# Oligodontia Associated With Osteopetrosis: A Rare Case Report

*C. Vinay, MDS R. Santosh Kumar, MDS*
*K.S. Uloopi, MDS V. Madhuri, BDS*
*R. Chandrasekhar Rao, MDS*

## ABSTRACT

Osteopetrosis is a descriptive term that refers to a group of rare hereditary disorders of the skeleton characterized by increased density of the bones. It was first described in 1904 by Albers –Schonberg, a German radiologist. This disease varies greatly in its presentation and severity ranging from neonatal onset with life threatening conditions such as bone marrow failure to incidental finding of osteopetrosis on radiographs. Dental abnormality may be attributed to pathological changes in the bone. Dentists should be aware of patients with the disease because of its effect on osteoclast function which results in impaired wound healing. This paper presents the case of a five-year-old girl exhibiting oligodontia associated with osteopetrosis.
**(J Dent Child 2011;78:53-6)**

**Received August 7, 2009; Last Revision November 20, 2009; Revision Accepted November 22, 2009.**

**Keywords: dental development, oral medicine, oral pathology, oligodontia, osteopetrosis, albers-schonberg disease**

Osteopetrosis is a descriptive term that refers to a group of rare hereditary disorders of the skeleton characterized by increased density of the bones. It was first described in 1904 by Albers – Schonberg, a German radiologist. This disease varies greatly in its presentation and severity ranging from neonatal onset with life threatening conditions such as bone marrow failure to incidental finding of osteopetrosis on radiographs. Dental abnormality may be attributed to pathological changes in the bone. Dentists should be aware of patients with the disease because of its effect on osteoclast function which results in impaired wound healing. This paper presents the case of a five year old girl exhibiting oligodontia associated with osteopetrosis. Osteopetrosis, also known as Albers-Schonberg disease, is a hereditary disease characterized by sclerotic, brittle, radiopaque bones with associated hematological and neurological abnormalities.[1] It was first described in 1904 by Albers-Schonberg, a German radiologist.[2] The term osteopetrosis was later introduced by Worth in 1963. Due to the dense, rock-like appearance of the bone, it is commonly referred to as "marble bone disease."[3] The estimated prevalence ratio is 1:100,000 to 1:500,000.[2] Mutations in at least 10 genes have been identified as causative in humans.[4] Nearly 50% of the cases are due to mutations in the ATP6i gene.[5]

Osteopetrosis results from either dysfunction or lack of osteoclasts, resulting in marked thickening of the cancellous and cortical bone along with encroachment or even disappearance of the marrow space.[6] The failure of osteoclasts to resorb bone leads to impaired bone remodeling. There is a decrease in the blood-forming potential of the marrow for which the body attempts to compensate by having other extra osseous structures with hematopoietic potential, such as the spleen and liver, take over this function. This might eventually lead to anemia, thrombocytopenia, and hepatosplenomegaly. Cranial nerve dysfunction, due to the proliferation of

*Dr. Vinay is professor, Department of Pediatric Dentistry, Dr. Santosh Kumar is assistant professor in Department of Prosthodontics, Dr. Uloopi is professor in Department of Pediatric Dentistry, Dr. Madhuri is postgraduate student in Department of Pediatric Dentistry and Dr. Rao is professor in Department of Pediatric Dentistry, all at the Vishnu Dental College, Bhimavaram, India.*
*Correspond with Dr. Vinay at vinaychandrappa@yahoo.co.in*

the bone surrounding the nerve foramina, has been described as a severe form of the disease.[1]

The dental findings may include: delayed tooth eruption; absence of a few teeth; unerupted and malformed teeth; enamel hypoplasia; abnormal pulp chambers; tendency to early decay; defects of the periodontal membrane; thickened lamina dura; and early tooth loss due to caries.[1]

The purpose of this paper was to present the case of a 5-year-old girl who exhibited oligodontia associated with osteopetrosis including its dental management.

## CASE REPORT

A 5-year-old Asian girl presented to the Department of Pediatric Dentistry, Vishnu Dental College and Hospital, Bhimavaram, Andhra Pradesh, India, with the complaint of unerupted teeth. This child was born to parents with a history of second-degree consanguinity (her parents and maternal and paternal grandparents had consanguineous marriages). No abnormalities were reported, however, in their extended family. Health history revealed that the pregnancy and birth were uneventful. The dental history given by the parents suggested the congenital absence of most of the primary teeth, although family history regarding absence of teeth was not contributory. The girl had an elder sibling who died a few days after birth due to unknown causes; a medical record regarding the late sibling's illness was unavailable.

On general examination, the child appeared to be malnourished with a short and thin stature. She was 2 feet 10 inches tall and weighed only 24 pound. The child presented skeletal deformities, including genu valgum, and a prominent pigeon chest (Figure 1). Her head morphology and facial appearance appeared normal. No signs of mental retardation or neurological problems were detected. Extraoral examination revealed hypoplasia of the premaxilla and a retruded mid-face (Figure 1).

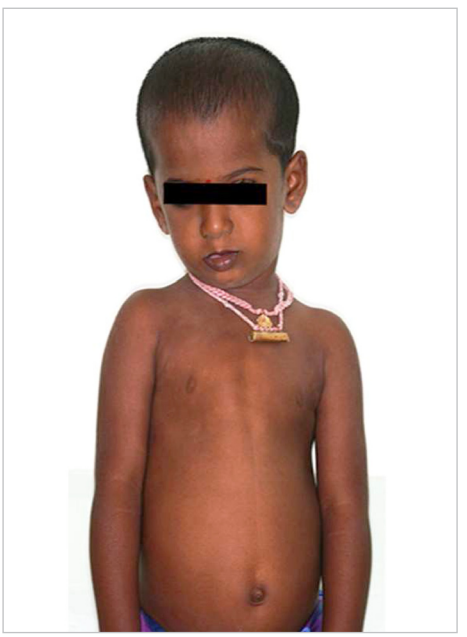

*Figure 1. Extra oral photograph showing pigeon chest, hypoplasia of the premaxilla and retruded mid face.*

Intraoral examination during the first visit revealed the presence of the teeth E, F, O, P, and S. The alveoli were of normal size and the primary maxillary central incisors were grossly decayed, yet the remaining erupted primary teeth appeared normal in size, shape, and color (Figures 2a-2c). The orthopantomograph (Figure 3) revealed hypoplastic unerupted permanent tooth buds along with diffuse sclerosis of the nasomaxillary complex and mandibular bone.

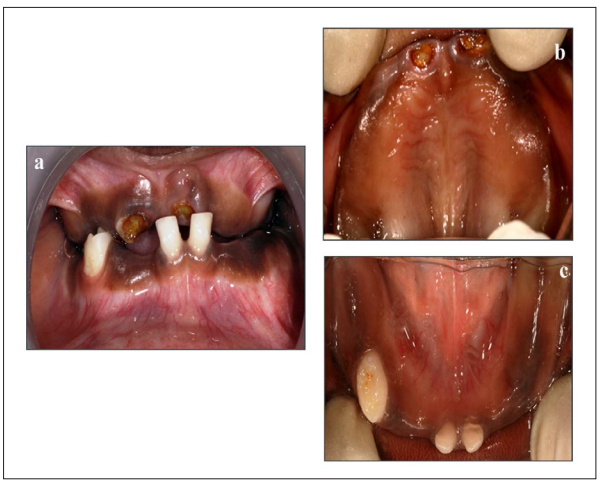


*Figure 2a–c. Intraoral photographs revealing normal alveolus with the presence of only five deciduous teeth ie, E, F, O, P and S including grossly decayed maxillary central incisors.*

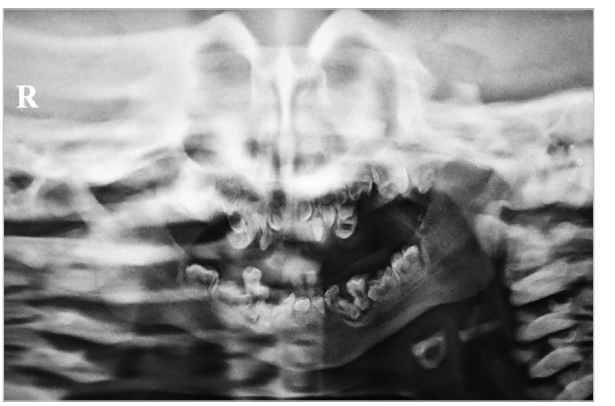


*Figure 3. Orthopantomograph (OPG) showing the congenital absence of all fifteen deciduous teeth and the presence of all permanent tooth buds with certain degree of hypoplasia.*

The child was referred to pediatric dentists for further investigations and treatment. Ultrasonography revealed mild splenomegaly, but no cardiac abnormalities were detected based on echocardiogram. Several radiographic images were obtained. A lateral view of the spine (Figure 4 ) indicated Rugger-jersey spine. A radiograph of the pelvis (Figure 5 ) illustrated diffuse sclerosis with Erlenmeyer flask deformity of the femoral shaft.

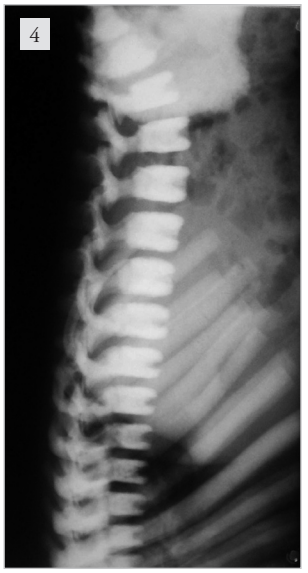

Figure 4. Lateral radiographic view of spine presenting Rugger jersey sign with increased bone density.

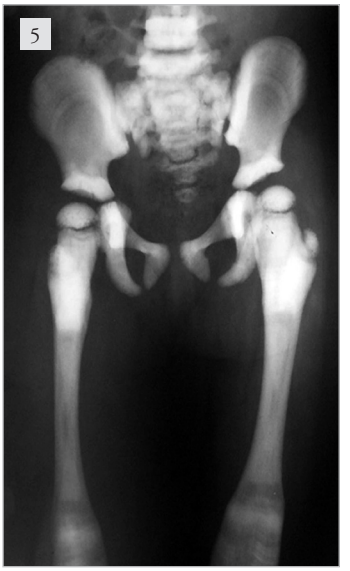

Figure 5. X-ray pelvis illustrating Erlenmeyer flask deformity of the femoral shift and diffuse sclerosis of bones.

Blood investigations showed normal differential count, although the patient's hemoglobin level was estimated to be 6.8 mg. The serum calcium and serum phosphorous levels were found to be normal, while the alkaline phosphate level was 258 U/L. Based on the overall investigation, the medical report suggested a diagnosis of osteopetrosis.

Complete oral rehabilitation was planned with the aim of restoring the form, function, and esthetics influencing the child's overall physical and psychological development. Extraction of the grossly decayed maxillary central incisors was planned followed by placement of a removable prosthesis to replace congenitally missing teeth. Because the vascular supply to the jaws and wound healing are compromised in osteopetrosis, avascular necrosis and infection after dental extractions may lead to osteomyelitis. Since the teeth were asymptomatic and the parents were unwilling for extraction, the grossly decayed primary maxillary central incisors were spared from extraction.

After recording jaw relations, a maxillary overdenture and a mandibular partial denture, with openings to accommodate teeth O, P, and S, were fabricated and inserted (Figures 6 a-d). A permanent resilient liner was used to increase comfort for the underlying soft tissues. The child currently is following strict oral hygiene regime. Regular 3-month recall protocol has been maintained to monitor the growth of the dental arches and eruption status of the permanent teeth. At 6-month follow up visit, the parents have reported better masticatory and speech efficiency of the child and also improved interaction with her peer group due to better esthetics.

## DISCUSSION

Osteopetrosis encompasses a clinically and genetically heterogeneous group of conditions that exhibit the trait of increased bone density on radiographs.[4] Although this disease is rare, it has been reported in most ethnic groups, especially where consanguinity is common,[5] as reported in the present case. Abnormal remodeling of primary woven bone to lamellar bone results in brittle bone that is prone to fracture.[5] The decreased blood-forming potential of the marrow is compensated by the liver and spleen, thus resulting in splenomegaly and hepatotomegaly.[7] This pathology contributed to the mild splenomegaly reported in the present case.

The diagnosis of osteopetrosis is clinical and largely depends on the skeleton's radiographic appearance. The classical radiographic features of osteopetrosis include: diffuse sclerosis affecting the skull, spine, pelvis and appendicular bones; bone modeling defects at the metaphyses of long bones (eg, funnel-shaped appearance); bone-in-bone appearance, especially in the vertebrae and phalanges; focal sclerosis of the skull base, pelvis, and vertebral end plates; and Rugger-jersey spine.[4] The child studied exhibited diffuse sclerosis of bones with widened metaphyseal regions of the long bones, Erlenmeyer flask deformity of the femoral shift, and Rugger-jersey spine, which collectively strongly support the diagnosis of osteopetrosis.

Osteopetrosis is divided into 3 types: (1) severe infantile—malignant autosomal recessive; (2) intermediate mild autosomal recessive; and (3) benign autosomal dominant.[8] The intermediate type of osteopetrosis, which is characterized by deficiency of carbonic anhydrase II, is a milder form of the disease, diagnosed late in infancy

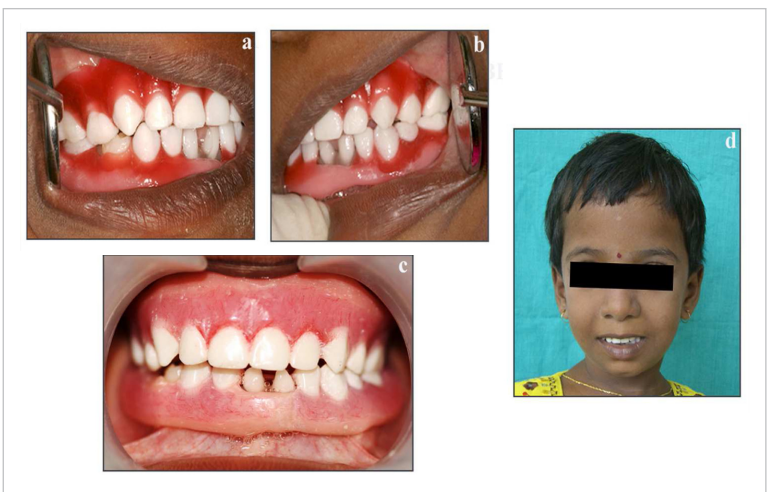


Figure 6a-d. Post operative Photographs screening maxillary over denture and mandibular partial denture in occlusion.

or early childhood.[9] This type usually displays classical radiographic features with or without hematologic complications.[7] The child in the present case report demonstrated analogous findings.

Dental findings concerning osteopetrosis include: noneruption or delayed eruption; hypodontia; enamel hypoplasia; abnormal pulp chambers; tendency to early decay; defects of the periodontal membrane; thickened lamina dura; and early tooth loss due to decay.[1,8,10,11] Although no enamel hypoplasia was demonstrated in this patient's erupted teeth, radiographically, her unerupted permanent teeth showed a certain degree of hypoplasia. A rare aberration in the number of teeth was observed (ie, 15 primary teeth were absent). The presence of permanent tooth buds, despite the congenital absence of primary precursors, is, however, quite atypical. According to Friede et al., teeth aplasia is due to progressive inadequacy in the supply of nutrients to the developing tooth germ caused by increased bone density.[10,12]

The medical management of osteopetrosis is aimed at decreasing or arresting hyperostosis, correcting anemia and thrombocytopenia, and treating infections. Oral cellulose phosphate, prednisolone, a low calcium diet, a high dose of calcitriol, and recombinant human interferon gamma has been reported to be effective in some patients.[13] Bone marrow transplantation has shown evidence of engraftment with correction of anemia, thrombocytopenia, and leukoerythroblastosis within 12 weeks of transplant.[10]

The main objective of dental management includes periodic prophylactic treatment to maintain a fragile oral health status and rehabilitation in the instance of developmental anomalies such as missing teeth. Preventive measures must be continuously and vigorously practiced. Special care should be taken to avoid serious adverse effects during invasive surgical procedures in this high-risk group of patients.

## REFERENCES


1. Droz-Desprez D, Azou C, Bordigoni P, Bonnaure-Mallet M. Infantile osteopetrosis: A case report on dental findings. J Oral Pathol Med 1992;21:422-5.
2. Lam DK, Sándor GK, Holmes HI, Carmichael RP, Clokie CM. Marble bone disease: A review of osteopetrosis and its oral health implications for dentists. J Can Dent Assoc 2007;73:839-43.
3. Filho AM, de Castro Domingos A, de Freitas DQ, Whaites EJ. Osteopetrosis: A review and report of two cases. Oral Dis 2005;11:46-9.
4. Stark Z, Savaririyan R. Osteopetrosis. Orphanet J Rare Dis 2009;4:5.
5. Wilson CJ. Autosomal recessive osteopetrosis. Orphanet Encyclopedia. "www.orpha.net/data/patho/GB/uk.malosteo.pdf"; 2003.
6. Barbaglio A, Cortelazzi R, Martignoni G, Nocini PF. Osteopetrosis complicated by osteomyelitis of the mandible: A case report including gross and microscopic findings. J Oral Maxillofac Surg 1998; 56:393-8.
7. Ruprecht A, Wagner H, Engel H. Osteopetrosis: Report of a case and discussion of the differential diagnosis. Oral Surg Oral Med Oral Pathol 1988; 66:674-9.
8. Bakeman RJ, Abdelsayed RA, Sutley SH, Newhouse RF. Osteopetrosis: A review of the literature and report of a case complicated by osteomyelitis of the mandible. J Oral Maxillofac Surg 1998;10:1209-13.
9. Whyte MP. Carbonic anhydrase II deficiency. Clin Orthop Relat Res 1993;294:52.
10. Jälevik B, Fasth A, Dahllöf G. Dental development after successful treatment of infantile osteopetrosis with bone marrow transplantation. Bone Marrow Transplant 2002;6:537-40.
11. Waldron CA, Neville BW, Damn DD, Allen CM. *Oral and Maxillofacial Pathology.* Philadelphia, Pa: Saunders; 1995:444-5.
12. Friede H, Manaligod JR, Rosenthal IM. Craniofacial abnormalities in osteopetrosis with precocious manifestations: Report of a case with serial cephalometric roentgenograms. J Craniofac Genet Dev Biol 1985;3:247-57.
13. Toranzo-Fernandez JM, Noyola-Frias MA, Hernandez-Duarte SP. Infantile osteopetrosis: A case report with osteomyelitis of the maxilla. J Clin Pediatr Dent 2002;27:77-80.