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Craniometaphyseal Dysplasia

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Craniometaphyseal dysplasia (CMD) is an extremely rare genetic disease marked by progressive thickening of the craniofacial bones and aberrant development of the metaphyses in long bones. As a result of diffuse hyperostosis of the skull base, neurological symptoms

associated with cranial nerve compression, such as reduced vision, cranial nerve palsy, and deafness can occur. Craniofacial abnormalities are prominent and include hypertelorism, frontonasal bossing, a broad nasal root, prognathic mandible, and defective dentition [1]. In this paper, the authors report a case of CMD associated with facial dysmorphism and mild hearing loss in a 4-year-old girl. The clinical aspects, pathogenesis, and management of CMD will be reviewed.

A 4-year-old female was referred from the Department of Otorhinolaryngology to our department to examine her abnormal facial appearance and history of nasal obstruction and mild hearing loss. Her head circumference was in the 96th percentile, with notably wide-set eyes, a broad nasal root, a prominent forehead, and mouth breathing due to narrow nasal passages (Fig. 1). She was born normally at term with a weight of 3.1 kg. The pregnancy and neonatal period were uneventful. She had no relevant family history of skeletal or craniofacial abnormalities. The craniofacial bones were noted to have salient sclerosis and hyperostosis (Fig. 2). The distal femur was notable for a narrow diaphysis and widened metaphysis, resulting in an "Erlenmeyer flask"-shaped appearance (Fig. 3). A facial computed tomography scan exhibited reduced pneumatization of the bilateral mastoid air cells, diffuse cortical thickening of the craniofacial bones, obliteration of the paranasal sinuses, and narrowing of the cranial nerve foramina due to diffuse sclerosis of the cranial base (Fig. 4). Serum alkaline phosphatase was minimally elevated at 392 IU/L. Other characteristics were normal. The patient is cur-



Fig. 1. Frontal view of the patient: hypertelorism, paranasal bossing, and widening of the nasal bridge are visible.



Fig. 2. Adenoid view of the skull shows marked sclerosis and hyperostosis of the skull base, maxilla, and mandible.



Fig. 3. Plain radiograph of the femur with narrow diaphysis and widened metaphysis, resulting in an "Erlenmeyer flask"-shaped appearance.

rently being observed without medication or surgical intervention.

The term craniometaphyseal dysplasia was coined by Jackson et al. [2] in 1954, and refers to a genetic bone disease with overgrowth of the craniofacial bones and metaphyseal widening of the long bones. The majority of previously described CMD cases are inherited via an autosomal dominant (AD) pattern. Rarely, CMD is suspected to have autosomal recessive (AR) inheritance when unaffected parents have more than one child with the condition. Mutations in the progressive ankylosis protein homolog (ANK) human gene (ANKH) gene cause AD CMD, and a candidate locus for AR CMD on chromosome band 6q21-22 has been identified. The ANKH gene provides instructions for making a protein present in bone that transports a molecule called pyrophosphate out of cells. AD CMD is characterized by aberrant alteration of the metaphyses, mild-to-moderate overgrowth of the cranial bone, and various cranial nerve compression presentations. Individuals with typical uncomplicated AD CMD have a normal life expectancy. AR CMD is typically more severe than the AD form [3]. In general, it has been reported that the craniofacial abnormalities of AD CMD can be alleviated with growth, which suggests a possible correlation with hormone levels. The AR form has ongoing clinical manifestations with a very poor prognosis.

Bilateral choanal narrowing secondary to bony

sclerosis and paranasal bossing is a common presentation in infancy. These characteristics are inclined to improve with growth. The craniofacial abnormalities of CMD are conspicuous and involve hypertelorism, frontal and paranasal bossing, a broad and flat nasal bridge with a saddle deformity, and prominent mandible. Two-thirds of cases complain about nasal and ear symptoms. It is advisable to assess hearing and visual function to appreciate the influence of cranial nerve compression, and to monitor the general progression of the disease. Metaphyses of long bones are widened at the distal end of the femur with a thinned cortex and decreased bony density.

Medical and surgical treatments can be considered for the management of CMD. Two main regimens can be used to treat CMD. The first regimen is calcitonin. Calcitonin inhibits bone resorption primarily through osteoclast suppression and secondarily by impeding bone formation through feedback coupling to limit osteoblast activity [4]. The other potential treatment is a low calcium diet and calcitriol. Calcitriol stimulates the resorption of bone by promoting recruitment and differentiation of monocytic precursors to form multinucleated osteoclasts on actively remodeling bone surfaces [5]. A low calcium diet is likely to prevent further deposition of minerals in bone and allow the calcitriol-induced osteoclasts to reduce the skull bone mass. Surgical treatment is applicable in cases of severe bony overgrowth of facial bones, as well as compression of a nerve canal or narrowed foramen magnum. However, surgical procedures can be technically difficult and bone regrowth is common. Millard et al. reported the first two cases of craniofacial surgery in CMD. The first surgery was performed on a 17-year-old boy and an aesthetic result was achieved with marked improvement in his quality of life. They attempted the second surgery with a combined craniofacial operation and intracranial bony decompression on the first patient's younger sister but reported mortality within 24 hours due to postoperative narrowing of the foramen magnum. Richards et al. do not recommend surgery, as the advantages are likely to be short in duration and the technical difficulties, and risk factors, such as sclerotic mastoid and obscured bony landmarks due to bony overgrowth are tremendous [5]. Therefore, surgery can be considered for palliative purposes to relieve severe symptoms by cranial nerve compression.

In the present case, developmental abnormalities of the craniofacial skeleton and long bone were not severe. Other clinical manifestations may be alleviated

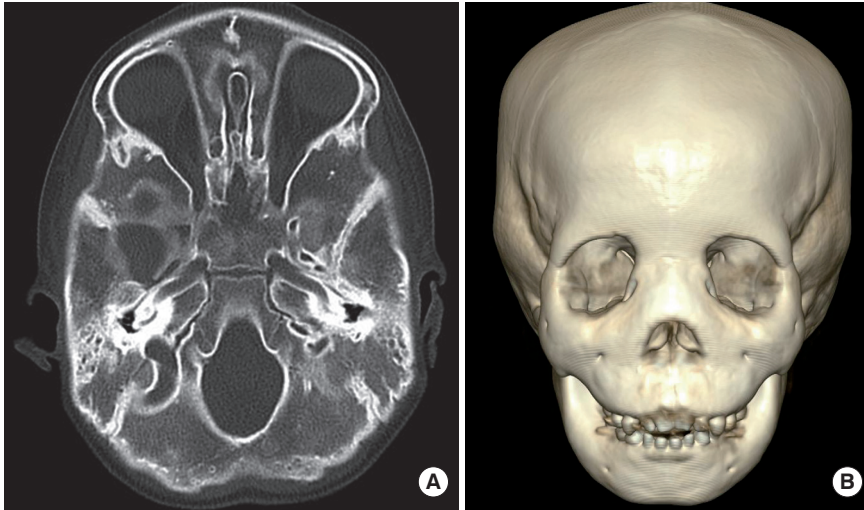


Fig. 4. (A) Axial view of a facial computed tomography (CT) scan shows diffuse thickening of the cranial base, reduced pneumatization of both mastoid air cells, and narrowing of the cranial nerve foramina by hyperostosis. (B) Three-dimensional facial CT scan shows unusually prominent zygoma, maxilla, and mandible. Increased interorbital distance and a widened nasal bridge can also be seen.

with growth. CMD is a rare disease that is often misdiagnosed. An accurate and early diagnosis and knowledge of the natural history of CMD is important for establishing preventative treatment regimens and proper management of complications as well as estimating the prognosis.

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Neuroendocrine Differentiation of Primary Mucinous Carcinoma of the Cheek Skin

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Primary mucinous carcinoma of the skin (MCS) is a rare sweat gland tumor, with approximately 175 cases reported in the literature since the first case was described by Lennox et al. [1] in 1952. Although MCS is usually slow growing, it often shows locally aggressive behavior and a high rate of local recurrence following simple excision. The clinical appearance and differential diagnosis of MCS vary, but histopathologically MCS is similar to metastatic carcinomas, specifically of the breast and colon. The recognition of MCS is essential for preventing an erroneous diagnosis of