

**TITLE: UNUSUAL OTOLOGICAL MANIFESTATIONS IN
CAMURATI-ENGELMANN'S DISEASE**

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ABSTRACT

Camurati-Engelmann's disease (CED) is a rare hereditary disorder affecting mainly the diaphysis of long bones but multiple cranial nerve deficits may also develop secondary to bony sclerosis of their foramina, including visual loss, facial palsy, deafness, vestibular disturbances and sensory deficits along the distribution of the trigeminal nerve. Deafness has been reported in about 18% of these cases due to narrowing of the internal auditory canals caused by bony encroachment on nerves and vessels. We report an extremely rare case of a patient with CED who presented with deafness due to gross abnormalities affecting both middle ear and cochlea. The issues relating to the management of these patients with temporal bone involvement are discussed.

Keywords: Camurati-Engelmann's disease, Hereditary deafness, internal auditory canal

INTRODUCTION

Camurati-Engelmann's disease (CED) or progressive diaphyseal dysplasia (PDD) is a rare hereditary disorder primarily characterized by symmetrical diaphyseal sclerosis and fusiform enlargement of long bones as well as cranial hyperostoses, particularly at the skull base. This condition was first reported by Camurati in 1922 and further delineated by Engelmann in 1929.^{1,2} Neuhauser *et al* (1948) coined the term progressive diaphyseal dysplasia, stressing the diaphyseal location and the progressive features of the disorder.³

The clinical manifestations of CED usually begin between the ages of 4 and 10 years and equally affect both sexes, with no racial predilection.⁴ The clinical triad of bone pain, muscle weakness and unsteady gait are frequently cited as the most common features.⁵ Although the femur, tibia and humerus are the most commonly affected sites, the cervical, thoracic and lumbar vertebrae can be involved leading to limitation of spinal mobility.⁶

The skull and facial bones are involved in 50% of case.⁶ Neurological symptoms are caused by hyperostosis and sclerosis of the skull base, with stenosis of the basal lamina and orbital apices, which can lead to compression of the cranial nerves.^{4,7,8} Another possibility is that the blood supply to the cranial nerves can be impaired producing venous occlusion and neural oedema, resulting in ischemia of the nerve.⁴ Cranial nerve involvement has been described in relation to the optic nerve,⁷ auditory nerve,^{6,9} trigeminal nerve,¹⁰ and facial nerve⁶, leading to visual impairment, hearing loss, facial numbness, and facial palsy respectively. Sensorineural deafness has been reported

as a result of bone dysplasia narrowing the internal auditory canal (IAC) and compressing the eighth nerve complex ¹¹.

We report a patient with this disease who presented with deafness. The otological manifestations of CED are reviewed and issues relating to the management of these patients with temporal bone involvement are discussed.

CASE REPORT

A 15 year-old girl with CED, was referred to Addenbrookes Hospital, Cambridge for management of severe progressive deafness which was first detected at the age of three. She was born 6 weeks prematurely. Subsequently she had a long history of intermittent joint pain and associated joint swelling in her knees, elbows and right wrist, myopia and recent onset of severe dental problems. There was no family history of bone deformities. She attended a special unit at school because of her deafness, learning difficulties and behavioural disturbance. Her hearing progressively deteriorated in both ears and she was fitted with bilateral hearing aids. The continued deterioration in her hearing resulted in amplification being less useful and she was at risk of become totally deaf.

Examination revealed dysmorphic facies with mid facial and mandibular hypoplasia, abnormal dentition and some dental loss. Distal and proximal interphalangeal bony swellings were noted, with short incurving little fingers and mild camptodactyly. There was bilateral reduction in hip rotation and knee extension. Generalised muscle weakness and a waddling gait were noted but neurological examination was normal. Facial nerve function was normal as was Romberg's and Unterberger's stepping tests. Otological examination demonstrated intact tympanic membranes with extensive tympanosclerotic plaques.

Pure tone audiometry revealed bilateral mixed hearing loss with air conduction thresholds averaging 98db in the right ear and 85 db in the left, and bone conduction averages of 45dB in both ears. Aided maximum speech discrimination at 60-70dB was

80% levels with lip reading and 47% without lip reading. Tympanometry showed a type B trace on the right side and a type A trace on the left side.

Laboratory tests including erythrocyte sedimentation rate (ESR), C - reactive protein (CPR), rheumatoid factor (RhF), autoantibodies and anti-neutrophil antibodies were all negative. Bone biochemistry blood tests (as calcium, phosphate, parathyroid hormone (PTH) levels) were all normal for a girl of her age.

Plain radiographic film appearances were consistent with diaphyseal dysplasia showing irregular cortical thickening of the diathesis of the long bones and more confluent sclerosis in phalanges of the hands and feet. The skull vault was uniformly thickened and there was sclerosis of the base of the skull. The spinous processes in the cervical region were sclerotic and the ribs and clavicles were slightly widened.

The initial computed tomography (CT) scan performed at the referring hospital was interpreted as demonstrating narrowing of both internal auditory canals (IACs), and this thought to be a possible cause of her deafness. In view of these findings and the possibility that surgical decompression of the cochlear nerves in the IACs might be indicated, she was referred to Addenbrooke's Hospital.

The high resolution temporal bone CT scans on bony settings performed at Addenbrookes did show some evidence of IAC stenosis but also revealed a thickened bony overgrowth with abnormal "ground glass appearance", extensively involving the temporal bones and skull base. These appearances were in keeping with fibro-osseous dysplasia (figures 1 and 2). There were some abnormalities within the semicircular canals which were otherwise near normal in size and configuration. At least 2 turns of the cochlea were identified on each side although both were partly obscured by the dense

bone of the petrous pyramid. Both middle ear clefts were small and the ossicles difficult to identify. The mastoid bones on each side were dense and poorly pneumatized.

High resolution imaging in this case revealed fibro-osseous dysplasia narrowing the internal auditory canals which would have explained the neural deafness. The imaging also revealed similar involvement of the cochlea producing a sensory component to the deafness. Finally involvement of the ossicles in the middle ear added the conductive component and hence the overall “mixed deafness” in this case. Surgery to the pathology at the most medial of these three sites would not improve hearing where there was a significant reduction in cochlear reserve due to involvement of the cochlea as well as the conductive component due to middle ear pathology.

DISCUSSION

The exact incidence of deafness in CED is uncertain because few authors have mentioned hearing loss in their reports. Of the 120 reported cases of CED, only 18% were stated to have a degree of deafness,¹¹ thus it is possible that this figure may be higher. More than 50% of these patients presented with pure sensorineural hearing loss, of which half were profoundly deaf. Twenty-two percent of cases are said to have a mixed type of deafness.¹²

The conductive deafness in CED can result from narrowing of the tympanic cavities, fixation or adhesions of the ossicles to the middle ear walls, and in addition, narrowing of the eustachian tube resulting in otitis media with effusion¹¹.

The aetiology of the sensorineural deafness however, is primarily thought to be due to narrowing of the internal auditory canal (IAC) caused by bony encroachment on nerves and vessels. There are several acquired conditions of the petrous temporal bone that may result in progressive IAC stenosis and these needs to be considered in the differential diagnosis of Camurati-Engelmann's disease (table 1). The most important pathologies include systemic disorders such as Paget's disease, cranial hyperostosis (van Buchem's disease), osteopetrosis (Albers Schonberg disease), familial hyperphosphatemia, fibrous dysplasia, otosclerosis and local conditions such as osteomas or exostoses of the IAC. Paget's disease leads to generalised bone involvement, increased serum alkaline phosphatase and a cotton wool appearance on imaging¹³. Fibrous dysplasia is characterised by areas of homogenous ground-glass appearance surrounded by dense cortical bone. Diffuse involvement of the entire temporal bone by the fibro-

osseous process with or without raised intracranial pressure, and involvement of other bones may be seen¹⁴. In osteopetrosis, there is excessive formation of immature bone that results in thickening of the bony cortex and narrowing or loss of the medullary cavity but the diaphyses, metaphyses and epiphyses are involved in contrast to Camurati-Engelmann's disease where only the diaphysis is involved¹⁵. Otosclerosis and osteomas of the temporal bone affect twice as many women as men and usually have their onset at puberty and the postmenopausal period, respectively. The former presents with a vascular otospongiotic overgrowth of osseous tissue in the otic capsule, (the fissula ante fenestrum of the oval window) and sclerosis of the labyrinthine capsule. The internal meatus may be narrowed by the otosclerotic focus. Although osteomas are most commonly encountered in the external auditory meatus, they may rarely occur in the internal meatus and compress the contents¹⁶. Bony lesions occurring in the IAC are not easily accessible to biopsy for histopathological examination.

It is important to distinguish whether the site of the lesion is within the middle ear, the otic capsule or the internal auditory canal, a combination of these or involvement at all these sites. When there is progressive bony compression of the auditory nerve in the internal auditory canal and this is the only site of pathology causing a neural defect, then decompressive surgery may be indicated. A prerequisite for this is a normally functioning auditory nerve prior to disease manifestation. Imaging is thus vital and high resolution axial and coronal CT scan on bony windows is the method of choice.

The site of involvement, the degree of compression and the risk/benefit assessment dictates management. The IAC may be decompressed via the middle fossa approach but the risks of this major surgery with regard to total loss of auditory function

and the risk to facial nerve must be explained fully to the patient as well as the small risk of post-operative epilepsy (1%)¹⁷ especially in view of the implications with regards to driving a motorised vehicle.

In this case imaging demonstrated that all these sites were involved, the middle ear, the otic capsule and the IAC and this accounted for the mixed (conductive and sensorineural) deafness. Middle fossa decompression of the cochlear nerve in the IAC would not have improved the hearing and was not indicated. The mixed deafness in this case with a 40dB average bone conduction and 85dB average air conduction was treated by the fitting of binaural hearing aids. However, she gained little benefit from her amplification as her deafness progressed. Due to the diffuse involvement of the temporal bone, middle ear surgery in the form of an ossiculoplasty was also not indicated.

In some cases of CED, there may be place for cochlear implantation in advanced cochlear pathology in the presence of a normal auditory nerve in an adequately patent IAC. If there is significant IAC stenosis with compression of the auditory nerve, cochlear implantation may not be indicated^{18,19}.

Consideration may also be given to the fitting of a bone anchored hearing aid (BAHA). This could offer some benefit in terms of amplification by passing the conductive element of the deafness direct binaural amplification of both bone conduction thresholds and offer optimal results in terms of amplification. However, a major concern might be the difficulty associated with inserting an implant into what is obviously abnormal bone. Also, subsequent osseointegration of the implant could not be guaranteed and the risks of implant failure may be higher than normal. There would be similar concerns over cochlear implantation particularly in view of the bone conduction

thresholds at present but this may be an option in the future. If her deafness progresses, a BAHA will be worth considering initially. If her thresholds continued to deteriorate due to abnormalities of the cochlea, then a cochlear implant may eventually become indicated.

The Camurati-Engelmann's syndrome is an extremely rare pathology and may afflict the temporal bones producing a decrease in auditory acuity which may be conductive, sensory or neural, a combination or all these. Imaging is critical in the evaluation of these patients since management is totally dependent on the sites of involvement. Surgery may not be indicated for diffuse disease in which case amplification with hearing aids, BAHA and cochlear implantation may have to be considered to rehabilitate these patients and improve their quality of life.

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SUMMARY

- CEA is a rare hereditary disorder affecting mainly the diaphysis of long bones but deafness has been reported in about 18% of these cases.
- The aetiology of deafness in CED disease is primarily thought to be due to narrowing of the IAC caused by bony encroachment on nerves and vessels. Interestingly in our case, there were gross abnormalities affecting both middle ears, and cochleas.
- The otologic manifestations of CED are reviewed and issues relating to the management of these patients with temporal bone involvement are discussed.

TABLES

Table 1: Causes of Internal Auditory Canal (IAC) Stenosis

Congenital malformation

1. Aplasia of IAC (VIII nerve agenesis)
2. Hypoplasia of IAC

Acquired

1. Extensive Otosclerosis
 2. Air cell in petrous apex
 3. Vitamin A deficiency
 4. Hyperostosis and bony tumours
 - a. Exostosis
 - b. Osteoma
 5. Generalised skeletal diseases
 - a. Cranial hyperostosis (van Buchem's disease)
 - b. Paget's disease
 - c. Camurati-Engelmann syndrome (diaphyseal dysplasia)
 - d. Turricephaly
 - e. Infantile cortical hyperostosis (Caffey's syndrome)
 - f. Osteopetrosis
 - Benign, dominant (Albers-Schonberg disease)
 - Malignant recessive
 - g. Hypophosphatemic rickets
 - h. Fibrous dysplasia (Albright's syndrome)
-
-

FIGURES AND LEGENDS

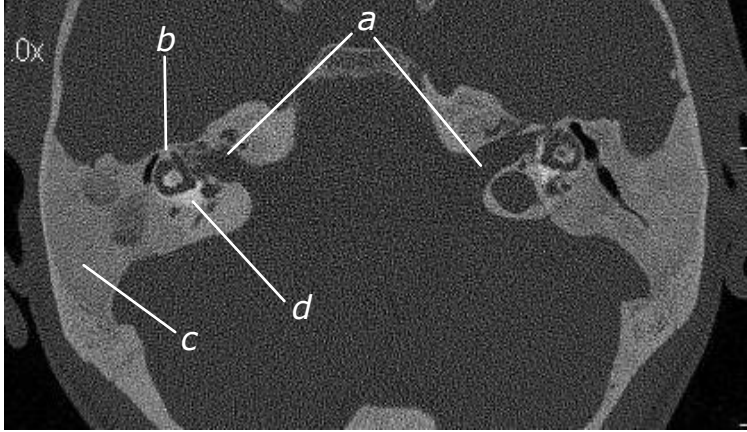


Figure 1

High resolution, axial temporal bone CT scan (bony settings). Wide internal auditory canals (a), and lateral semicircular canals (b). Dense temporal bones (c), with little pneumatization and hyperdense bone making up the otic capsule (d).

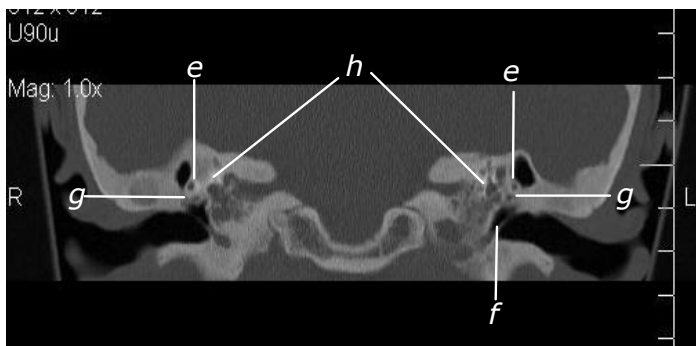


Figure 2

Coronal CT scan (bony settings). Fallopian canals (e) – both partly visible in the epitympanium. There is a very narrow middle ear space (f), no ossicles and the scutum is in continuity with the medial wall (g). The otic capsule bone appears very abnormal (h).