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Hyperacusis and Williams syndrome

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Abstract
Abnormal sensitivity to environmental sounds is a short-term feature often observed following insertion of grommets. Here we describe a child with this symptom who was found to have Williams syndrome, a condition in which hyperacusis is observed in 95 per cent of patients.

Key words: Middle ear ventilation; Hyperacusis

Introduction

Discomfort in the presence of moderately loud sounds is not an uncommon characteristic of presbycusis. This phenomenon, better known as loudness recruitment is distinct from hyperacusis which implies discomfort or a startled response to sounds of an intensity which would not be considered threatening or loud by the average person. In addition, while loudness recruitment is usually accompanied by a sensorineural hearing loss, most people with hyperacusis have virtually normal hearing.

Williams syndrome is a condition in which hyperacusis is a prominent diagnostic feature.

Case report

A 21-month-old girl was referred to the paediatricians for developmental delay. The child was unable to walk unaided and the GP had noted she looked 'peculiar' and that her speech was poor. She was the first child of normal parents and had been born full-term at normal delivery. The 'odd look' of the child was confirmed by the paediatrician who discovered that the child also had a squint, marked epicanthic folds, drooling of saliva and hyperteleorism. She was found to be a friendly little girl with poor fine motor skills and dysmorphic features. At a chronological age of 23 months she was found to have a performance age of 14 months on a Denver developmental screening test. Her chromosomes were normal 46 XX and no abnormal polysaccharides were found in her urine. Haemoglobin, urea and electrolytes, liver function tests and TSH were within the normal range. Blood and urine amino acids were normal.

With continued care at the child development unit she made good progress and was noted to be playing and running well. She was seen by an ophthalmologist for her strabismus and correction spectacles were prescribed. Her speech improved and she later also developed a recurrent rectal prolapse which required reduction under general anaesthetic. She had some difficulty in swallowing solids which was attributed to bulbar incoordination.

A year later she was referred to the ENT clinic with intermittent otalgia, drooling and snoring accompanied by a clear nasal discharge. Examination revealed dull tympanic membranes and collapsed nasal alae. On a performance test a 35–40 dB loss was found on the left side and impedance testing gave a flat trace on the left but normal responses on the right side. Six months later adenoidectomy and bilateral myringotomies were carried out. Fluid was found on the left side and a grommet inserted. Four months later her mother noticed the child was suffering from 'absences', perhaps from a lack of concentration. Suspecting petit mal epilepsy an EEG was arranged and this proved to be normal. The 'absences' could not be precipitated by hyperventilation.

Six months after surgery the child was reviewed in the ENT clinic and on this occasion her mother mentioned the child's distress in response to ordinary sounds. She was found to clasp her hands over her ears in response to every day sounds including the ringing of a doorbell or telephone. Reassurance was given that the child was probably not used to normal environmental sounds having had hearing loss for a long time. The left grommet was *in situ*, her nasal airway had improved and her continued drooling was noted.

The child was subsequently seen by a geneticist and a diagnosis of Williams syndrome made on the characteristic facial appearance (Figure 1a and b). Her past feeding difficulties were attributed to unrecognized hypercalcaemia. A systolic murmur was found in the aortic area and though a supravalvular aortic stenosis was suspected clinically only trivial tricuspid regurgitation was found on echocardiography. When last seen the grommet had been extruded and an audiogram showed normal hearing on both sides.

Discussion

Williams syndrome is a rare disorder the incidence of which is 1 in 50 000 (Arnold *et al.*, 1985) to 1 in 20 000 (Udwin and Yule, 1988). The facial appearances are characteristically described as being elfin-like and include epicanthus, a wide nasal base and a full nasal tip, a long philtrum and a protruding lower lip (Williams *et al.*, 1961). Other associated abnormalities include cardiovascular abnormalities, usually supravalvular aortic stenosis, prenatal and postnatal growth retardation, developmental delay and occasionally infantile hypercalaemia (Beurin, 1972; Jones and Smith, 1975). A stellate pattern is often noted in the irides of these patients, and is said to be of diagnostic significance (Holmstrom *et al.*, 1990). Other eye findings include epicanthic folds, short palpebral fissures, strabismus, a median eyebrow flare, megalocornea and a preponderance of blue eyes (Stewart and Prescott, 1976; Preus, 1984). Oral manifestations include missing teeth, microdontia, enamel hypoplasia and

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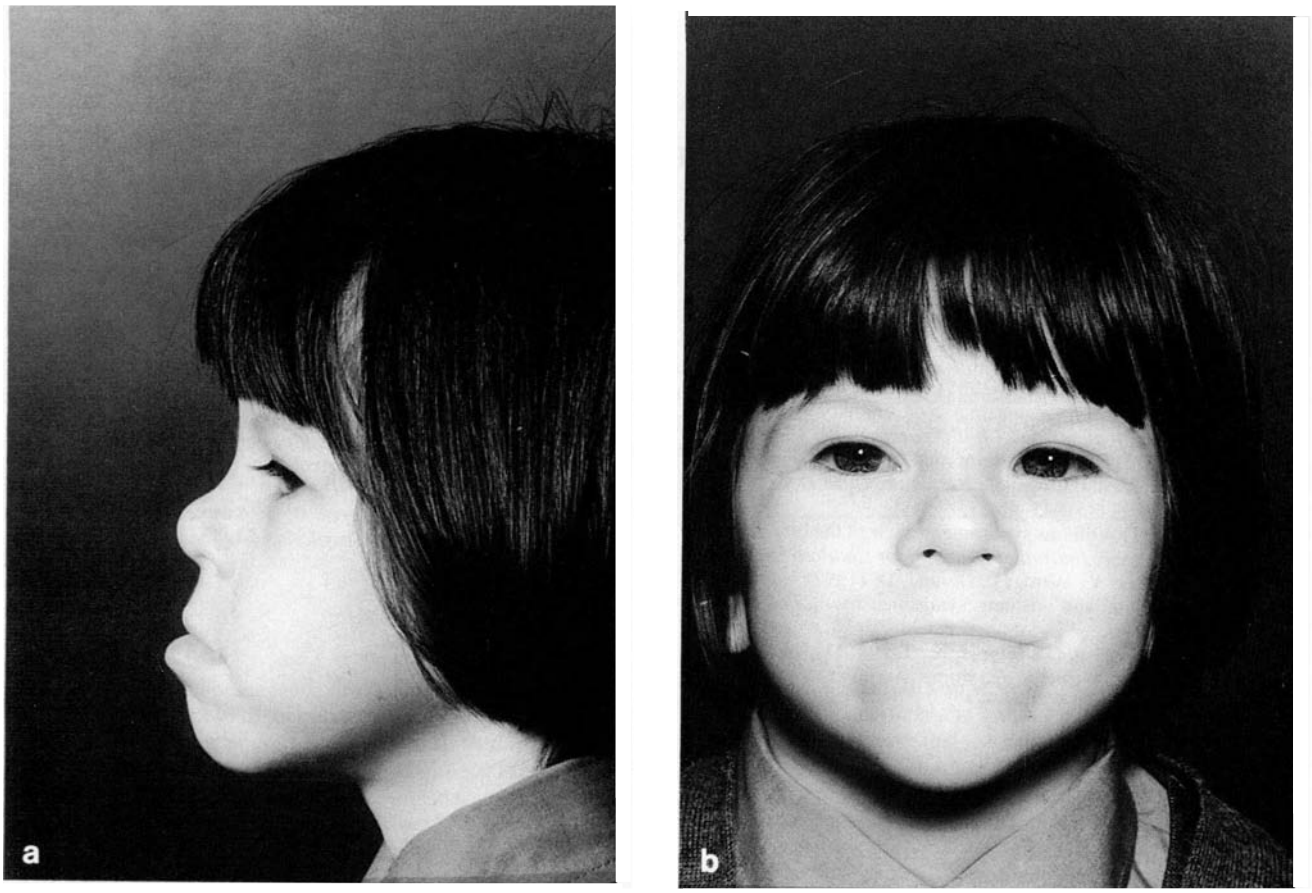

FIG. 1

(a) Side view of characteristic facial appearance of Williams syndrome; (b) front view.

caries (Boraz, 1991). The ears of patients with Williams syndrome are said to be large protruding and giving the illusion of being low set (Kelly and Barr, 1975). The mental status varies from mild to moderate retardation. Other features reported are unreasonable anxiety, poor peer interaction, pervasive hyperactivity, excessive demands for attention and maladaptive behaviour. A child with Williams syndrome is typically socially outgoing, friendly and loquacious (Klein *et al.*, 1990).

Most cases of Williams syndrome have been sporadic although the few reports of familial occurrences have led to speculations about dominant X-linked, multifactorial or heterogenic causality. The cause is unknown and few cases of chromosomal abnormalities have been identified (Colley *et al.*, 1992). Most of the research into the aetiology of Williams syndrome has centred around vitamin D sensitivity and impaired calcitonin secretion. Calcitonin-gene-related product is an important neuropeptide which may be responsible for some of the cardiovascular and central nervous system features observed in Williams syndrome (Jones, 1990). The diagnosis in the absence of hypercalcaemia can be difficult to establish and a diagnostic index using 50 characters has been said to have an accuracy of 99 per cent (Preus, 1984).

Nearly 95 per cent of patients with Williams syndrome suffer from hyperacusis (Klein *et al.*, 1990). Audiological investigations into the cause of hyperacusis are hampered by the difficulty in performing threshold audiograms or speech discrimination in patients who are either too young or uncooperative because of developmental delay. However it has been shown that 83 per cent of patients with Williams syndrome were reported to be frightened or bothered by sounds against three per cent in a control group (Klein *et al.*, 1990). Hyperacusis is not a phenomenon peculiar to Williams syndrome alone but has been noted in several patients with developmental delay caused by other genetic disorders. In a survey of 100 children undergoing bilateral insertion of grommets for otitis media with effusion at this hospital, 47 per cent of children reported hyperacusis of varying severity which lasted for two to 40 days. The sounds causing distress in Williams syndrome vary from a firecracker or fire engine siren to the ringing of a telephone. This hyperacusis may be severe enough to prevent their acceptance of normal travel or household activities such as vacuum cleaning or mowing the lawn. A feature that characterizes Williams syndrome is the behavioural response to loud sounds. Reactons vary from covering the ears with hands and crying or cringing. The mechanism for hyperacusis is not known and the proposed causes include hyperacuity of hearing, distortion in the neural coding of the auditory input that may cause abnormal growth in loudness, or failure of the central nervous system to habituate the startle response. Evidence of calcification in the labyrinth has been reported in one patient (Beurin, 1972).

The incidence of otitis media is also said to be higher in children with Williams syndrome (Klein *et al.*, 1990). In this study which involved a questionnaire survey none of the parents reported permanent hearing loss. No relationship could be established between the degree of hyperacusis and the frequency of otitis media. Hyperactivity in Williams syndrome may be linked to the prevalence of otitis media.

The management of hyperacusis in these children requires a rearrangement of household routine so that noisy activities can be avoided when the child is present. Ear plugs may be worn if the child is quite distressed. The plugs may impede speech development and their use should be for short periods only. Equipment for use in the house should be selected according to its noise level. Behavioural conditioning techniques have been described and are said to be quite successful (Meyerson and Frank, 1987). If repeated exposure to a particular sound is allowed with the child exercising some control over the source, for example a vacuum cleaner, the child's fear may diminish. A

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tape recording of sounds can also be quite useful in this regard by gradually increasing the volume of a distressing sound. Regular audiological investigations are required when otitis media is detected and grommets inserted when indicated.

Conclusion

Williams syndrome is a rare condition in which hyperacusis is a common symptom. Other congenital anomalies are often associated and the diagnosis is usually based on the characteristic facial appearance. Relief from distressing hyperacusis can be obtained by gradual, controlled introduction to loud sounds.

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