The Journal of Laryngology and Otology
March 1991, Vol. **105**, pp. 210–212

Clinical Records

Sensorineural hearing loss and the Marinesco-Sjögren syndrome

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Abstract
Sensorineural hearing loss has not been previously reported in patients with the Marinesco-Sjögren Syndrome. Two siblings are described where hearing was initially normal but subsequently deteriorated.

Introduction

Marinesco-Sjögren Syndrome (MSS) is an autosomal recessive disorder characterized by mental retardation, cerebellar ataxia and congenital cataracts. The syndrome was described by Marinesco *et al.*, in 1931 in a sibship with four affected individuals. Subsequently, Sjögren (1950) reported 14 additional cases and established its autosomal recessive mode of inheritance. Although designated MSS, Superneau *et al.* (1985) recently drew attention to an earlier description by Maravscik in 1904 of three affected siblings.

The syndrome, though rare, has been reported from various countries throughout Europe, America and Asia in white and negroid races (Garland and Moorhouse, 1953; Nyberg-Hansen *et al.*, 1972; Yamanaga *et al.*, 1980). Additional features described have included short stature, atrophic depigmented hair and skeletal abnormalities. Hypergonadotrophic hypogonadism has been shown to be associated with MSS and is possibly closely linked (Skra and Berg, 1977).

Reports of language development have indicated that this is delayed in most cases and speech is slow and dysarthric (Sjögren, 1950; Garland and Moorhouse, 1953; Alter *et al.*, 1962; Nyberg-Hansen *et al.*, 1972). Not all case reports have made reference to auditory acuity but in those individuals in whom hearing has been tested there has been no record of impairment (Alter and Kennedy, 1968; Hakamada *et al.*, 1981). Two siblings will be described whose hearing was initially normal but subsequently deteriorated.

Case report

The children (Fig. 1) were the sole offspring of the non-consanguineous marriage of two Caucasians. CH, a male, was born in 1973 and KH, a female, was born in 1975. The father had a normal child from a previous marriage.

Both children were born at term by spontaneous normal delivery. CH weighed 3.7 kg at birth and KH 3.4 kg. CH was noted to have cataracts at the age of four months and KH at birth and subsequently there were removed surgically. Nystagmus was present in both children but particularly marked in the younger.

Development was delayed in all aspects—locomotor, cognitive, linguistic and social. This was evident from early childhood. Sitting was not achieved until nine months (KH) and 18 months (CH) and the children walked at three years. Balance problems were evident, especially in the younger child. Poor growth was apparent in the first two years with heights and weights just below the 3rd percentile. Growth velocity slowed subsequently. Cognitive function was assessed as within the severely retarded range (I.Q. < 50). KH had treatment periodically for asthma but otherwise neither child was receiving regular medication.

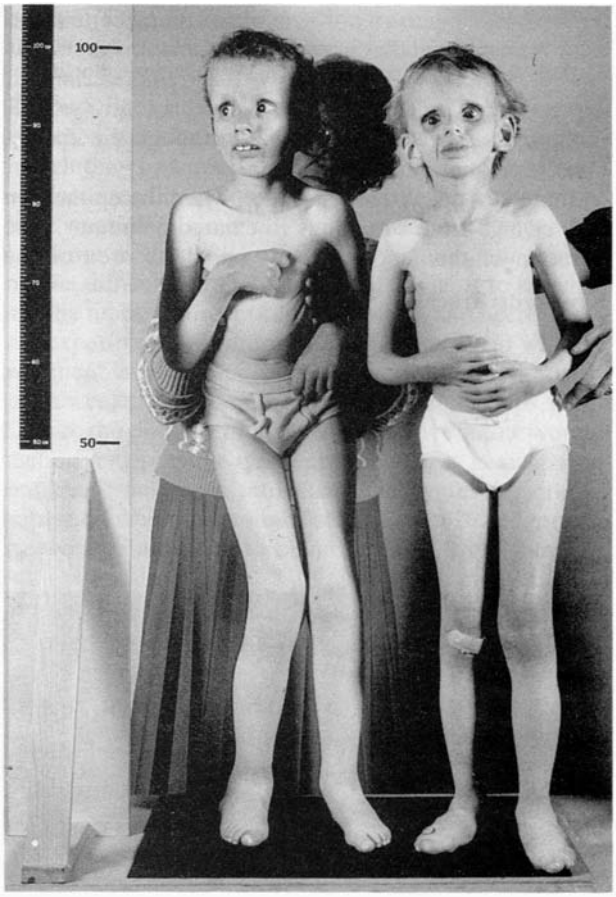

Fig. 1
CH at 14 years and KH at 12 years.

Accepted for publication: 21 November 1990.

Physically the children were of short stature and both had thin facies. Horizontal nystagmus was apparent in both but in KH it was more marked. Both children were partially sighted. Ears were low set and the pinnae appeared normal. There were no aural appendages. Hair was blonde, fine but not unduly sparse. Both were ataxic and walked with a broad-based gait. Physical signs were more marked in the lower limbs than the upper, with reduced muscle bulk, hypotonia and muscular weakness apparent. The cardiovascular and respiratory systems were found to be normal. KH had ankle eversion and flat feet. At 13 years, CH had developed pubic hair and there was an increase in penile length. At 11 years, KH showed no growth of secondary sexual characteristics.

Laboratory tests were normal. These included blood counts, urinary analysis, enzyme studies, hormone estimations, electrolytes and karyotypes.

Speech and hearing

Speech was late appearing with the first meaningful words spoken at 3–4 years and sentences 5–6 years. Their speech was not scanning or dysarthric but both had articulation defects. At the age of 4½ years, CH developed a slight conductive hearing loss which improved after conservative treatment. His hearing remained normal until the age of seven years when it was noticed to have deteriorated. An audiogram showed a bilateral sensorineural hearing loss of 50 dB HL. Auditory brain stem electric response tests were consistent with a loss of cochlear origin. Hearing remained stable until the age of 12 years when there was a rapid deterioration until he had no detectable hearing on the audiometer (Fig. 2).

KH developed a sensorineural hearing loss of 35 dBHL at the age of six years and hearing stabilized until the age of 11 years when it was noticed to deteriorate over a few months. Behavioural tests indicated responses to low frequency stimuli at 85 dBA but no response to high frequency sounds. An auditory brain stem electric response test showed responses at 100 dB HL. Normal waveforms and normal interpeak latencies suggested that the loss was cochlear in origin. Four months later her hearing deteriorated rapidly until no responses could be elicited in behavioural tests or using electric response audiometry. Deterioration of hearing was not associated with pyrexia or with any other neurological changes. Both parents had normal audiograms and there was no family history of hearing loss, mental retardation, cataracts or neurological conditions.

Discussion

The occurrence of a sensorineural hearing loss in both of the children described could indicate pleotropism, genetic linkage or the association of two recessive traits. The probability of a chance association in both children of two unrelated genes would not be high. MSS is rare and, whereas recessive genes causing a deteriorating type of sensorineural hearing loss have been reported, they are infrequently encountered. It would seem unlikely that both parents were heterozygous for both genes.

The absence of previous descriptions of individuals with MSS having a hearing defect does not exclude the possibility of pleotropism or linked genes. Delayed language development has featured in the majority of case reports of individuals with MSS. Poor cognitive ability could have provided adequate explanation for this delay, but in the absence of information in most reports about auditory acuity, it is not known whether hearing impairment could be a reflection of the difficulty of assessing auditory acuity in individuals with severe mental retardation prior to the advent of electric response audiometry. Recent investigations of hearing thresholds of mentally-handicapped populations have indicated a higher prevalence of hearing impairment in these groups than in the general population (Nolan *et al.*, 1980). Nevertheless, in those instances of MSS where auditory function has been recorded, hearing has been normal (Alter and Kennedy, 1968; Hakamada *et al.*, 1981).

The relationship of the sensorineural hearing loss to MSS in the two children described is not yet clear, but the finding indi-

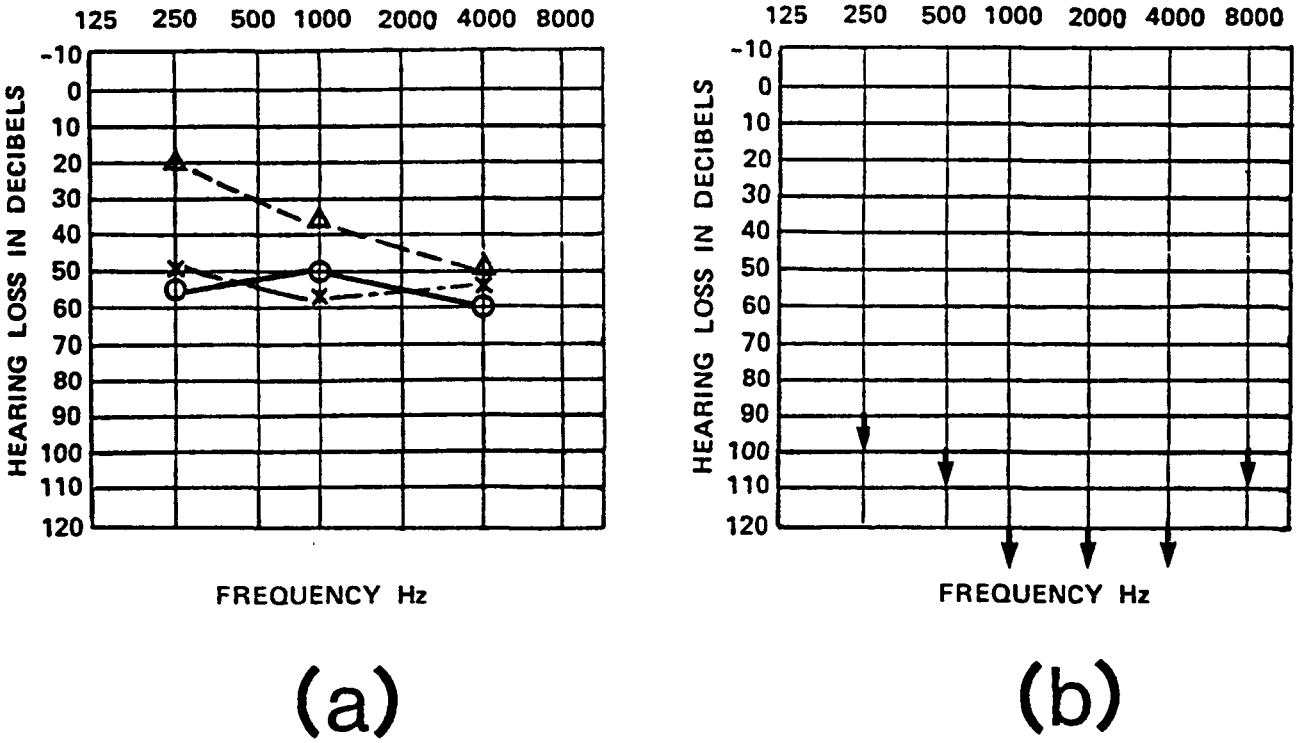

FIG. 2

(a) CH's audiogram at 7 years (O–AC thresholds R ear; X–AC thresholds left ear; △—unmasked bone conduction thresholds). (b) CH's audiogram after deterioration of hearing levels at 12 years.

https://doi.org/10.1017/S0022215100115385 Published online by Cambridge University Press

cates that auditory evaluation should be part of routine management procedure with these patients.

Acknowledgements

The author wishes to thank Dr D. A. Price for medical information. Professor R. Harris for reading the paper, the parents for permission to write about their children and Dr M. Super for the photographs.

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Key words: Deafness, sensorineural; Marinesco-Sjögren syndrome