Osteosclerotic bone dysplasia of the craniofacial bones

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ABSTRACT

Melorheostosis is a rare sclerosing bone dysplasia which is characterized by a localized, diffuse thickening of the cortical bone mainly affecting diaphysis of long bones, and bones of hand and feet unilaterally. Involvement of the maxillofacial region is rare and the diagnosis is mainly based on radiographic features. We report a rare case of a 5 year old girl with Osteosclerotic Bone Dysplasia (Melorheostosis) of multiple craniofacial bones.

Key Words: Melorheostosis, dysplasia, craniofacial, cortical bone, multiple

Introduction

In 1922, Leri and Joanny described a rare nonhereditary sclerosing bone dysplasia of unknown etiology, characterized by a localized, diffuse thickening of cortical bone, mainly the long bones of the upper and lower extremities, and those of the hands and feet. The condition, also known as Leri disease, may affect one bone (monostotic), many bones in one extremity (monomelic), or multiple bones (polyostotic). Involvement of bone and adjacent soft tissues leads to deformities which limit movement. The radiographic appearance of this disorder is described as similar to “dripping wax” or “flowing candle wax”. Hence it was named Melorheostosis, which is derived from the Greek words for limb (melos) and flow (rhein). Melorheostosis has been referred to as Leri disease or syndrome, osteosis eburnians monomelica, and rhizomonomelorheostosis. The incidence of this dysplasia has been...
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Reported to be 0.9 cases per million. [2,3,5,9,10] It does not show a gender or age predilection and in 40-50% of cases, it is evident usually by the age of 20 years. [2,5,10]

Melorheostosis has been associated with abnormalities of mesodermal origin like glomus tumors, vascular nevi, hemangiomas, arteriovenous malformations, lymphangiectasia, lymphangiomas, neurofibromatosis, hypophosphatemic rickets, linear scleroderma etc. [2,3] Histologically, the sclerotic bone consists of dense but regular bone tissue. Clinically, it may present with pain and/or joint or soft tissue swelling. [3]

Although approximately 400 cases of melorheostosis have been reported in the literature, its occurrence in the maxillofacial region is extremely rare. Less than 10 cases of melorheostosis in maxillofacial region have been reported in the English literature. [3,5] There have been no known cases of sensory or motor function impairment in these reported cases. [3] Majority of the cases reported are symptomless. The most common symptom reported is varying severity of pain and occasional paresthesia, but no loss of sensory or motor function. Other symptoms reported include gradual bone expansion, stiffness of joints, focal pigmentation on the skin, localized hypertrichosis, thickening of the skin, and scleroderma. [2,5]

This case was the first such reported case in our department and showed involvement of multiple maxillofacial bony structures.

Case Report
A 5 year-old female patient presented with a history of glabellar soft tissue fullness and nasal bone abnormality. The expansion was asymptomatic. No lymphadenopathy was noted. No significant family history of the disease was known. A medical CT had been done at the age of 1 month and eventually been diagnosed as Melorheostosis.

A non-contrast thin slice cone beam volumetric computed tomography scan of the head and neck was obtained extending from the level of frontal bone to C3-C4 to evaluate the extent of structural changes. Cone beam CT (CBCT) reconstructions demonstrated multiple high density/sclerotic foci of compact bone in the midline and extending into the left side of the sphenoid, frontal, and ethmoid bones, anterior maxilla, and anterior mandible along with enlargement of these bones. (Figure 1, Figure 4).

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Fig. 3 Sclerotic and expansile bone in maxillary and mandibular anterior region

Fig. 4 An extensive dense radiopaque mass in the left anterior mandible with enlargement and thickening of the cortical bone

Fig. 5 Sclerotic mass in the sphenoid, ethmoid and maxillary sinuses

Transitional dentition was noted with multiple primary teeth and unerupted permanent teeth. The bone in the region of teeth #s 21-25 and 9-10 was very sclerotic with evidence of expansion leading to a possible delay or failure of eruption of these teeth. (Fig.2) A malformed supernumerary tooth was noted in the anterior maxilla, apical and palatal to the developing #9. (Fig.3) The sphenoid sinus and the left maxillary sinus appeared to be inadequately pneumatized. (Fig.5) Mucosal thickening was noted in the bilateral maxillary sinuses, ethmoid air cells, and sphenoid sinus with apparent blockage of the right ostiomeatal unit. No ossification of the adjoining soft tissue or skin abnormality was reported.

Periodic evaluation was recommended to assess the progress of the condition. Surgical correction will be delayed until maximal growth has been achieved, unless patient becomes symptomatic.

Discussion
The etiology of melorheostosis is not clear. [1-6,11,12] It is known to develop slowly and without symptoms during childhood. [3] Various theories have been proposed which include vascular disturbances, inflammatory and degenerative processes, defects in embryogenesis, and abnormalities in innervation. [3,4,10] The incidence of associated vascular anomalies is about 5%. [5] Melorheostosis is a rare sclerosing bone dysplasia which is characterized by a localized, diffuse thickening of the cortical bone. This is a non-genetic developmental anomaly of mesodermal origin with an early childhood onset, predominantly affecting the appendicular skeleton. It is most common in the diaphysis of long bones, and bones of hand and feet unilaterally. [3,5,6,10,13] Exclusive craniofacial skeleton involvement by melorheostosis is very rare. Literature search was conducted using PubMed, Query, Web of Science, and CINAHL databases using the following terms: melorheostosis, jaw, mandible, maxilla, craniofacial, and dripping candle
wax. It was found that although over 400 cases have been reported, cases involving the cranio-facial region are very few. Anderson et al. compiled a list of 8 published reports in the English-language literature in November 2013 in Table I. These cases involved the cranio-facial region. [10] ("Current case" in the table, is the 9th case reported by Anderson et al.) Our case makes it the 10th such case. (Table 1)

Per above table [10], most cases were noted in females as a swelling in the maxilla or mandible with no associated pain or displacement of teeth.[10] Our patient is also a female and presents with nonsymptomatic involvement of multiple facial bones.

Freyschmidt in 2001 [12], described four distinct types and one combination type of melorheostosis based on radiological appearance. Type A is characterized by an osteoma-like appearance. Type B represents the classic “flowing” hyperostosis. Type C shows a myositis ossificans-like pattern. Type D demonstrates features similar to osteopathia striata. Type E is a mixture of patterns. According to this classification, involvement of the craniofacial regions would fall under Type E, the mixed pattern.[10,12,13] The case we report falls under Type E due to involvement of multiple bones in the craniofacial region. Melorheostosis is not associated with any specific biomarkers and laboratory findings show normal levels of serum calcium, phosphorous, and alkaline phosphatase.[5,10]

Various clinical features appear prior to any radiologic signs and symptoms. Radiologic signs are the primary basis of diagnosis of this condition. Radiographic features of melorheostosis involving the maxillofacial region show high attenuation, irregular foci of abnormal sclerotic bone involving the facial bones in a sequential manner along with overlying soft tissue prominence of the affected bones.[3,5,6,10,11] This condition in the jaws can closely resemble other bone tumors like osteoma and parosteal osteosarcoma.[10,14] Histopathology is nonspecific and offers no definitive diagnosis. Computed tomography is not regularly conducted for diagnosis, but can help in localizing the lesions. Magnetic resonance imaging (MRI) is not necessary in most such cases.[5,10] However, affected bones show a decreased signal intensity on MRI, and a moderate and asymmetric uptake is noted in a radionuclide bone scan.[13]
Primary goal of treatment is symptomatic relief and depends on the effect of the bony growth on the surrounding structures; including cosmetic deformity. Non-steroidal anti-inflammatory analgesics can be administered for bone pain. Surgical intervention is advised to be delayed until skeletal maturity is reached. Physical therapy in cases of appendicular involvement is also helpful in conjunction with analgesic relief. Parents of the patient were advised to continue regular follow up and report any changes, especially if any symptoms develop. Prognosis of this condition depends on the anatomical location, extension into soft tissues, and soft tissue changes. It does not shorten life span. The disease exhibits a slow, chronic course, with periods of exacerbation and arrest. Malignant transformation is rare. When alveolar processes of the mandible and maxilla are involved, eruption of teeth and orthodontic movement could pose clinical challenges. The differential diagnosis for increased bone density conditions which may simulate osteoslerotic bone dysplasia as presented in this young patient include sclerosing osteomyelitis, fluorosis, fibrous dysplasia, parosteal osteosarcoma and Caffey disease. Sclerosing osteomyelitis may affect any age, sex or race and it tends to occur more often in middle aged African-American females. Typically, it manifests as acute exacerbations of pain and swelling, and the cortical bone may show significant expansion. Radiographically, the lesion is poorly defined with early radiolucent zones in association with sclerotic masses. Osteomas present with a smooth well defined margin. Skeletal fluorosis causes an increase in the volume of the cancellous bone with irregular deposits of osteoid tissue. Fibrous dysplasia is usually monostotic and occurs in the mandible with the typical radiopaque-radiolucent ground glass or orange peel appearance. Parosteal osteosarcoma has to grow circumferentially to the bone’s surface, presents with a dense osteoid matrix and a cleavage plane separates the lesion from the cortex. Caffey disease or infantile cortical hyperostosis is a self-limiting disorder which can involve the mandible and presents as lamellated periosteal reaction with soft tissue swelling. In the current case, the patient was not symptomatic. She had a bony hard swelling, gradually increasing in size. There was no reported history of associated discomfort or obvious motor and sensory nerve deficits.

Conclusion
Melorheostosis is a rare bone disorder which is very uncommon in the craniofacial region. We report a case of craniofacial melorheostosis involving maxilla, mandible, sphenoid, ethmoid and frontal bones which was first diagnosed in our patient at the age of 1 month showing probability of congenital presentation. The follow up visit at the age of 5 years confirmed the diagnosis. Regular follow up and clinical monitoring for disturbances in the eruption of teeth was recommended. Challenges in orthodontic tooth movement maybe anticipated due to dense sclerotic bone in the alveolar processes.

References


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