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Case study

A rare cause of facial nerve palsy in children: Hyperostosis corticalis generalisata (Van Buchem disease). Three new pediatric cases and a literature review

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ABSTRACT

Differential diagnosis of facial nerve palsy in children is extensive. We report on three pediatric cases presenting with facial nerve palsy caused by hyperostosis corticalis generalisata (Van Buchem disease). This autosomal recessive disease is characterized by progressive bone overgrowth, with narrowing of the neuroforamina in the skull causing cranial neuropathies. These three new cases of Van Buchem disease are of interest because of exceptionally early presentation of symptoms. Furthermore, this is the first report describing bilateral papilledema in a child with Van Buchem disease. Head computerized tomography (CT) scan revealed thickened calvarium, skull base and mandible in all three children, with narrowed facial nerve canals. Bone mineral density (BMD) was markedly increased at all measured points and biochemical markers of bone formation were significantly elevated. Diagnosis of Van Buchem disease was genetically confirmed. The cases are unique in that these are the first well-documented pediatric cases of Van Buchem disease.

1. Introduction

Early recognition, diagnosis and treatment of facial nerve palsy in the pediatric population is important. Recent data suggest that it is possible to identify a specific cause in over 70% of pediatric cases.[1] Differential diagnosis of facial nerve palsy in children is extensive. Common etiologies of acute onset facial nerve palsy in children are acute otitis media or

Abbreviations: HB, House–Brackmann; CT, computerized tomography; BMD, bone mineral density; PINP, procollagen type I N-terminal propeptide (PINP); Bone AP, bone-specific alkaline phosphatase; sCTx, carboxyterminal telopeptide alpha1 chain of type I collagen.

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mastoiditis, infection with *Borrelia burgdorferi* and varicella zoster virus (Ramsey Hunt syndrome). Congenital facial nerve palsy occurs in developmental defects or trauma. In gradual onset facial nerve palsy cholesteatoma or other space occupying lesions should be considered.

We present three children with recurrent facial nerve palsy caused by hyperostosis corticalis generalisata (Van Buchem disease), a rare genetic disorder of bone metabolism. All three children presented with symptoms of facial nerve palsy before the age of three years.

2. Case study

Three patients presented with recurrent facial nerve palsy. Clinical and laboratory findings of the three cases are summarized in Table 1.

Patient 1, a 3-year-old boy (Fig. 1A and B) presented with several episodes of unilateral facial weakness, the first of which was at the age of 9 months. The symptoms appeared within one day and facial asymmetry slowly recovered partially in the following two to three months. Most episodes were preceded by a common cold. In two years four similar unilateral episodes, both right-sided and left-sided occurred. Neither hearing abnormalities nor symptoms of increased intracranial pressure were reported. At neurological examination bilateral facial nerve palsy: House–Brackmann (HB) grade VI on the right side and grade V on the left side was apparent. Head circumference was near the 50th centile.

At pure tone audiometry he had mild bilateral conductive hearing loss (30–40 dB). Ophthalmologic examination showed bilateral papilledema and increased optical nerve sheath diameter. Head computerized tomography (CT) scan revealed thickened calvarium, skull base and mandible, with narrowed facial nerve canals. Ventricles were not enlarged. Bone mineral density (BMD) at the lumbar spine was vastly increased (+10 SD). The biochemical markers of bone formation osteocalcin, procollagen type I N-terminal propeptide (PINP) and bone-specific alkaline phosphatase (bone AP), were significantly increased in comparison to age and sex matched reference values. However, the biochemical marker of bone resorption carboxyterminal telopeptide alpha1 chain of type I collagen (sCTx) was in the normal range.

One of his sisters (patient 2, Fig. 1C–F) also had recurrent facial nerve palsy. Their parents were consanguineous with a positive family history for hyperostosis corticalis generalisata (Van Buchem disease). Diagnosis of Van Buchem disease was suspected and genetically confirmed in both patients by the finding of a 52-kB homozygous deletion 35 kB downstream of the SOST gene on chromosome 17q12-a21. This deletion was also found in a third pediatric case (see Table 1 for details) from another family in the same village.

Although evidence for effectiveness of this therapy is still scarce,[2] two of our patients were treated with prednisone (1–2 mg/kg/day) for two weeks as soon as a new episode of facial nerve palsy occurred. Levels of biochemical markers of bone formation were monitored during and after treatment. Effective monitoring of facial nerve function proved to be extremely difficult, despite the parents taking daily photographs of their children's faces. Patient 1 and 2 received

Table 1 – Summary of clinical and laboratory findings of three pediatric cases of Van Buchem disease.

	Sex, age	Severity facial nerve palsy,[a] age of onset	Hearing loss	Ophthalmological findings	Head CT	BDM[b]	Osteocalcin[c] (ng/ml)	PINP[c] (ng/ml)	Bone AP[c] (units/l)	sCTx[c] (pg/ml)
Case 1	Boy 3	VI/V 9 months	Conductive (bilateral 30–40 dB)	Bilateral papilledema	H + N	LS + 10 SD	93.3↑	2165.0↑	274.5↑	2473
Case 2	Girl 6	IV/III 2 years	Conductive (right 10–20 dB; left 50 dB)	Normal	H + N	LS + 5.2 SD PF + 7.5 SD TB + 9.5 SD	136↑	2295.0↑	262.4↑	3308
Case 3	Girl 4	V/V at birth	Mixed[d] (mild)	Normal	H + N	Not performed	130↑	1185.0↑	262.6↑	1437

↑Significantly increased in comparison to age and sex matched reference values (reference available on request).
CT = computerized tomography, BDM = bone mineral density, PINP = procollagen type I N-terminal propeptide, bone AP = bone-specific alkaline phosphatase, sCTx = telopeptide alpha1 chain of type I collagen, H = hyperostosis (calvarium, skull base, mandibula), N = narrowing of facial nerve canals, LS = lumbar spine, SD = standard deviation, PF = proximal femur, TB = total body.
a Severity of facial nerve palsy at time of presentation, House–Brackmann grading (right/left).
b BDM at lumbar spine, proximal femur and total body in standard deviations above the mean, adjusted for age and sex.
c Serum values at time of presentation.
d Mixed: conductive and sensorineural hearing loss.