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Case Report

Juvenile Paget's Disease with Paranasal Sinus Aplasia

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Juvenile Paget's disease (JPD) is a rare skeletal disorder that's characterized by bone demineralization and elevated levels of serum alkaline phosphatase. JPD involves the paranasal sinuses in extremely rare cases. We report here on a 25-month-old Asian male who was diagnosed of JPD associated with aplasia of the paranasal sinuses, but not the ethmoid sinuses. The patient was successfully treated by surgery and we undertook no medical intervention. This appears to be the first reported case of JPD associated with bilateral paranasal sinus aplasia.

Key Words. *Paget disease of bone, Osteitis deformans, Paranasal sinuses, Paranasal sinus neoplasms*

INTRODUCTION

Paget's disease of the bone is a chronic disease of the skeleton. This disease is relatively common in older people, and it occurs in approximately 3-4% of the population aged over 50 yr. In contrast, Juvenile Paget's disease (JPD) is rare, with fewer than 30 reported cases according to our literature search. JPD is a metabolic bone disease that presents during the first 2 yr of life as generalized skeletal deformities associated with elevated serum alkaline phosphatase activity of a bony origin (1).

The term "JPD" was first used to describe an unusual bone disease in an 11-yr-old boy (1). Other terms for this disease include fragile bones and macrocranium, hyperostosis corticalis deformans juvenile, chronic idiopathic hyperphosphatasia, chronic progressive osteopathy with hyperphosphatasia, osteochalasia desmalis familiaris, familial osteoectasia, hereditary hyperphosphatasia, and congenital hyperphosphatasia (2). While the disease process is usually polyostotic and it can affect any bone in the body, sinus aplasia has not previously been associated with JPD.

The present article describes a case of 25-month-old Korean male with JPD associated with paranasal sinus aplasia.

CASE REPORT

A 25-month-old Korean male was hospitalized for the evaluation and treatment of nasal stuffness. The patient was tall for his age (95.2 cm, 97th percentile, he weighed 17.5 kg (97th percentile) and had a disproportionately large head (52 cm head circumference). The patient had marked facial deformities including a large forehead, prominence of both cheek bones and leontiasis features cause by maxillary enlargement.

The nasal endoscopic findings showed obstruction of the nasal cavity due to hypertrophy of both inferior turbinates (Fig. 1A). Both tonsils were enlarged (grade II) and adenoid hypertrophy (grade III) was observed on the skull lateral view. The laboratory tests showed a highly elevated serum alkaline phosphatase concentration of 1,082 IU/L (normal range: 50-150 IU/L), and a total hydroxyproline urinary excretion rate of 330 mmoL/24 hr, which was slightly above normal. All the other laboratory tests showed normal results.

Plain radiographs of the skull showed thickening and sclerotic changes in the facial bone and skull vault, and unformed obliterate paranasal sinuses (Fig. 1B). The chest appeared normal. The lower extremities revealed slight anterior bowing of the femora, reduced mineralization, loss of normal trabeculation, and coarse strand-like bone trabeculation.

CT scans of the skull showed thickening of the facial bone and skull vault, and non-development of the paranasal sinuses, while the ethmoid sinuses appeared normal. There were thickening of the nasal septum and both inferior conchae, resulting in narrowing of the nasal cavities on CT scans (Fig. 1C).

Bone scans revealed increased radiotracer uptake in the facial bones and skull base, and this suggested a high rate of bone turnover (Fig. 1D).

To relieve the nasal symptoms, the patient underwent two surgical procedures: a submucosal partial inferior turbinectomy and an adenotonsillectomy. Histological examination of the inferior nasal concha showed an extremely thin cortex and an abnormal spongiosa. There wers increased cellularity with irregularly arra-

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nged cement lines, loose fibrovascular tissue within the intertrabecular space. The trabeculae were lined with active osteoblasts and osteoclasts, numerous osteoclasts, and an increase in osteoblastic bone formation. These findings were consistent with Paget's disease (Fig. 2).

The patient is now 4-yr-old. The nasal stuffiness has improved, there have been no additional symptoms, and no further or ongoing treatment has been required. The patient no longer shows problems relating to the sinus aplasia.

DISCUSSION

Swoboda coined the term JPD in 1958 to emphasize the similarities between congenital hyperphosphatasia and Paget's bone disease (3). Both disorders are characterized by bowing of the extremities, increased serum alkaline phosphatase activity of a boney origin, elevated urinary hydroxyproline levels and abnormal cortical remodeling of highly vascularized bone (3).

JPD is a bone modeling disorder, and the basic pathology is believed to be due to a blockage in the transformation of coarsely woven bone into mature lamellar bone (4). The pathogenesis of JPD is unknown. The increased serum alkaline phosphatase

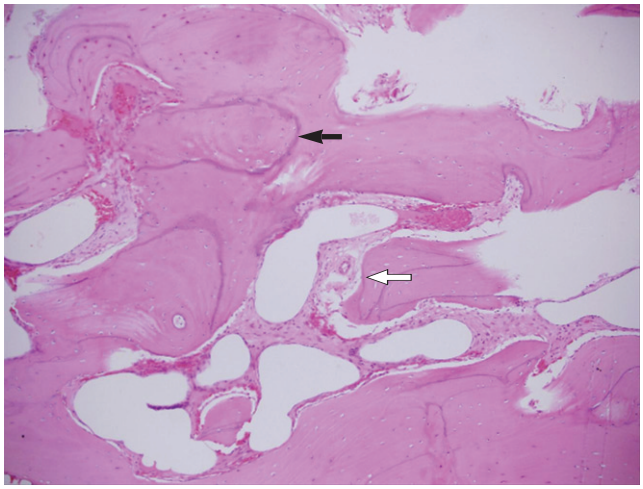

Fig. 2. Biopsy from the inferior nasal concha showing increased cellularity with irregular cement lines (black arrow) and loose fibrovascular tissue within the intertrabecular space (white arrow) (× 200, H&E stain).

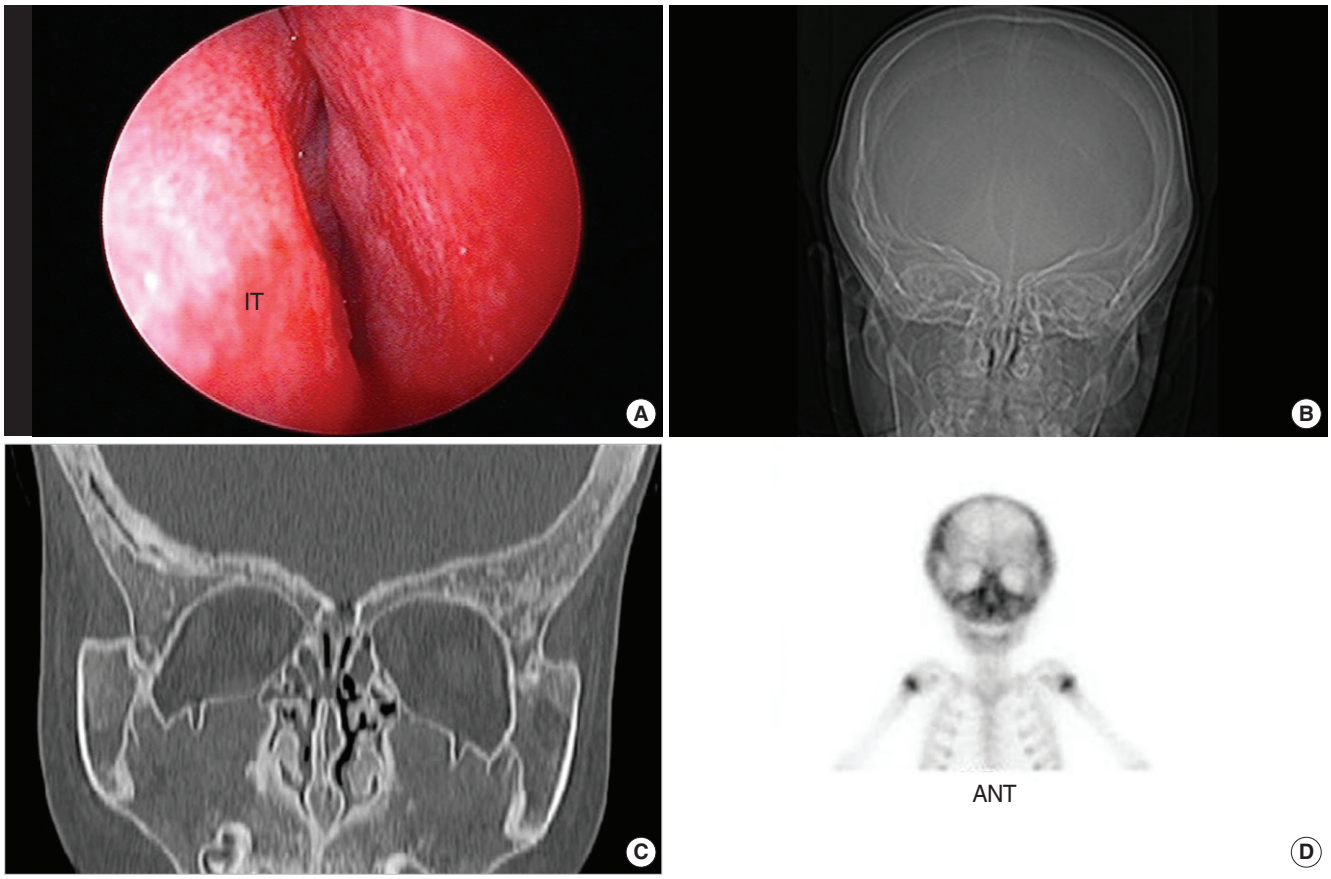

Fig. 1. (A) Endoscopic view showing hypertrophic change of inferior turbinate & nasal cavity narrowing of right side. (B) Plain radiography showing thickening and sclerotic changes in the facial bone and skull vault, and unformed obliterate paranasal sinuses. (C) Coronal computed tomogram revealing complete agenesis of both paranasal sinuses but not the ethmoid sinuses. (D) Bone scan showing marked uptake of radionuclide in both paranasal sinuses, especially the maxillary sinuses. IT: inferior turbinate.

activity is a reflection of increased bone turnover, and excessive bone turnover is responsible for the decreased levels of mature lamellar bone (4).

The clinical findings of patients with JPD are increased bone fragility, bowing deformities, a large head, premature loss of teeth, kyphosis, scoliosis, and progressive loss of muscular power with delayed and clumsy walking or failure to walk (5, 6). In the present case, the paranasal sinuses were obliterated, resulting in obstruction of the upper air passages and leontiasistic deformity of the facial bones.

Radiographic analysis of patients with JPD shows skeletal bone changes, including widened skull bones, generalized demineralization, expanded osteoporotic long bones with coarse trabeculation and short tubular bones (5, 6). In addition, the calvarium is irregularly thickened. Involvement of the paranasal sinuses in JPD is extremely rare (7). The present patient had complete aplasia of the maxillary, frontal and sphenoid sinuses, and such paranasal sinus aplasia has not previously been associated with JPD.

The laboratory findings for the present patient were consistent with those of the previous reports, including elevated serum alkaline phosphatase, acid phosphatase activity and urinary peptide-bound hydroxyproline (2). The elevated peptide-bound hydroxyproline excretion was consistent with the elevated levels of alkaline and acid phosphatase in the blood, and this was evidence of rapid bone matrix turnover (6).

The histological findings of JPD include intensive metaplastic fibrous bone formation and increased osteoblast and osteoclast activity (5). In the current case, the bone biopsy findings were similar to the previously reported findings, and they confirmed the patient's increased bone turnover.

The administration of calcitonin for JPD can slow the disease progression by promoting normal bone formation, resulting in an enhanced quality of life (2, 4, 5, 8). Furthermore, calcitonin is reported to not only arrest the disease progress, but it can also reverse the long bone abnormalities in terms of external dimensions and shape (8). Treatment with diphosphonate (pamidronate) may be equally effective (9-11). While JPD generally requires medical treatment, such a strategy was not applied in the present case due to a lack of general symptoms. The patient underwent a submucosal partial inferior turbinectomy and an adenotonsillectomy, and then the patient's nasal stuffness improved. Continued observation of the patient did not provide any evidence of disease progression or complications.

The present report is the first to describe a case of JPD associated with paranasal sinus aplasia, and the patients nasal symptom was successfully treated with surgery.

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