

Early Radiodiagnosis Of Craniometaphyseal Dysplasia – A Case Report

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Abstract:

Craniometaphyseal dysplasia is a rare genetic craniotubular bone remodeling disorder characterized by progressive hyperostosis, undertubulation of the long bones, causing metaphyseal deformities of the long bones, and sclerosis of cranio-facial bone. We are reporting a case of craniometaphyseal dysplasia in a 12-year-old indian child, highlighting the importance of radiological diagnosis of this rare genetic disorder.

Keywords: Craniometaphyseal dysplasia; Radiodiagnosis; Differential diagnosis

Introduction:

In 1954 Peter Jackson¹, an English physician and colleagues reviewed disorders of osseous modelling and identified a specific syndrome which comprised dysplasia of the metaphyses, sclerosis of the base of the skull, and overgrowth of the craniofacial bones. They coined the term "craniometaphyseal dysplasia" (CMD). It has an autosomal dominant form of inheritance which has been linked to chromosome 5, while recessive form results in more severe cases. Patients with CMD have a mutation that affects the ANK protein that is involved with mechanisms transporting minerals from the cells to the extracellular environment resulting in osteoblast/osteoclast dysfunction².

Deposition of mineralized bone and failure to degrade it prevents normal remodelling of the long bones, the skull base and the facial bones, with progressive expansion.

Radiographic changes are age-related and mostly visible during childhood. Radiographic findings may be sclerosis of the skull base, petrous temporal bone and along the skull sutures, Obliteration of paranasal sinuses, facial asymmetry and widening of the metaphyses of the long bones³.

Case report

A 12 year old boy born of a consanguineous marriage approached us with complain of progressive deafness, visual impairment and disfigurement of face. he was first child and was born by cesarean section with cephalic presentation without any associated abnormalities. The child had an enlarged skull with prominent frontal bossing, wide forehead, parietal protuberance, low set ears (Fig. 1).



Fig. 1: Enlarged skull with frontal bossing and typical facial features

The face showed a depressed bridge of the nose, wide separation of the orbits, and a characteristic broad flat nose, and hypoplastic mesodontic teeth (Fig. 1). Both eyes had showed coarse nystagmoid movements. There were central irregular opacities in both corneae. There were swellings over the wrist, elbow, and knee joints. The child was also having marked bilateral coxa valgum and genu valgum (Fig. 2).



Fig. 2: Bilateral coxa vulga and genu vulgum

A full blood count and studies of the blood chemistry were non-contributory.

The following radiological features were classical to CMD. Skull- Skull showed diffuse hyperostosis, with sclerosis of the mastoids and the paranasal sinuses which were not pneumatized (Fig. 3).



Fig. 3: Skull antero-posterior

Thorax- There was abnormal widening of the clavicles, at their medial ends. The ribs were also widened, particularly at the costochondral junctions. But, no sclerotic changes were present in the clavicles or ribs and the scapulae were uninvolved (Fig. 4).



Fig. 4: Antero-posterior view of thorax

Spine- No specific changes were present in the cervical, dorsal or lumbosacral regions of the spine.

Upper limbs- Modelling of the humeral metaphyses was abnormal, with widening of metaphyses at both the ends. The cortex was thin, with widening of the medulla, which had fine trabecular pattern. The diaphyses were sclerosed. Similar changes were present in the radii and ulnae. In hand abnormal modelling, with widening of the metacarpal and phalangeal metaphysis while sclerosis of diaphysis was present (Fig. 5).



Fig. 5: Skiagrams of upper limb

Lower limbs- Characteristics radiological changes seen in proximal and distal metaphyses of both femoral bone. Where splaying produced the classical 'Erlenmeyer flask' deformity. Similar changes occurred in the distal and proximal tibiofibular metaphysis (Fig. 6).



Fig. 6: Skiagrams of lower limb

MRI of brain showed compression of posterior cranial fossa structures, bilateral optic nerve atrophy as a result of optic canal stenosis, bilateral atrophy of auditory nerve because of stenosis of auditory canal (Fig. 7).

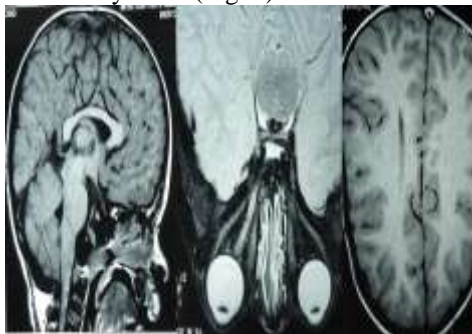


Fig. 7: MRI of the brain

Discussion

Though it is not always clinically possible to differentiate between the two (dominant and the recessive) forms of CMD, others craniotubular bone disorders can be differentiated from CMD. Among the others craniotubular disorders, followings may be considered as few important differential diagnoses:

- (a) Craniodiaphyseal dysplasia : This is usually associated with mental retardation, deafness and lack of sexual maturity. Typically it involves diaphyses instead of metaphyses. There is massive generalized hyperostosis with severe craniofacial deformity, and diaphyseal endostosis^{4,5}.
- (b) Metaphyseal dysplasia (Pyle's Disease) : There will be no skull base manifestations. Facial alterations may be supraorbital prominence and obtuse mandibular angle. Expanded pelvis and thoracic cage are associated features⁶.
- (c) Frontometaphyseal dysplasia (FMD) : Frontometaphyseal dysplasia described by Gorlin and Cohen (1969) as a syndrome consisting hypertrophy of bony supra-orbital ridge, hirsutism, long-bone alterations and conductive deafness. Besides manifestations in common with CMD, antimongoloid palpebral fissures, deficiency in mandibular growth, narrowing of the ribs and pelvis and sometimes mental retardation are usual presenting features of this syndrome. Patients with FMD develop alterations of

craniofacial bones, long bones, digits, vertebrae, iliac crest and pelvis⁷.

(d) Pyknodisostosis : Other than characteristic features of short stature, patients with pyknodisostosis also exhibit blue sclerae, dystrophic nails and rudimentary phalanges. Affected persons resemble each other, having very small faces and micrognathia. Constriction of cranial foramina is not observed in this condition⁸.

(e) Melorheostosis : Unilateral sclerosis and thickening of bones are presenting features. It also affects various soft tissues.

(f) Osteopetrosis : Among the three different forms of osteopetrosis, the predominant feature is increase in bone density. All of them have normal facies.

(g) Endosteal hyperostosis : There is widening of the mandible and diaphyses of tubular bones along with sclerosis of the skull.

(h) Sclerosteosis : This is characterized by overgrowth and sclerosis of the skeleton. Mandibular prognathism, maxillary hypoplasia, excessive height, syndactyly and elevated intracranial pressure are also manifestations of this disorder. In conclusion CMD can be easily diagnosed radiologically with its characteristic features and can be differentiated from other dysplasias. We reported our case to help in early and easy radiodiagnosis of this syndrome to help in early management.

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