destruction seen in our patient did not have consequences for higher binaural hearing functions other than linguistic comprehension. Among the important three levels where bilateral interaction primarily occurs—the superior olivary complex, the nuclei of the lateral lemniscus, and the inferior colliculus—the third is the largest component and the target of all ascending connections from the lateral lemniscus. Binaural mechanisms necessary to obtain spatial representation occur essentially at these subthalamic levels. It is known that neurons involved in analysing interaural time or level differences are found abundantly in the central inferior colliculus, but also in subdivisions of the superior olivary complex. Thus an intact superior olivary complex suffices for analysis of interaural time or level differences to localise a non-verbal sound.

There is evidence that the neurons of the inferior colliculus—by contrast with those of the superior olive—have more specific functions and respond to interaural intensity changes within a restricted range. This is one argument in favour of the hypothesis that the inferior colliculus possesses “higher” functions than the lower nuclei. The alternation of excitatory and inhibitory response of neurons with their ability of dynamic variation of these responses according to different stimulus conditions is the fundamental working principle of the auditory brainstem nuclei. This is supported by a suggestive “gating process” at the inferior colliculus level that may block “irrelevant” sensory input which would be essential for linguistic comprehension. It has been shown that the inferior colliculus incorporates units that respond only to dynamic aspects of a stimulus such as frequency modulated tones in the spoken word. 4 These physiological aspects could be named “auditory imaging” for the clinical picture in our patient.

The inferior colliculus is thought to be the generator of wave V of BAEPs as documented in numerous human and animal studies. 1, 3 It is suggested that a unilateral inferior collicular lesion can be accompanied by normal wave V of BAEPs. 1 Yet completely normal BAEPs in cases of bilateral destruction of the inferior colliculus in humans have not been reported to our knowledge.

We can think of two possible explanations. The first is a technical aspect. As documented in the recent publications of Lapras group BAEPs are less sensitive in quadrigeminal plate disease than middle latency auditory evoked potentials. Due to the limited experience in this field it can only be speculated that our patient might have shown the expected abnormalities. 1

On the other hand, some reports postulate an anatomically diffuse second auditory brainstem pathway outside the primary lemniscal projections called the medial extralemniscal auditory pathway. 1 Bogner et al. favour the existence of this parallel projection system in the “questionable” case of preserved wave V of BAEPs with bilateral inferior collicular destruction. 1

We conclude that our patient allows arguments for the possible existence of a parallel extralemniscal auditory brainstem pathway and a subsequent destruction of superior colliculus in the processing of verbal communication.

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Congenital cerebellar ataxia, mental retardation, and atrophic retinal lesions in two brothers

The congenital cerebellar ataxias constitute a rare group of syndromes which are difficult to classify. 1-3 Most patients show a non-specific clinical picture characterised by cerebellar ataxia, motor delay, nystagmus, and, often, mental retardation and limb spasticity. The course is not progressive and usually improves with age. Autosomal recessive, autosomal dominant, and X linked recessive inheritance have been suggested in different families.

We recently examined two brothers affected by congenital cerebellar ataxia; they are the only two sons of a healthy white mother who had married her father’s brother; the younger sister is apparently normal.

The oldest patient, the first born of three siblings, is 52 years old and in early life showed a mildly delayed psychomotor development. Walking started at 18 months and ataxia was then noticed. At the age of 18 he developed severe visual problems because of bilateral keratoconus. At the age of 31, he was admitted to hospital. Hodgkin’s lymphoma was detected which responded to chemotherapy. In the past years, cluster headache, memory impairment, vertigo, and mild self-injurious behaviour were noted.

He came to our attention because of worsening in memory function. The facies was particular with long ears, downverted palpebral fissures, bulbous tip of the nose, posterior rotation of the ears with adherent earlobes, overcrowded teeth, scrotal tongue, and high arched palate. He had severe visual loss with a deficit of fixation, bilateral rotary nystagmus, bilateral cataracts, gross retinal atrophic lesions, and bilateral dystrophic changes of the iris.

An IQ of 62 was found on the Wechsler adult intelligence scale. 1 Examination showed cerebellar dysarthria, cerebellar ataxia with limb and trunk ataxia and ataxic gait, mild hypertonia of the lower limbs, hyperexcitable tendon reflexes, and Babinski’s sign on the left. Brain MRI showed a pronounced reduction in size of the vermis and of the cerebellar hemispheres and a mild cerebral cortical atrophy (figure).

An ECG was normal and no signs of muscular or peripheral nerve system involvement were found on EMG.

The second patient is a 48 year old man who also showed a moderately delayed psychomotor development. Walking started at about 5 years of age and, also in this case, ataxia was then noticed. Similarly to his brother, he came under our attention because of worsening in memory function. He had long ears, over-arched palate, thick lower lip, and dorsal hyperkeratosis. Alternating exotropia, horizontal
Pure word deafness after resection of a tectal plate glioma with preservation of wave V of brain stem auditory evoked potentials.

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