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## CLINICAL VIGNETTE

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It is our hope that this section will stimulate dialogue and an exchange of experience. The perspective of "observation" should not be displaced.

The Editors

### AIR-BONE GAP IN PATIENTS WITH X-LINKED STAPES GUSHER SYNDROME

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#### ABSTRACT

It is often possible to elicit the stapedius reflex in patients suffering from recessive X-linked progressive mixed deafness syndrome with stapes gusher. The presence of an air-bone gap in the audiogram and the ability to elicit the stapedius reflex are conflicting. Measurements were performed on two patients who were suffering from this syndrome, to establish whether the hearing loss was of the mixed or purely sensorineural type. It was argued that, owing to congenital malformations, the audio-vestibular system might act as a more than normally efficient transducer, to convert skull vibrations into inner ear fluid motions, leading to bone conduction thresholds that are better than expected. The results of tone and speech audiometry, stapedius reflex measurements, and brainstem evoked response audiometry in this study showed a pattern similar to that generally seen in patients with purely sensorineural hearing loss. This supports the hypothesis that the air-bone gap in the audiogram does not have the usual significance of a conductive hearing loss component.

A stapes gusher is a dreaded complication of stapes surgery.<sup>1-4</sup> Therefore it is important to recognize the potential presence of this complication in advance. In some patients, opening the stapes footplate can result in perilymph gushing: the so-called stapes gusher. Surgical exploration has shown that perilymph is supplied to the vestibulum via the internal auditory canal (IAC) (cerebrospinal fluid), and that insertion of a tamponade into the IAC stops the stapes gusher.<sup>1</sup>

Investigations on the pedigree of a family in which several members had a stapes gusher showed that they were suffering from a recessive X-linked progressive mixed deafness syndrome.<sup>2,3</sup> To date, in this family, nine males from three generations are known to be suffering from this syndrome. Two of them underwent surgery, resulting in a stapes gusher. Polytomography and computed tomography (CT) showed characteristic malformations of the audio-vestibular system in all nine patients.<sup>2</sup> Audiometry

consistently supported the presence of an air-bone gap, which indicated a middle ear component in the hearing loss of these patients. Surprisingly, if the degree of hearing loss was not too great, it was possible to elicit a stapedius reflex: eight of the nine patients were tested, in five of them an evident stapedius reflex was found.<sup>2</sup> Although stapedius reflexes have occasionally been documented in patients with certain types of middle ear disorders,<sup>5</sup> it is an unlikely combination.

Because a stapedius reflex can be elicited, the presence of a conductive hearing loss component may be questioned. Additional support for the hypothesis that the hearing loss is purely of the sensorineural type was found in a previous report in which the speech recognition of some patients with this syndrome was found to be poor, at least poorer than expected on the basis of mixed hearing loss. More specifically, maximum speech recognition scores of between 30% and 70% were found in two patients who had an average hearing loss of 75–90 dB HL with a sensorineural component of 50–65 dB HL.

How can the presence of an air-bone gap in the audiogram of these patients be explained? It may be indirectly related to the reported congenital anatomic malformations of the audiovestibular system, more specifically, the degree of asymmetry between the scala tympani and the scala vestibuli. As described by Tonndorf,<sup>6</sup> stimulation of the cochlea via bone conduction is strongly influenced by this degree of asymmetry; therefore, the conflicting results concerning the presence of a stapedius reflex and the presence of an air-bone gap may be ascribed to the (malformed) audiovestibular system, which acts in these patients as a more than normally efficient transducer, to convert skull vibrations into cochlear fluid motions. As a result, the bone conduction thresholds are better than would be expected, leading to an air-bone gap in the audiogram that does not have the usual significance of a conductive component in the hearing loss. To find out whether or not the hearing loss was purely of the sensorineural type, audiometric tests were performed on two of the affected males of this family suffering from the X-linked progressive mixed deafness syndrome with stapes gusher. These included tone and speech audiometry, the determination of loudness discomfort levels, tympanometry, and stapedius reflex measurements. Auditory brainstem response (ABR) was included because absence of a total wave delay in the ABR recordings would support the present hypothesis.<sup>7</sup>

#### MATERIAL AND METHODS

Pure-tone audiometry was performed using standard procedures and equipment (Interacoustics AC 5 audiometer, calibrated according to ISO 389) (Interacoustics, Assens, Denmark). For the speech performance-intensity curve or speech audiogram, standard Dutch PB word lists were used consisting of

10 monosyllables each. Phoneme scores were obtained. The presentation level is expressed in decibels above the speech reception threshold (SRT) for subjects with normal hearing. The maximum presentation level with the present set-up was 100 dB SRT.

Acoustic impedance and contralateral stapedius reflex measurements were performed with a clinical acoustic impedance meter (Amplaid 720, Amplaid, Milan, Italy). The test signals for the reflex measurements were pure tones of 0.5, 1, 2, and 4 kHz at levels ranging from 80 to 120 dB HL.

For the ABR measurement, silver-silver chloride electrodes were attached to the forehead and the ipsilateral and contralateral mastoids. The ground electrode was placed on the patient's arm. Responses to 1024 stimuli were filtered (100–3000 Hz), averaged, and stored (Medelec ER94a, Medelec, Surrey, England). The stimuli, generated by the Medelec AS 10, were delivered by headphones (TDH-39P with MX 41/AR cushion, Telephonics, Huntington, New York). They consisted of either condensation or rarefaction clicks of 0.1 ms duration. The repetition rate of the clicks was 15 per second and the stimulus level was either 80 or 90 dB nHL. For each test condition, the measurements were repeated at least once. The ABR latencies were compared to normative population values, which were gathered in a previous normalization study. Normative values at stimulus intensities of 50, 70, 80, and 90 dB nHL were available; intermediate latencies were calculated by linear interpolation. Both patients underwent CT.

#### Patients

Patient JK was a 35-year-old affected member of the family with the X-linked progressive mixed deafness syndrome with stapes gusher. At 3.5 years of age, he was diagnosed as having a hearing loss of the mixed type, because of the air-bone gap that was found. The average air conduction threshold at 0.5, 1, and 2 kHz pure-tone average (PTA) of either ear was 50 dB and the average bone conduction threshold at the same frequencies was 30 dB. Hearing aids were fitted. At 14 years of age, he underwent stapes surgery on the right ear. The operation could not be completed because of a stapes gusher. After surgery, a 10-dB increase was found in the air conduction as well as the bone conduction thresholds. Fifteen years later, the right ear was found to be deaf after a traffic accident with head injuries.

In Figure 1, the actual tone and speech audiograms are presented. Mixed hearing loss was found in the left ear, with PTA of 80 dB HL. The right ear was found to be deaf: above 500 Hz, no air conduction or bone conduction thresholds could be obtained up to the maximum stimulation levels of the audiometer (the left ear was masked using the insert earphone). Tympanometry revealed a reduced middle ear pressure of –140 daPa in the left ear and –180 daPa in the right ear. Compliance was within the

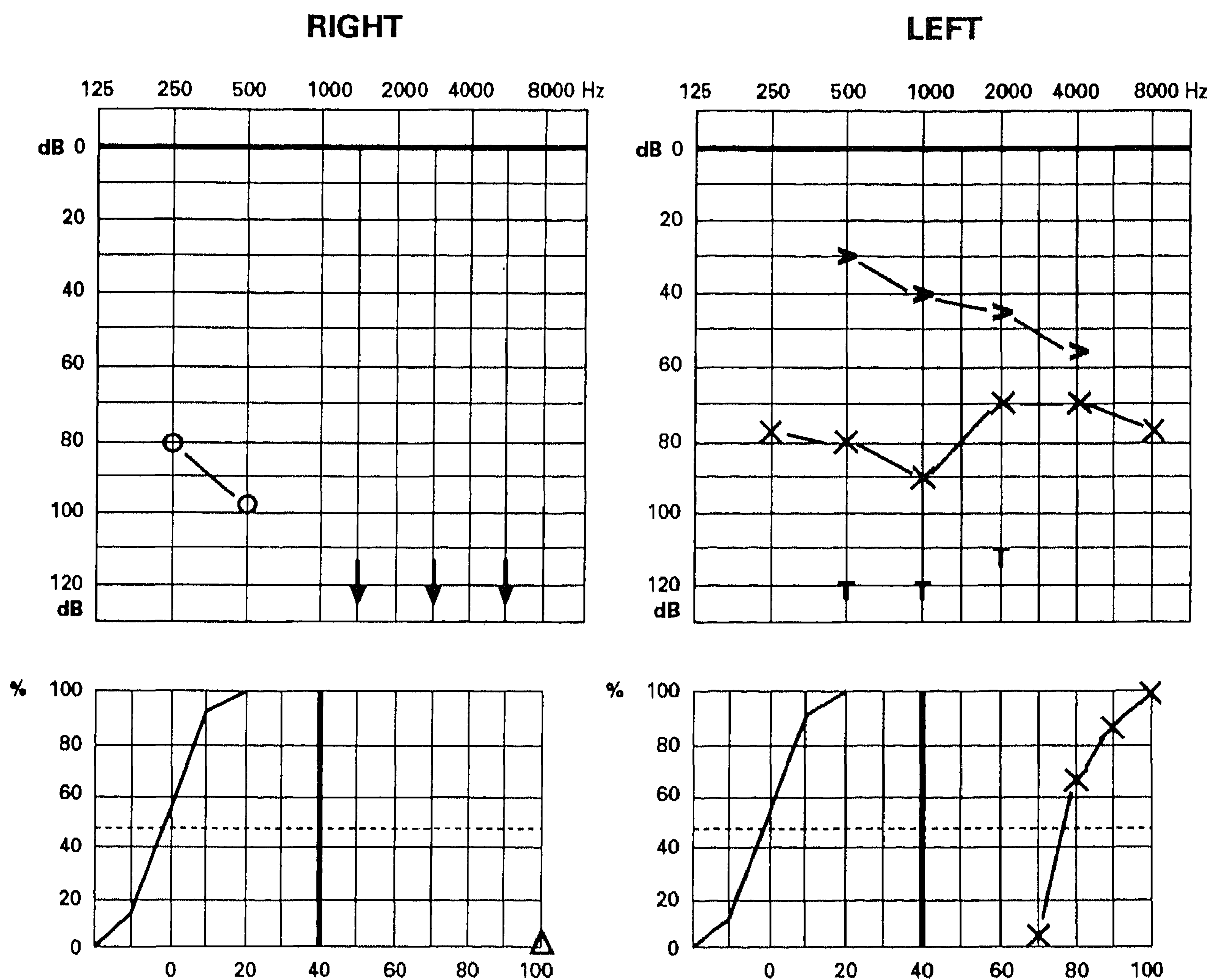


Figure 1. Tone and speech audiograms of patient JK. T refers to the loudness discomfort level.

normal range. The contralateral stapedius reflex of the left ear could not be elicited, owing to the deaf right ear. The SRT in the left ear was found to be in good agreement with the PTA, and a maximum phoneme score of 100% was obtained. The size of the air-bone gap in the frequency region of the ABR click (i.e., the average air-bone gap at 2 and 4 kHz) amounted to 20 dB. Loudness discomfort levels were found at several frequencies.

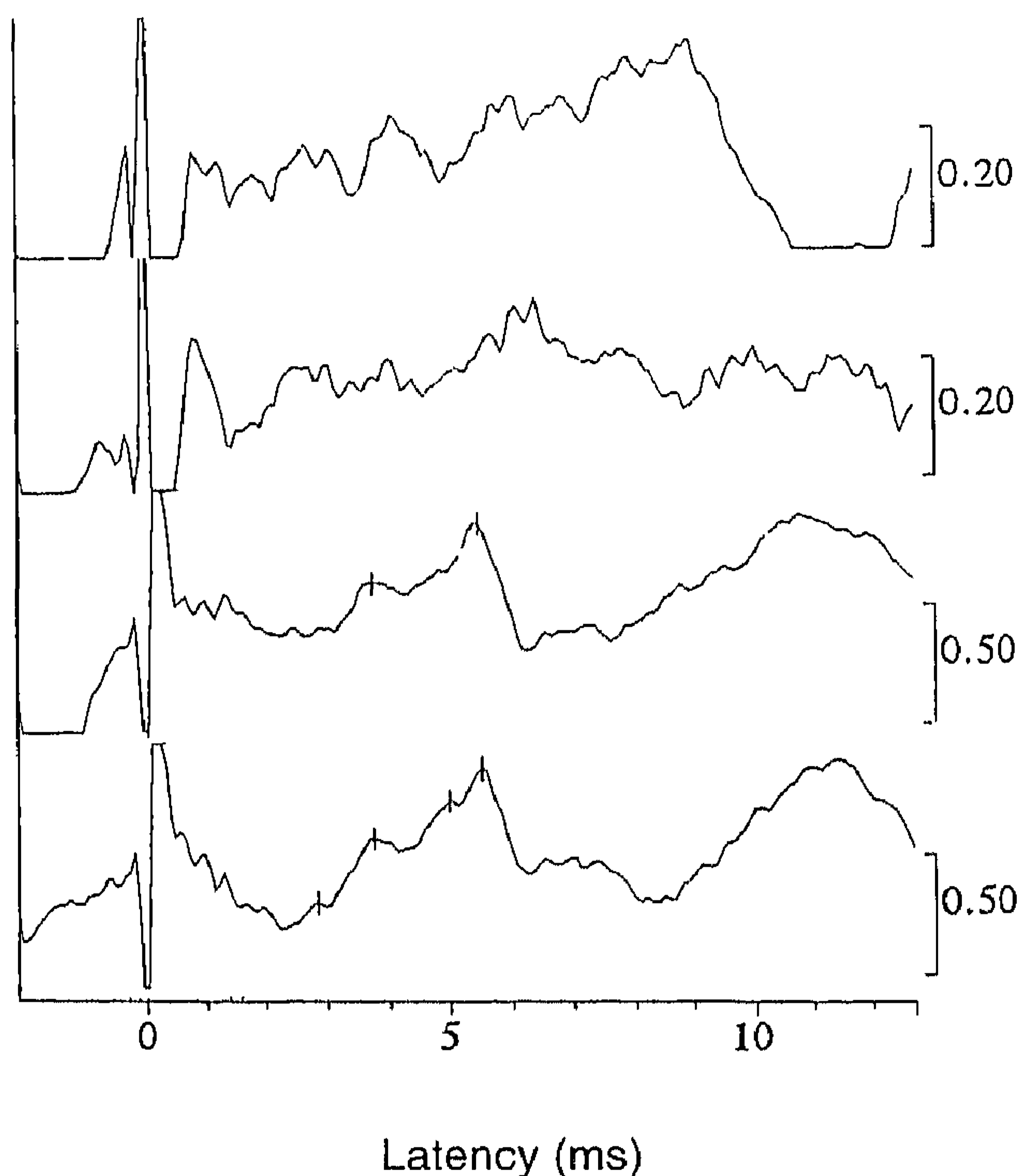
The ABR showed no reproducible responses in the deaf right ear but good reproducible responses in the left ear (Fig. 2). The latencies of waves I, III, and V were found to be within the normal range (mean  $\pm 2$  SD) for all the measurement conditions: rarefaction and condensation clicks at 80 and 90 dB nHL (Fig. 3).

In accordance with previous findings,<sup>2</sup> CT showed small cochlea and lateral dilatation of the internal acoustic meatus.

Patient FP was a 31-year-old cousin of patient JK, suffering from the X-linked progressive deafness syn-

drome with stapes gusher. He did not undergo surgery. Hearing loss of approximately 50 dB was diagnosed when he was 3.8 years of age. In the following years, the hearing loss increased to a level of 75 dB on both sides. He had been using binaural hearing aids since the diagnosis.

In Figure 4 the actual tone and speech audiograms of this patient are presented. Symmetrical hearing loss was noted, with an air-bone gap in both ears, and PTA of 82 dB HL. Tympanometry revealed normal middle ear pressure and compliance. The contralateral stapedius reflex could be elicited at several test frequencies at levels exceeding 100 dB HL. In both ears, the SRT was in fair agreement with the PTA, and the maximum phoneme score (at the maximum output level of 100 dB SRT) was 100% for the right ear and 70% for the left ear. The average air-bone gap at 2 and 4 kHz was 25 dB for the left ear and 22.5 dB for the right ear. No loudness discomfort levels were found up to 120 dB HL. The ABR recordings showed reproducible and normal responses for



**Figure 2.** ABR recordings of patient JK, obtained with rarefaction clicks at 90 dB nHL, showing the first and the verification recordings of the right ear (*upper curves*) and the left ear (*lower curves*).

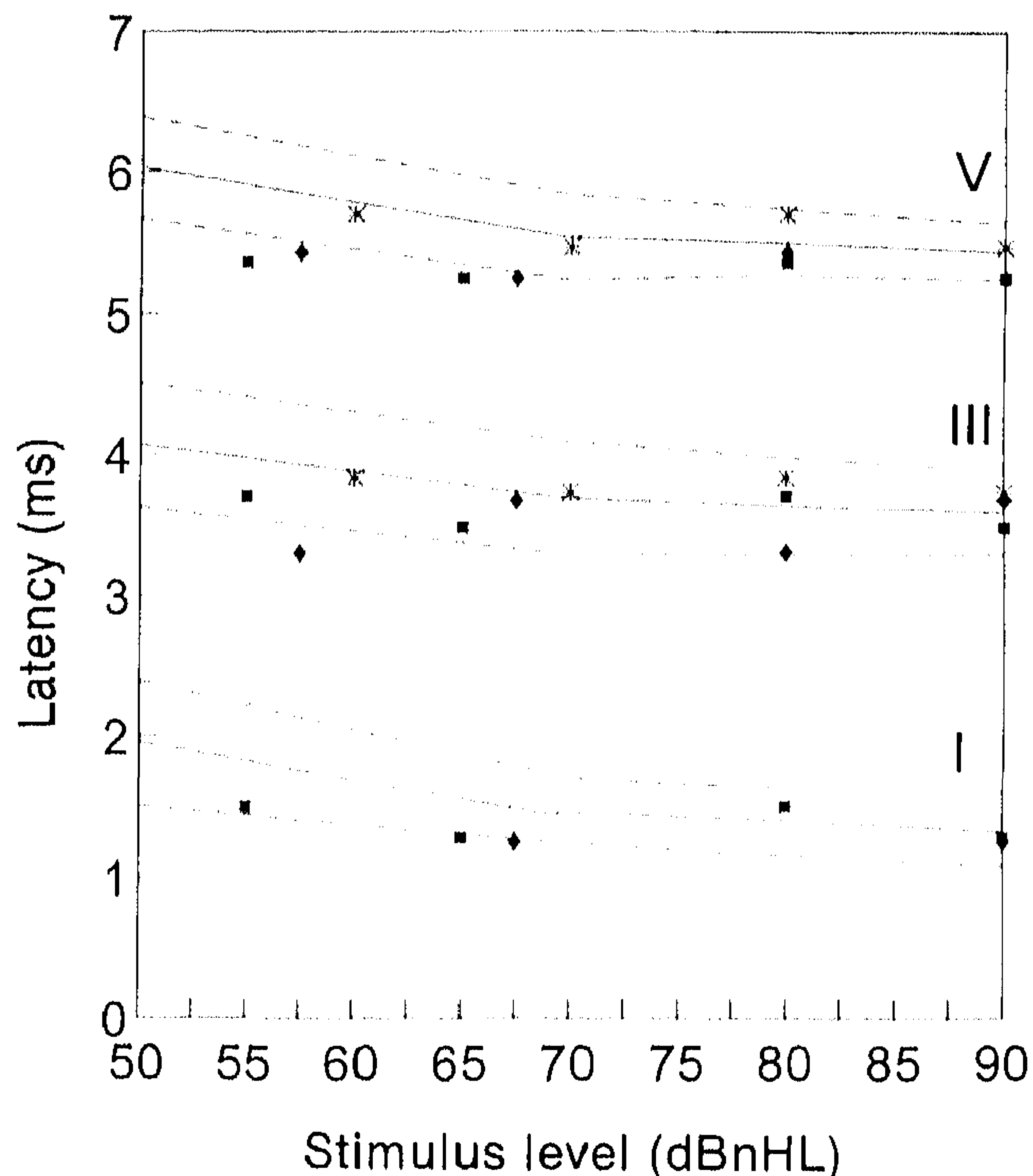
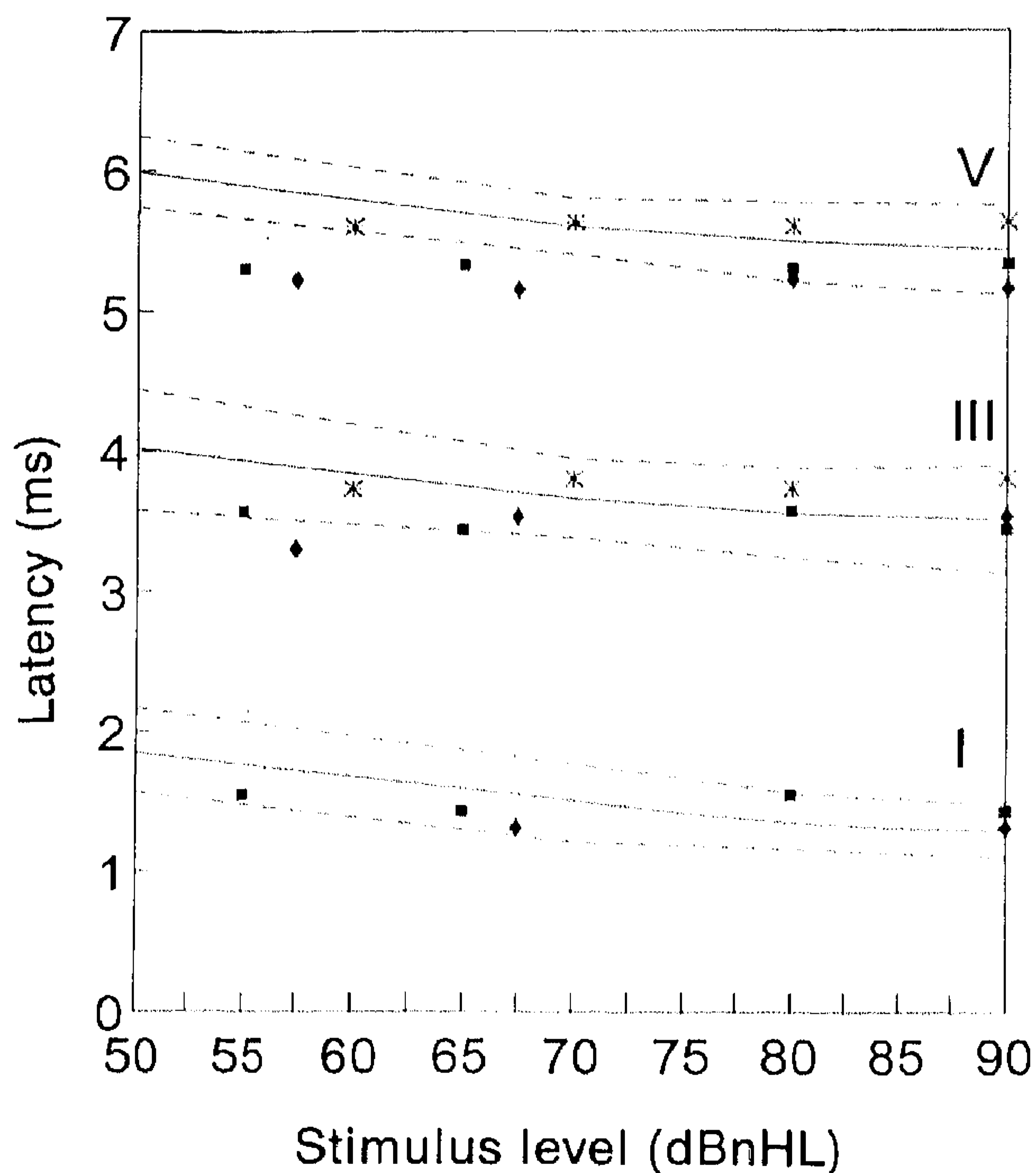
both ears (Fig. 5). The latencies were within the normal range for both ears and in all stimulus conditions (see Fig. 3).

Computed tomography showed the characteristic malformations of the audiovestibular system.

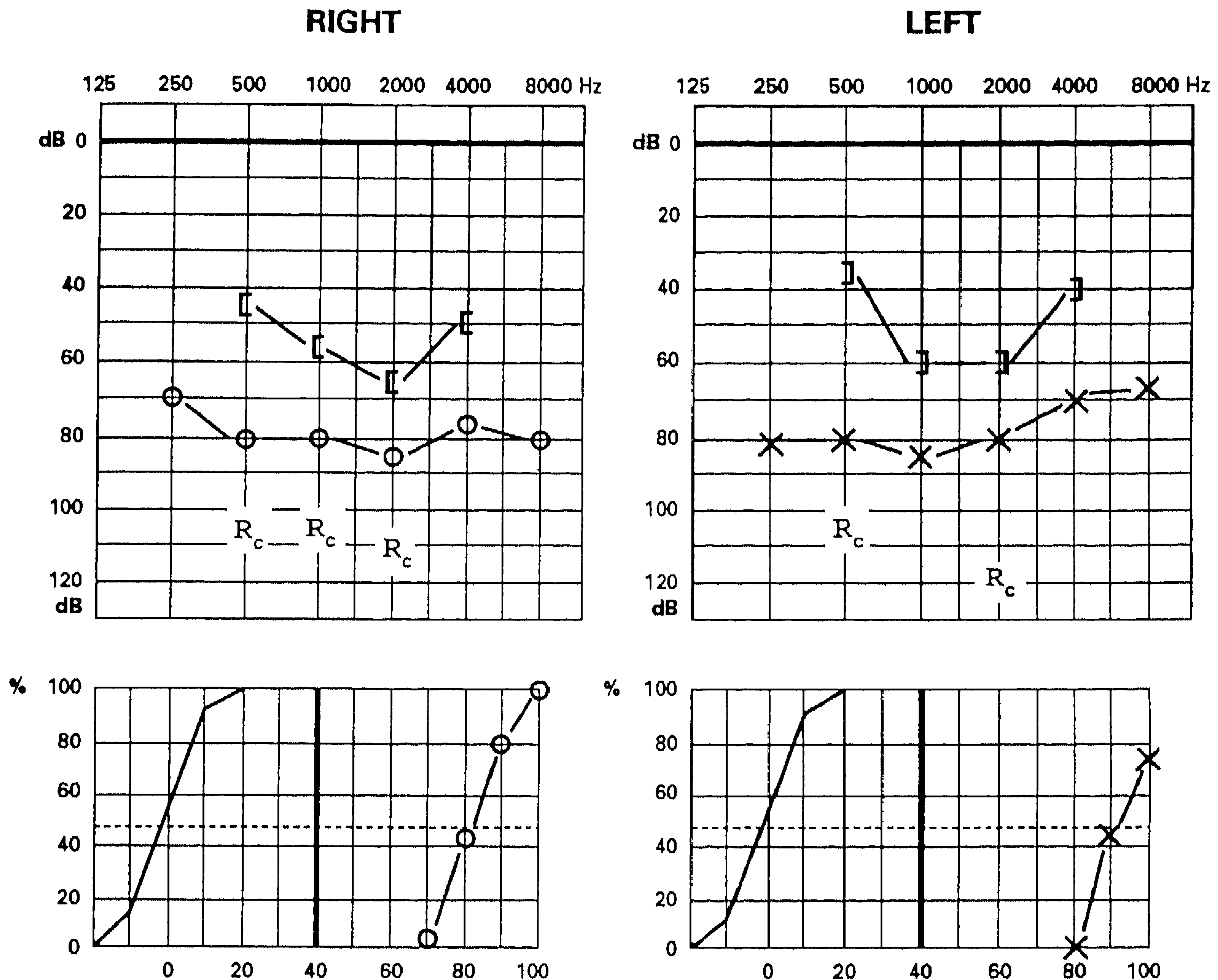
### DISCUSSION

Both patients were suffering from the X-linked progressive mixed deafness syndrome with stapes gusher. In patient JK, an air-bone gap was found, but not a stapedius reflex. This was not surprising because stimulation of the right ear was impossible owing to deafness, and stimulation of the left ear could not elicit a reflex in the right ear, because the stapes tendon had been cut during surgery. Patient FP had the conflicting findings of an air-bone gap and the presence of a stapedius reflex. The ability to elicit the stapedius reflex showed recruitment, as generally found in cochlear lesions. The stapedius reflex thresholds were found at approximately 30 dB SL. This is in agreement with the value expected in sensorineural hearing loss that exceeds 70 dB HL.<sup>8</sup>

To gather more evidence in favor of the hypothesis, the ABR measurement was performed. If the air-bone gap is attributable to a conductive component, the ABR latencies have to be prolonged, depending on the width of the air-bone gap.<sup>7,9</sup> Unfor-



**Figure 3.** ABR latencies of waves I, III, and V of the patients for stimulation by rarefaction clicks (*left*) and condensation clicks (*right*). The full lines represent the normative values for males, the broken lines indicate  $\pm 2$  SD. The patients' ABR latencies are presented twice, once at the actual stimulation level (ASL, 80 or 90 dB nHL) and once at the ASL minus the air-bone gap (corrected stimulation level, or CSL; range, 55–70 dB nHL). The symbols refer to (♦) right and (■) left ear, patient FP; (\*) left ear, patient JK.



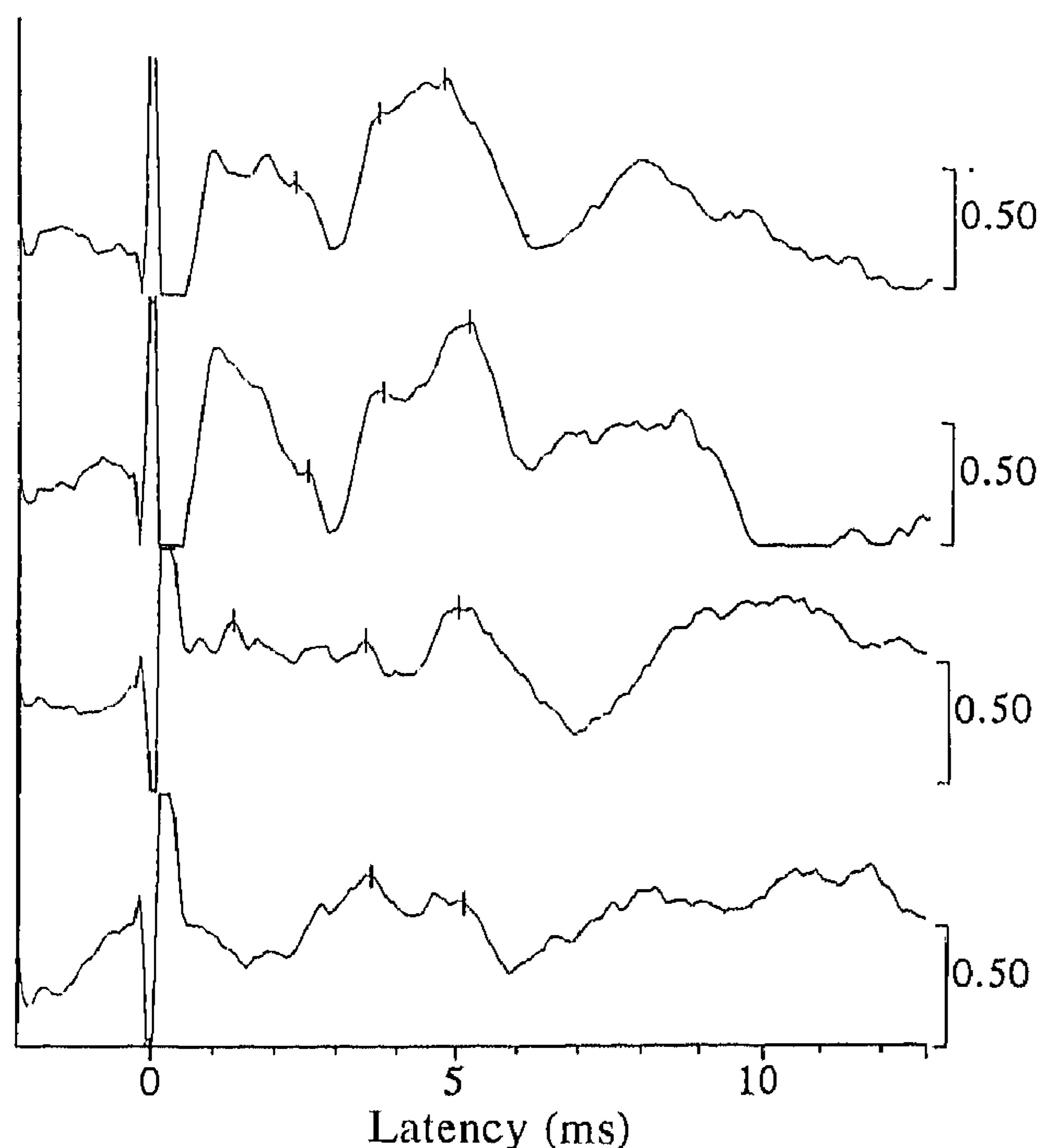
**Figure 4.** Tone and speech audiograms of patient FP.  $R_c$  refers to the stapedius reflex thresholds obtained with contralateral stimulation.

tunately, the air-bone gap is most pronounced in the low frequency region. In the frequency region predominantly stimulated by the clicks normally used during ABR testing (2–4 kHz region<sup>10</sup>), the air-bone gap is small (around 20 dB).

The wave I, III, and V latencies of the ABR recordings of both patients at 80 and 90 dB nHL, presented in Figure 3, were found to be within the normal range. If an air-bone gap exists in the frequency region of the ABR clicks (2–4 kHz region), the latencies should not be compared to the normal latencies at the actual stimulus level (ASL), but to the normal latencies at the stimulation level of the cochlea, namely, the ASL minus the air-bone gap [the so-called corrected stimulus level (CSL)].<sup>7</sup> Therefore, the same latencies are presented twice in Figure 3, once at the ASL and once at the CSL. It can be seen that almost all the latencies at the CSL were below the mean normal values, and several of the latencies were outside the 2 SD region. Table 1 presents the (signed) differences between the measured and nor-

mal latencies for waves I, III, and V, averaged across the subjects, stimulus intensity, and polarity. These differences are presented twice, once calculated at the ASL and once at the CSL. The difference at the ASL deviated nonsignificantly from zero for all three waves. At the CSL, the difference deviated significantly from zero ( $t$  test,  $p < .01$ ) for all three waves. This means that the latencies most closely matched those expected when there was no correction for an air-bone gap. Therefore, the present ABR results support the hypothesis that the hearing loss is purely of the sensorineural type.

The presence of an air-bone gap in the audiogram may be indirectly related to anatomic malformations of the cochlea and vestibulum as indicated previously. One of the modes of stimulation of the cochlea in bone conduction is the compressional response caused by vibrations of the cochlear shell, as described by Tonndorf.<sup>6</sup> The compressional response is strongly influenced by the mobility of the cochlear windows and the degree of asymmetry be-



**Figure 5.** ABR recordings of patient FP obtained with rarefaction clicks at 90 dB nHL, showing the first and the verification recordings of the right ear (*upper curves*) and the left ear (*lower curves*).

tween the scala tympani and the scala vestibuli, including the vestibulum. In patients with a stapes gusher, the vestibulum is often dilatated,<sup>1,2</sup> and the cochlea is smaller than normal. This means that the system is more asymmetric than normal and, in terms of Tonndorf's theory, will act as a more efficient transducer of skull vibrations into cochlear fluid motions. As mentioned, the mobility of the two cochlear windows and of the so-called third window also plays an important part in the compressional response.<sup>11</sup> The third window represents the aqueducts and vascular and neural channels of the cochlea. In the patients with the X-linked progressive mixed deafness syndrome with stapes gusher, the third window is larger than normal (which causes the gusher during stapes surgery). Because blockage of the third window leads to higher bone conduction thresholds,<sup>11</sup> it is supposed that a larger than normal third window may give rise to lower bone conduction thresholds. In conclusion, the conflicting results concerning the presence of a stapedius reflex and the presence of an air-bone gap, as generally found in patients with the X-linked syndrome with stapes

**Table 1.** Average (signed) Differences between the ABR Latencies and Normative Values\*

	<i>Difference in Latencies (ms) of Waves</i>		
	<i>I</i>	<i>III</i>	<i>V</i>
Observations	6	11	12
Stimulus level			
ASL	0.05 ± 0.11	0.05 ± 0.16	-0.07 ± 0.17
CSL	-0.19 ± 0.04 <sup>†</sup>	-0.19 ± 0.19 <sup>†</sup>	-0.34 ± 0.20 <sup>‡</sup>

\*Assuming that the inner ear is stimulated at (a) the actual stimulation level (ASL) and (b) the corrected stimulus level (CSL: ASL minus the air-bone gap).

<sup>†</sup> $p < .01$ ; <sup>‡</sup> $p < .001$

gusher, may be ascribed to anatomic malformations of the audiovestibular system that act as a more than normally efficient transducer of skull vibrations into cochlear fluid motions. As a result, the bone conduction thresholds might be better than expected, causing an air-bone gap in the audiogram that might not have the usual significance of a physically conductive component in the hearing loss.

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