CASE REPORT

Worth syndrome “mandibular osteosclerosis” as an incidental finding: a report of 2 cases

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This report presents two cases of Worth syndrome involving the mandible which were identified as an incidental finding on radiologic evaluation. Both patients were females who presented with enlarged mandibles. Radiologic evaluation revealed multiple bilateral mandibular enostoses, widened and thickened inferior cortical border of the mandible, with no other major clinical finding on examination. One of the patients received orthognathic surgery and healed uneventfully with no post-surgical complications. In this report, we reviewed possible differential diagnoses to this syndrome to facilitate identification among general dentists and oral and maxillofacial radiologists.

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Introduction

Worth syndrome, also known as endosteal hyperostosis, autosomal dominant type is a rare genetic bone disorder grouped under craniotubular bone disorders.^{1,2} It is characterized by generalized endosteal and periosteal hyperostosis, presenting as cortical thickening of bones, bilateral widening and prognathism of the mandible with increased gonial angle.³⁻⁸ This condition was first described by Worth and Wollin as hyperostosis corticalis generalist congenita in 1966.⁵ It was further evaluated by Gorlin and Glass in 1977, suggesting the name autosomal dominant osteosclerosis and distinguishing it from Van Buchem disease, another severe form of craniotubular bone disorder characterized by autosomal recessive inheritance pattern, elevated alkaline phosphatase, neurologic complications, increased head circumference, nasal obstruction, hypertelorism and exophthalmos.³ Heterozygous mutations in the low density lipoprotein receptor-related protein-5 gene on chromosome 11q13 have been identified in patients with endosteal hyperostosis.⁹ In this report, we describe two patients with Worth syndrome which were identified as an incidental finding on radiologic evaluation. One of the patients

received orthognathic surgery and healed uneventfully with no known complication.

Case reports

Case 1

A 51-year-old female presented to her orthodontist for evaluation of an anterior open bite. On clinical examination, the lower jaw exhibited obvious enlargement and slight facial asymmetry, prompting acquisition of a cone beam CT (CBCT) exam. The CBCT dicom file was sent to an oral and maxillofacial radiologist (KT) for evaluation. Radiologic findings included multiple bilateral mandibular enostoses, osseous hyperplasia and inferior cortical border widening of both right and left sides of the mandible, obtuse gonial angle, and steep mandibular plane angle ([Figure 1](#)). A midline palatal torus, and proclined mandibular incisors which formed an acute interincisal angle were also present. No other significant medical history or radiologic findings were identified. Subsequently, the patient underwent an uncomplicated orthognathic surgery to correct the open bite. Healing progressed normally with no post-surgical complications.

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Figure 1 Radiologic images of Case 1. Panoramic reformat (A), axial and coronal views (B), bone volume rendering (C).

Case 2

A 40-year-old female presented to her dentist for routine oral evaluation. On clinical examination, there was no facial asymmetry, but the mandible appeared enlarged and bilateral mandibular lingual tori were present. A CBCT scan was performed as part of the comprehensive examination, which revealed mandibular opacities. The CBCT dicom file was sent to an oral and maxillofacial radiologist (KT) for evaluation. Radiologic findings included multiple, irregularly-shaped, well-defined osteosclerotic lesions along the endosteal surface of the inferior mandibular cortical border from the right first molar to the left first molar, widened and thickened inferior cortical border, obtuse gonial angle, and steep mandibular plane angle (Figure 2). Bilateral mandibular lingual tori and proclined mandibular incisors, which formed an acute interincisal angle were also present. No other significant medical history or radiologic findings were identified. Clinical and radiologic findings of both cases are summarized in Table 1.

Discussion

Worth syndrome is grouped under craniotubular bone disorders, hyperostosis type.^{1,2} Genetic craniotubular bone disorders were originally divided into two board

groups by Gorlin et al. in 1969: dysplasias and hyperostoses.^{1,2} Dysplasias are caused by defect in bone remodeling, while hyperostoses are caused by active overgrowth of bone. Examples of craniotubular dysplasias includes: Pyle's disease, craniometaphyseal dysplasia, craniometaphyseal dysplasia, frontometaphyseal dysplasia, osteopetrosis, dysosteosclerosis, and oculodontoosseous dysplasia. While, craniotubular hyperostoses includes: Van Buchem disease, sclerosteosis, Worth syndrome, Camurati-Engelmann disease, and congenital hyperphosphatasia.^{1,2} The clinical findings in Worth syndrome are typically found in the mandible.^{1,3-8} The changes in the mandible have been reported to usually start at puberty and plateau with cessation of growth, and tori on the palate or mandible have been reported.^{3,5}

Worth syndrome involving the mandible is a rare disease condition which is infrequently encountered by a general dentist or oral maxillofacial radiologist making its diagnosis a challenge. To aid the decision for a diagnosis of Worth syndrome, a summary of the clinical and radiologic findings are presented in Table 1. The differential diagnosis for a patient presenting with clinical findings similar to Worth syndrome in the mandible includes Van Buchem disease, Paget disease, cherubism, fibrous dysplasia, and florid osseous dysplasia.

Van Buchem disease is an autosomal recessive bone disorder. A severe form of craniotubular bone disorder

Figure 2 Radiologic images of Case 1. Panoramic reformat (A), axial and coronal views (B), bone volume rendering (C).

characterized by hyperosteoses of the skull, mandible, clavicles, ribs, spine, diaphyseal cortex of long and short bones, and elevated serum alkaline phosphatase.¹⁰ This produces neurologic complications, increased head circumference, nasal obstruction, hypertelorism and exophthalmos. Our patients did not present with any of these symptoms and no radiologic finding was identified in the skull.

Paget disease (PD) is a heterogeneous disease having both genetic and environmental aetiology. Genetic mutation of sequestosome 1 which encodes the p62

protein has been identified in patients with PD.¹¹ It is rarely diagnosed in patients younger than age 40 years. Serum alkaline phosphatase and urinary hydroxyproline levels are elevated in patients with PD. Jaw involvement of PD is present in 10–15% of cases, the maxilla is more frequently involved than the mandible and skull involvement produces increased head circumference, palatal expansion and neurologic complications. Radiographically, it presents as areas of irregular increased density interspersed by radiolucent areas having a cotton-wool appearance.¹² The maxilla was

Table 1 Summary of the clinical and radiologic findings of Worth syndrome cases

	Case 1	Case 2
Clinical findings	<ul style="list-style-type: none">• Slight facial asymmetry• Enlarged mandible• Palatal torus	<ul style="list-style-type: none">• No facial asymmetry• Enlarged mandible• Bilateral mandibular tori
Radiological findings	<ul style="list-style-type: none">• Multiple bilateral mandibular enostoses• Osseous hyperplasia and widening of the inferior cortical border of the mandible• Obtuse gonial angle• Steep mandibular plane angle	<ul style="list-style-type: none">• Multiple well-defined irregularly-shaped osteoscleroses along the endosteal surface of the mandible• Widening and thickening of the inferior cortical border of the mandible• Obtuse gonial angle• Steep mandibular plane angle
Genetic findings	<ul style="list-style-type: none">• Identification of mutations in the LDL receptor-related protein-5 gene on chromosome 11q13 was not performed	<ul style="list-style-type: none">• Identification of mutations in the LDL receptor-related protein-5 gene on chromosome 11q13 was not performed

LDL, low density lipoprotein.

not involved and no symptoms were reported in our patients.

Cherubism is an autosomal dominant inherited disease process characterized by remodelling whereby giant cell-containing fibrous lesion replaces the jaw bone. Mutations in SH3BP2 protein have been identified in patients with cherubism. The lesion typically affects children as young as age 2–5 years involving both bilateral posterior mandible and maxilla. It more commonly involves the mandible than the maxilla. Radiographically, it presents as expansile multilocular radiolucent lesion of the jaw bones with thinning of the cortex.¹³ Jaw images of our patients did not present with multilocular radiolucencies.

Fibrous dysplasia (FD) is a developmental dysplastic anomaly of bone-forming mesenchymal tissue; the cancellous bone is replaced by fibrous tissue and abnormal bone. There are two basic clinical forms: monostotic (involving a single bone) and polyostotic (involving multiple bones usually associated with a syndrome). Monostotic FD occurs more frequently in the jaws with the maxilla being more commonly involved than the mandible. Jaw involvement of FD usually presents with swelling or deformity, some cases are asymptomatic and are discovered as incidental findings on radiographic evaluation for some other reason.

Radiographically, FD typically presents as a ground-glass appearance.¹⁴ Our patients' images did not present with a ground-glass appearance.

Florid osseous dysplasia is an extensive form of osseous dysplasia involving multiple quadrants of the jaws, limited to the tooth-bearing segments. Florid osseous dysplasia is usually seen in middle-aged females of Black or Asian descent. Expansion of the jaw is rare and radiographically presents as lytic to sclerotic lesion depending on its stage of development.¹⁵ The lesions in our patients were not limited to the tooth bearing segments of the jaw but also involved the inferior cortical border of the mandible. In conclusion, we report two rare cases of Worth syndrome identified as an incidental finding on radiologic evaluation and reviewed possible differential diagnoses. A definitive diagnosis of Worth syndrome in these patients would require extensive genetic work-up and family tree analysis. Based on published reports in the literature, our patients presented with classic radiologic and clinical features of Worth syndrome.

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