To the editor:

Sir, X-linked adrenoleukodystrophy (X-ALD) is a hereditary metabolic disease with an incidence rate of 1:16,800.1 Mutations in the ABCD1 gene in X-q28 cause the presence of high levels of very long chain fatty acids (VLCFA) in the serum and tissues. X-ALD presents with adrenal insufficiency as well as severe cognitive and neurologic disability. Diagnosis of X-ALD requires the measurement of the concentration of VLCFA in plasma together with genetic analyses. We report a case of a 13 year-old boy with X-ALD who presented initially with only a hearing loss and attention deficit. This report indicates the importance of the word recognition score (WRS) in quiet for diagnosis of X-ALD.

A 13 year-old boy presented with a hearing loss and attention deficit occurring over a two-month period. His routine audiological tests (pure-tone audiometry, tympanometry and word recognition in quiet) indicated normal hearing in the right ear, mild hearing loss in the left ear, and normal middle ear function in both ears. His diagnosis was sensorineural hearing loss, and he was referred to an audiology technician for a hearing aid fitting.

The audiology technician noted that the patient's WRS in quiet was 72% at 60 dB HL for the right ear, which is uncommon for an ear with normal pure-tone thresholds, suggesting the presence of a retrocochlear lesion. Normal cochlear and auditory brainstem function was confirmed from otoacoustic emission (OAE) measurements and the auditory brainstem evoked response (ABR). Poor performance in word recognition in quiet with normal OAE and ABR indicated the possibility of a central lesion after the auditory nerve.

We performed the Mandarin hearing in noise test (HINT)2 with the patient, which adaptively finds the signal/noise ratio (SNR) where only half the sentences are recognized (the speech reception threshold (SRT)). It is a more difficult measure of speech recognition that challenges the entire auditory system and might provide clearer evidence of a central lesion, since the auditory nerve and the cochlea appeared normal. The patient was unable to recognize any of the sentences in the test, even at SNRs well above the average SRT for normals. This result confirmed the likelihood of a central lesion. MRI was performed to diagnose the cause of the lesion. The MRI showed the characteristic pattern associated with X-ALD (Fig. 1). High levels of VLCFA, particularly hexacosanoic acid (C26:0) and tetracosanoic (C24:0), in the patient's plasma (Table 1) and mutations in the ABCD1 gene finally confirmed the X-ALD diagnosis.

Hearing difficulties in the presence of a normal audiogram are common initial symptoms of pediatric male X-ALD patients.3 MRI often provides initial diagnostic evidence4 and determines whether chemistry and genetic testing are needed. However, not all patients with hearing difficulties need MRI, which is costly and time-consuming. The presentation of this patient, both...
hearing loss and attention deficit are nonspecific, they are even common in children and adolescents. But poor performance in speech audiometry is unusual and strongly recommends further examination.

Speech audiometry played an important role in diagnosis of this X-ALD patient, ignoring the abnormality of it further examinations would not have been performed. This case highlights the importance of speech audiometry, more attention should be paid to it, especially the word recognition in quiet.

References

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Fig. 1. MRI shows bilateral symmetrical change in the trigone region of the periventricular white matter, which appear as a butterfly pattern. The lesions involved the occipito-parieto-temporal lobe.

Table 1  
The concentration of VLCFA in plasma.

| VLCFA     | Patient | Normal       |
|-----------|---------|--------------|
| C26:0µmol/L | 1.21    | 0.54–0.80    |
| C24:0/C22:0 ratio | 1.04    | 0.73–0.99    |
| C26:0/C22:0 ratio | 0.037   | 0.007–0.013  |

a Hexacosanoic acid.  
b Tetracosanoic.  
c Docosanoic acid.

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