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CASE REPORT

Successive generations with inherited craniofacial fibrous dysplasia

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Abstract Craniofacial fibrous dysplasia is a benign fibro-osseous lesion of bone that only affects the bones of the craniofacial complex. Here, we report a case of craniofacial fibrous dysplasia in a 16-year-old Thai male who presented with mild swelling and tenderness at the mandibular right first molar area and ipsilateral nasal congestion. Conventional and cone-beam CT radiographic examinations were performed. The radiographs revealed multiple mixed radiolucent and radiopaque lesions involving most of the craniofacial bones. The first biopsy from the right mandibular area was diagnosed as juvenile ossifying fibroma, whereas a biopsy from the right maxillary area was diagnosed as fibrous dysplasia. The defects appeared to have a genetic basis, because his mother and younger brother had the same clinical and radiological findings. Furthermore, the family history given by his mother revealed that several other members of her family had similar clinical signs and symptoms. We diagnosed this case as inherited craniofacial fibrous dysplasia on the basis of previously reported clinical, radiographic and histologic findings as well as family history.

Keywords Monostotic fibrous dysplasia · Polyostotic fibrous dysplasia · Craniofacial fibrous dysplasia · $G_s\alpha$ gene

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Introduction

Fibrous dysplasia is a congenital nonhereditary pathologic condition of bone characterized by replacement of normal bone with fibrous connective tissue, followed by the development of multiple mineralized masses in this tissue. Most previous reports have indicated that the etiology of fibrous dysplasia is linked to a mutation in the $G_s\alpha$ gene located at chromosome 20q13.2-13.3 [1, 2]. The extent of fibrous dysplasia is believed to be related to the point during embryonic or postnatal development when the postzygotic mutation in $G_s\alpha$ occurs [3, 4]. The severity and extent of $G_s\alpha$ mutation-associated diseases are also related to the degree of proliferation of the mutated cells within the clone during migration, growth, and differentiation as well as the ratio of mutated to normal cells at the affected anatomical sites [3, 5].

Fibrous dysplasia may affect single (monostotic) or multiple (polyostotic) bones. The monostotic form is more common, occurring in 75–80% of fibrous dysplasia cases [6]. The polyostotic form is divided into three types: (1) craniofacial fibrous dysplasia, in which only the bones of the craniofacial complex, including the mandible and maxilla, are affected; (2) Lichtenstein–Jaffe type, in which multiple bones of the skeleton are involved, with instances of café-au-lait pigmentation of the skin and rare endocrinopathies; and (3) Albright's syndrome, characterized by a triad of severe polyostotic fibrous dysplasia (mostly