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Vestibular nerve dysfunction and decompression in Engelmann's disease

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Abstract
Vestibular nerve dysfunction as the major neuro-otological symptom in one of the osteopetrosis group of bone disorders is unusual. We describe a patient with Engelmann's disease who presented in this manner and who benefited from an eighth nerve decompression procedure. Surgical decompression of the internal auditory meatus to relieve vertigo has to our knowledge not been previously reported in this condition.

Key words: Osteopetrosis; Vestibular diseases

Introduction

Progressive diaphyseal dysplasia or Camurati-Engelmann disease is a rare genetic bone disorder characterized by progressive sclerosis of the diaphyses of the long bones, associated with a marked thickening of the skull. It was first described by Engelmann in 1929, and is predominantely inherited by an autosomal dominant mechanism. It is part of the osteopetroses group, all of which show abnormality of skeletal modelling and increased bone density (Table I) (Brookes and Booth, 1987). The genetic craniotubular hyperostoses differ slightly in that the main problem is bony overgrowth rather than defective modelling.

A common finding in all osteopetroses is the involvement of the skull base. Temporal bone dysplasia with dysfunction of the facial and eighth nerves can be the first indication of a bone disease or the cause of referral to the neuro-otologist of a patient in whom the diagnosis has already been made. Facial nerve palsy is usually the main presenting feature, and often responds well to surgery (Hamersma, 1977; Hamersma and May, 1986; Benecke, 1993). We present a case of Engelmann's disease in which the vestibular symptoms due to bony narrowing of the internal auditory meatus (IAM) were predominant and were successfully treated by surgical decompression. To our knowledge this is the first such case in the English literature.

Case report

A 31-year-old woman with a long standing diagnosis of Engelmann's disease was referred by her neuro-opthalmologist in 1991. She was being regularly reviewed on account of a mild visual disturbance due to bilateral low grade papilloedema. This was static and presumed to be either secondary to raised intracranial pressure or direct optic nerve compression, caused by bony overgrowth of the skull base. The main neuro-otological symptom was that of dizziness, comprising true rotational vertigo and imbalance with a tendency to fall to the left side. These episodes had begun two years previously and occurred sporadically, often associated with headache, and lasting minutes to a few hours. There was bilateral moderate fluctuating hearing loss and associated left tinnitus.

On examination, the tympanic membranes and facial nerve function was normal. She had an abnormally shaped skull with a degree of right ocular prominence which did not involve her eye movements in that she had normal convergence, gaze, saccadic and pursuit movements. Ocular counter rolling was normal, as was opto-kinetic nystagmus to a rotating drum. When performing Romberg's test she felt very unsteady with her eyes closed, though she did not fall to the side. Her gait was compromised by arthralgia affecting the left leg. She became ataxic on eye closure, with a tendency for the left leg to abduct. On attempted tandem gait she tended to fall to the left. She was unable to perform Unterberger's test because of her arthritis, but taking small step gaits she tended to rotate to the left with eye closure. On Hallpike positional testing with the head in the right-down position she developed rotational vertigo, after a latent period, not accompanied by positional nystagmus, but unusually associated with a rushing noise in the right ear, which resolved when the vertigo resolved. In the left head down position there was no positional nystagmus.

TABLE I
OSTEOPETROSIS GROUP OF BONE DISORDERS

Osteopetrosis
Benign, dominant form (Albers-Schonberg disease)
Malignant, recessive form
Genetic craniotubular dysplasias
Craniometaphysial dysplasia
Craniodiaphysial dysplasia
Frontometaphysial dysplasia
Dysosteosclerosis
Genetic craniotubular hyperostoses
Hyperostosis corticalis generalisata
Sclerosteosis
Congenital hyperphosphatsaia
Progressive diaphysial dysplasia (Engelmann's Disease)
Osteopathia stiata with a sclerotic skull

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FIG. 1
Coronal CT scan of the temporal bones illustrating gross bony overgrowth of the IAMs more marked on the left.

Audiometry revealed a right-sided mixed hearing loss of 70 dB in the lower frequencies and 30 dB in the higher, and a flat 30 dB sensorineural hearing loss in the left, with a 50 dB dip at 4 kHz. The stapapedial reflexes were absent bilaterally. Auditory brain stem evoked responses were absent on the left and significantly delayed on the right side. Electronystagmography (ENG) revealed no evidence of spontaneous nystagmus in the dark. Vestibular ocular reflexes were of low amplitude but symmetrical with complete suppression on fixation. She had good optokinetic and smooth pursuit eye movements. Caloric testing showed bilateral labyrinthine hypofunction more marked on the right side, and a right directional preponderance with enhancement in the absence of optic fixation. These findings were consistent with the presence of bilateral retrocochlear pathology due to eighth nerve compression, more marked on the left side. Some conductive loss probably due to ossicular involvement was also present in



FIG. 2
Post-operative coronal CT scan illustrating the extensive bony decompression.

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the right ear. Computed tomography (CT) of the temporal bones showed gross bony overgrowth of the skull base with markedly narrowed internal auditory canals (IAM) (Figure 1).

The possibility of surgical decompression of the IAMs was discussed, but the patient declined preferring to continue on medical treatment with low dose prednisolone. Over the next year, the episodes of vertigo and unsteadiness became more frequent and severe and were unresponsive to all medical therapy. She began to experience momentary sensations as if she was suddenly being pushed to the left side. The left tinnitus became louder intermittently, sometimes several times a day and was associated with increased dizziness and oscillopsia. Audiometry showed no deterioration in hearing and facial nerve function remained normal. In view of her disabling vestibular symptoms surgery was reconsidered and in 1993 exploration of the left vestibular nerves was carried out via a retrosigmoid approach. At operation the IAM was found to be extremely narrowed with new bone formation and the neural structures compressed in a slit-like meatus. The sclerotic bone in this disease is largely avascular, and, as expected, no untoward technical difficulties were encountered at operation, and wide decompression was achieved as illustrated by the post-operative CT scan (Figure 2).

Post-operatively there was an immediate improvement in the vestibular symptoms. This continued over the next year of follow-up, with some slight residual unsteadiness, possibly due to her impaired pre-operative contralateral labyrinthine function. Post-operatively there was initially some increased high frequency loss in the left ear. We suspected that this could well have been due to the local temperature changes induced by persistent drilling with a diamond burr adjacent to the dural sheath of the IAM overlying the cochlear nerve. An alternative explanation is that the operative loss was due to acoustic trauma from the drilling. Osteopetrotic bone is more dense than normal bone and is therefore more acoustically efficient in transmitting the sound energy from the drill to the cochlea. This improved over the next year resulting in a residual significant increased hearing loss only at 8 kHz. There was no significant change in the speech discrimination or speech reception threshold following surgery. Eighteen months post-operatively she is well and under occasional review.

Discussion

The osteopetrosis group of bone diseases comprises a number of similar but heterogeneous dysplastic and hyperosteotic osseous conditions. Engelmann's disease is characterized by the progressive nature of hyperostosis of the diaphyses of the long bones especially the femur and tibia. The base of skull is usually affected by bony overgrowth, with less severe involvement of the calvarium. Often there are associated neuromuscular abnormalities with muscular weakness and a broad based waddling gait. Familial studies of severely affected individuals have shown that there is probably variability in the extent of the disease, with some members having no clinical symptoms, but only diagnostic radiological changes in the long bones (Sparkes and Graham, 1972). Onset of the disease is usually in childhood in the severe form, but the diagnosis is often made later on in life in less affected individuals. Inheritance appears to be suggestive of an autosomal dominant trait, with the possibility of new mutations causing some of the cases (Sparkes and Graham, 1972). In this case the patient's parents had mild hand arthritis, and her father fibrosing alveolitis, but no other stigmata of the disorder. The aetiology of the disease is one of bony hyperostosis rather than defective modelling. This may be explained by the decreased number of osteoclasts that have been noted by some investigators (Allen *et al.*, 1970).

Neuro-otological symptoms from bony overgrowth around the internal auditory meatus (IAM), causing seventh and eighth nerve dysfunction, can be the first symptom of an osteopetrotic group disorder. Cranial neuropathies in general occur in seven to 15 per cent of these patients, with the optic nerve most commonly affected, and secondly, the facial nerve (Johnston *et al.*, 1968). The cochlear portion of the eighth nerve usually gives rise to more symptoms than the vestibular division. Hearing loss can be both sensorineural, due to nerve compression in the IAM, and conductive, secondary to ossicular involvement, overgrowth of the round or oval window, or narrowing of the external meatus. These patients are known to have reduced labyrinthine function on vestibular testing, but clinically this is usually less problematic (Hamersma, 1977).

Two mechanisms of neural dysfunction secondary to bony hyperostosis of the temporal bone around the IAM have been suggested. One theory is that direct compression from involved bone causes a gradual loss of function eventually resulting in total paralysis. Alternatively, a vascular mechanism may be responsible. Venous occlusion leading to nerve oedema, might cause compression and blockage of the arterial supply in the meatus and thereby neural ischaemia. Any cause of neural oedema, such as a viral infection, may also lead to arterial compromise and ischaemic damage (Miyamoto *et al.*, 1980). Both these mechanisms may occur concurrently, and could explain the occurrence of fluctuating as well as sudden seventh and eighth nerve palsies.

Gradual nerve dysfunction rarely responds well to medical therapy such as steroids. Some form of decompressive surgery is often needed. Facial nerve palsy is the most frequent indication for neuro-otological intervention. A review of the literature reveals four cases of Engelmann's disease which have required neuro-otological decompression of the IAM. Miyamoto *et al.* (1980) describe two cases; a 26-year-old man with a progressive right sided facial palsy, right-sided total and left-sided partial hearing loss. Bilateral middle cranial fossa explorations were carried out and slit-like IAMs decompressed. The facial palsy improved as did the left-sided hearing loss initially, but the hearing gain was not sustained. The authors mention some vertigo, but do not comment further. The second case was a 30-year-old woman with bilateral progressive hearing loss and no vertigo or facial palsy. Neural decompression via a middle cranial fossa approach stabilized the hearing. The authors state that this was the first such decompression for hearing loss to be performed for the osteopetroses. Sparkes and Graham (1972) report a 26-year-old man with a right facial palsy and deafness who benefited from decompressive craniotomy surgery. The approach and the possible presence of vertigo were not discussed. Benecke (1993) describes a 57-year-old man with a fluctuating left facial palsy and progressive left sensorineural hearing loss. A middle cranial fossa approach prevented further facial weakness and decline in hearing. Once again vestibular symptoms were not mentioned.

When there is facial nerve weakness a middle fossa approach is clearly the procedure of choice. This will facilitate optimum decompression of not only the facial nerve in the IAM, but also its narrow intra-labyrinthine segment which may be the primary site of neural compression. The nature of the skull base bony over-

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growth will, however, inevitably cause technical difficulties due to the loss of bony land marks and reduced space for supra-tentorial access. This will often necessitate increased temporal lobe retraction with an attendant increased risk of epileptic sequelae. Because the vestibular nerves occupy the posterior IAM compartment, we suggest that retro-sigmoid access is the approach of choice for patients with primary vestibular symptoms. Anatomical identification is facilitated by direct visualization of the neural structures as they exit at the porus and pass to the brain stem.

This case is the first, to our knowledge, where the vestibular symptoms have been the primary cause of neuro-otological presentation, and surgical decompression has been undertaken. Hearing loss was present but was less problematic and both sensorineural and conductive in character, undoubtedly for the reasons describe above. IAM decompression for facial nerve palsy in the osteopetroses has been documented (Hamersma, 1977; Hamersma and May, 1986; Benecke, 1993), with a favourable outcome. This case suggests that similar results may be attainable for vestibular symptoms, if problematic. However longer follow-up is needed, as the progressive nature of Engelmann's disease may alter the ultimate outcome.

References

Allen, D. T., Saunders, A. M., Northway, W. H., Williams, G. F., Schafer, I. A. (1970) Corticosteroids in the treatment of Engelmann's disease: progressive diaphyseal dysplasia. *Paediatrics* **46:** 523–531.

Benecke, J. E. (1993) Facial nerve dysfunction in osteopetrosis. *Laryngoscope 103:* 494–497.

Brookes, G. B., Booth, J. B. (1987) Diseases of the temporal bone In *Scott-Brown's Diseases of the Ear, Nose and Throat* 4th Edition. Butterworth Scientific, London. pp 340–380.

Engelmann, G. (1929) Ein Fall von Osteopathia hyperostotica (sclerotisans) multiplex infantalis. *Fortschritte auf dem Gebiete der Röntgenstrahlen und der Nuklearmedizin* **39:** 1101–1106.

Hamersma, H. (1977) Facial nerve paralysis in the osteopetroses In *Facial Nerve Surgery.* Aesulapius Publishing Co. Birmingham, Ala. pp 555–572.

Hamersma, H., May, M (1986) Osteopetrosis and facial palsy In *The Facial Nerve.* (May, M. ed.), Thieme Inc., New York, pp 469–476.

Johnston, C. C., Lavy, N., Lord, N., Vellios, F., Merritt, A. D., Diess, W. P. (1968) Osteopetrosis, *Medicine* **47:** 149–167.

Miyamoto, R. T., House, W. F., Brackmann, D. E. (1980) Neurotologic manifestations of the osteopetroses. *Archives of Otolaryngology* **106:** 210–214.

Sparkes, R. S., Graham, C. B. (1972) Camurati-Engelmann disease. Genetics and clinical manifestations with a review of the literature. *Journal of Medical Genetics* **9:** 73–85.

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