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CASE REPORT

"A Probable Case Of Craniodiaphyseal Dysplasia- A Rare Entity"

KAMDAR P K*, SHAH V**

ABSTRACT

Craniodiaphyseal dysplasia (CDD) is a rare sclerosing bone disorder, the severity of which depends on its phenotypic expression. Hyperostosis can cause progressive foraminal stenosis leading to palsy of cranial nerves, epilepsy and mental retardation. We report here, the only case of CDD who presented with fever accidentally and was found to have right facial palsy and craniofacial bone thickening on X-ray, which was diagnosed as craniodiaphyseal dysplasia. We believe that this patient represented one of the few examples of a mild form of adult Craniodiaphyseal dysplasia with facial changes and mild conductive deafness in both ears.

Key words – Craniodiaphyseal dysplasia

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Introduction

Craniodiaphyseal dysplasia is an autosomal recessive (OMIM- 218300) or autosomal dominant (OMIM- 122860) condition. It is characterized by very severe bone dysplasia and massive generalized hyperostosis and sclerosis, especially involving the skull and facial bones. Progressive bony encroachment upon cranial foramina leads to severe neurological impairment in childhood [1].

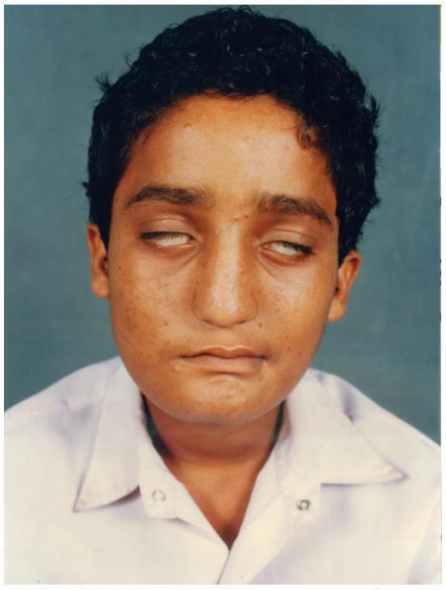
In 1958, Joseph et al2 described a child with severe sclerosis of the skull and facial deformity, noted the similarity of the features to a case reported previously by Halliday [3] and termed the condition as progressive craniodiaphyseal dysplasia. At that time, they considered it to be a variant of Camurati-Engelmann disease. Gorlin et a1[4] delineated the syndrome and cited published cases which had previously been described as cases of leontiasis ossea [3],[5] ,[6].

In 1974, MacPherson [7] described three cases of craniodiaphyseal dysplasia, pointed out the variable manifestations of the condition and emphasized the overlap in phenotype of CDD with some of the other craniotubular dysplasias and hyperostoses. He concluded that "perhaps the name craniodiaphyseal dysplasia should refer to a group of diseases". In our extensive literature search in Pubmed, OMIM, Google and others, we found nearly 30 cases with different phenotypic variations.

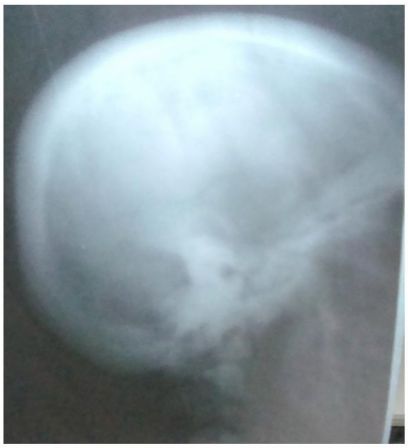
Case Report

A 14 year old male patient presented with a main complaint of fever for three days, the detailed history suggestive of visual disturbances and impaired hearing since the last 10 – 12 months prior to admission and gradually progressive. He was tall and moderately built and nourished. Facial appearance is not typical. However, his head was large, with a prominent forehead and a long face. There was neither flat nasal bridge nor hyperteloirism. There was bilateral ptosis and left side lower motor neuron facial palsy [Table/Fig 1].The overlying skin was apparently normal. The pulse rate, blood pressure and respiratory rates were normal. There was pallor. The cardiovascular and

respiratory systems were normal. The Central Nervous System (CNS) examination revealed left facial nerve palsy of the lower motor neuron type. The tone, power and reflexes were normal. The planter response was bilateral flexor. Both mother and father were asymptomatic and were not affected. There was no history of similar disease in any family members of the family including both brothers. His haemoglobin was 10gm%, total count was 4800/cu.mm and he was positive for Plasmodium vivax. Biochemical investigations including renal and liver function tests were normal. Blood sugar and electrolytes were within the normal range. His serum calcium was 9.8mg /dl and serum alkaline phosphate was 12 Units. His audiogram was suggestive of bilateral moderate conductive deafness. Though there was bilateral ptosis, his fundus examination was normal. The X-ray of his skull showed thickening of the entire skull and a prominent change at the base of the skull [Table/Fig 2] **.** The X-Ray of his tibia and fibula showed thickening of the cortex of diaphysis[Table/Fig 3] .All joints were normal. There was increased bone density along with thickening. There was undertubulation of the metacarpals, metatarsals and phalanges of the hand, especially those involving diaphysis, so that the bones appeared to be of the same diameter[Table/Fig 4] . The similar changes were seen in foot also. There was thickening of the cortex of the diaphysis of almost all the bones with epiphysis and metaphysis appearing relatively uninvolved. Because the main complaint was fever and he was positive for Plasmodium vivax, he was treated with antimalarial agents, he responded well, and was discharged after five days. We discussed the issue with the patient and his father. However; because of financial constraints and non availability of tests, they were not investigated with other imaging techniques and genetic testing.

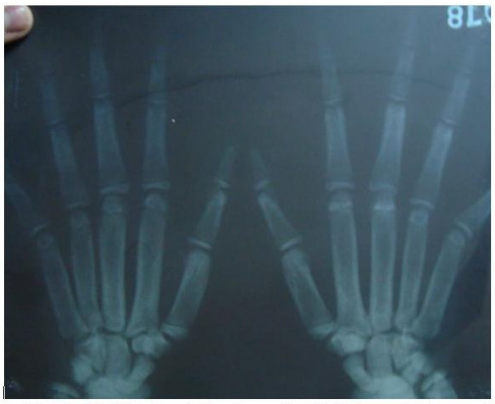


(Table/Fig 1) Bilateral ptosis and left side motor neuron facial palsy .



(Table/Fig 2) X-Ray skull showing thickening and prominent change at base of skull.

(Table/Fig 3) X-Ray Tibia and Fibula showing thickening of Cortex of Diaphysis.



(Table/Fig 4) X-Ray hand showing undertubulation of Metacarpals, Metatarsals and phalanges of hand.

Discussion

CDD is the most severe of all forms of sclerotic bone disorders. Although first reported by Halliday [3], Joseph et al [2] described the early features of the syndrome and named it progressive craniodiaphyseal dysplasia. Ghorlin, Spranger and Koszalka [4] described the syndrome in its entirety and identified previously published cases which had been described as cases of leontiasis ossea. The genetics of the disorder remain unclear, although it has been suggested that it may be autosomal recessive in inheritance, with an incidence of only 0.1/million [1],[3],[5],[9],[10]. Though Craniodiaphyseal dysplasia is an autosomal recessive condition, only a few cases have been described with autosomal dominant heritance [8],[9]. There are other disorders which overlap with Craniodiaphyseal dysplasia and are listed in OMIM lists.

In our patient, there was diffuse thickening of the base of the skull and Calvarium. X-Ray findings were suggestive of the thickening of the cortex of the diaphysis without involvement of the metaphysis or epiphysis predominantly. There was left sided facial palsy for which, the possible mechanism could be the mechanical damage of nerve fibres and/or impaired blood supply due to hyperostosis or due to narrowing of the middle ear cavity, internal auditory meatus and compression of the facial canal at the geniculate ganglion.

Radiologically, this syndrome has to be differentiated from other dysplasias such as Camurati-Engelmann disease, Van Buchem's dysplasia and craniotubular dysplasias [11],[12] .In Van Buchem's dysplasia, mandibular enlargement is the predominant manifestation in which the head circumference is normal and facial changes are manifested in the second decade.

In craniotubular dysplasia, there is metaphyseal widening and cortical thinning, giving rise to a club-shaped configuration of long bones, whereas in craniodiaphyseal dysplasia, the diaphyseal widening gives rise to a cylindrical appearance. However, the skull changes are nearly the same, except for prominence of the supra-orbital ridge. These cases can be easily excluded by radiological features in our case. The main difficulty in our case, was the problem of differentiating it from the Camurati–Engelmann disease. In the Camurati-Engelmann disease, the degree of cranio-facial involvement is mild, with major changes in the long bones and an early age of onset. Therefore, at first, a diagnosis of Camurati-Engelmann is more likely, however; the predominant involvement of the skull and its progressive nature favours Craniodiaphyseal dysplasia. Regarding age, there is one case report from Mumbai by S. Naique [13] in which the patient presented with quadruparesis at the age of 47 years. Though the age presentation may be late in our case, the progressive nature of the disease and prominent skull involvement made us to favour the diagnosis of Craniodiaphyseal dysplasia over Camurati-engelmann disease. One more feature of he Camurati-Engelmann disease is the characteristic body habitus, like thin limbs with little muscle mass, yet with prominent palpable bones [14], which were not observed in our case. Mutational analysis can clear the confusion, but it was not possible in our case and therefore, we concluded it as a probable case of Craniodiaphyseal dysplasia.

Limitations

We did not investigate this case with CT brain, MRI Spine and genetic studies like mutational analysis due to unavailability of such investigations in our institute at that time and due to financial constraints.

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