CASE REPORT

Worth syndrome as a diagnosis for mandibular osteosclerosis

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This case report presents an unusual incidental radiographic finding on a dental panoramic radiograph that caused diagnostic confusion. A 46-year-old female presented with symmetrical sclerotic cortical thickening of the anterior mandible visible on radiograph, with no other major clinical examination findings. The patient subsequently showed no change in radiographic appearance over a 6 year period. Based on this evidence, and the benign nature of the history, we propose a diagnosis of Worth syndrome. This report aims to clarify the difference in terminology and presentation of two conditions known to produce mandibular osteosclerosis: Worth syndrome and Van Buchem disease, both of which are often quoted synonymously in the literature.

Keywords: mandibular osteosclerosis; Worth syndrome; Van Buchem disease

Case report

A 46-year-old female was presented to an oral and maxillofacial surgery clinic via her dental practitioner with an unusual incidental radiographic finding (Figure 1). This showed symmetrical sclerotic cortical thickening of the anterior mandible, extending half way along the body. On clinical examination she was noted to have prominent bilateral lingual tori, with an irregular lower mandibular border. No obvious facial asymmetry was noted.

The patient had a medical history of a Grade 1 node negative ductal carcinoma of the right breast. This had been treated curatively 1 year previously with wide local excision, post-operative radiotherapy and adjuvant tamoxifen hormonal therapy. For this reason the patient was concerned regarding the possibility of metastatic lesions from her previous carcinoma.

Routine haematology and serum bone markers—alkaline phosphatase, osteocalcin and Type I procollagen peptide—were all negative. From the history, further radiographic imaging was not indicated. After extensive discussion with radiologists and a review of the literature, a presumptive diagnosis of Worth syndrome was proposed to explain this condition.

The patient was reassured and followed up 6 months later with a repeat dental panoramic radiograph. There was no change in the radiographic appearance. Long-term follow-up at 6 years revealed no further changes (Figure 2).

Discussion

Worth syndrome is an autosomal dominant osteosclerosis with radiographic evidence of sclerosis and oropharyngeal exostoses. When reviewing the world literature, there appears to be confusion and conflicting information regarding the naming and presentation of this condition. This report therefore aims to introduce and clarify the main clinical and radiographic features of this syndrome.

In 1966, Worth and Wollin first described a condition of benign and usually asymptomatic osteosclerosis associated with torus palatinus and torus mandibularis.1 Gorlin and Glass2 separated this syndrome from Van Buchem disease (hyperostosis corticalis generalisata), by noting the autosomal dominant inheritance pattern of Worth syndrome.

Both conditions are rare hereditary sclerosing bone dysplasias. Classically, Worth syndrome shows autosomal dominance, a benign disease progression and features confined to thickening of the mandible. It does not progress beyond teenage years.3 In contrast, the autosomal recessive Van Buchem disease exhibits bone hypertrophy of the skull base and mandible, with possible involvement of ribs, clavicle, spine and pelvis.
This can produce exopthalmos, hypertelorism, increased head circumference, cranial nerve involvement and nasal obstruction, with symptoms presenting in childhood and showing a progressive course. Elevated serum bone markers are observed in Van Buchem disease but not in Worth syndrome.4

Part of the diagnostic confusion stems from the fact that Van Buchem disease and Worth syndrome are often quoted synonymously in literature or by multiple descriptive terms. The terminology “autosomal dominant osteosclerosis” is commonly applied to Worth syndrome, and “hyperostosis corticalis generalista” or “endosteal hyperostosis” for Van Buchem disease. The authors also found the terms Type I (Van Buchem disease) and Type II (Worth syndrome) hyperostosis corticalis generalista used in various articles.

Several papers have linked mutations in the LDL receptor-related protein five gene, with conditions of increased bone density and oropharyngeal skeletal disease. It has been proposed that there is a link between Van Buchem disease, Worth syndrome, osteopetrosis Type I and osteoporosis-pseudoglioma syndrome.5 This is an area of ongoing research and in the future may assist in providing a definitive diagnosis.

In this case there was no visible facial deformity and the patient was happy to be discharged without additional investigation. Some case reports have described success with surgical recontouring of the lower border of the mandible, particularly when managing mandibular bone hypertrophy produced by the hereditary sclerosing bone dysplasias.6,7 They achieved a satisfactory aesthetic result, with minimum morbidity and acceptable long term stability.

This case demonstrates no clinical features of Van Buchem disease, as one would expect to see a progressive disease presenting in childhood along with deranged bone marker levels. However, there is no evidence of an obvious positive autosomal dominant inheritance pattern to corroborate Worth syndrome. As both conditions are known to show mandibular bony involvement, this case may either be Worth syndrome, a variant of Van Buchem disease, or simply extensive

Figure 1 Panoramic radiograph at patient presentation, showing symmetrical mandibular cortical thickening

Figure 2 Panoramic radiograph at 6 year follow-up, showing no change to previous radiograph
mandibular tori. A definitive diagnosis is difficult to obtain without detailed genetic testing and in-depth family tree analysis. A bone tissue biopsy would have ruled out metastatic spread, but in the likely case of benign bone sclerosis would not have further aided a diagnosis. With the benign radiographic appearance, and with no further advancement at 6 months and 6 years, the risk of surgery and the patient’s own wishes outweighed the benefits of an invasive procedure.

Although not presenting a definitive diagnosis, this case does, however, display a fascinating radiograph, with an irregular and benign pattern of bone deposition around the lower border of the mandible that has not been widely reported in the maxillofacial radiological literature.

References