Ramon syndrome: report of a rare case

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ABSTRACT Cherubism and gingival fibromatosis are frequently isolated conditions. Cherubism primarily affects the jaw bones causing painless bilateral swelling and disfigurement. When it co-exists with gingival fibromatosis, epilepsy, and mental retardation, it is referred to as Ramon syndrome, first described by Yachanan Ramon in 1967. Presently, we describe a particularly rare case of non-familial Ramon syndrome.

Key words: Cherubism; Epilepsy; Fibromatosis, gingival; Mental retardation

Introduction

Gingival fibromatosis is a rare disease of unknown etiology. This form of gingival enlargement usually appears early in childhood and is also known as idiopathic hyperplasia or hereditary gingival fibromatosis. Several theories have been advocated to explain the etiology of this condition. One such theory suggests that an inherited constitutional factor plays an etiological role. Dense, fibrous gingiva is the main oral manifestation of idiopathic hyperplasia. The enlarged gingiva is firm and pink, but is generally not erythematous, except at sites of local inflammation with a tendency to bleed.

Cherubism causes painless swelling of the maxillary and mandibular bones and facial disfigurement. With maxillary enlargement and consequent elevation of the orbital floor, the lower sclera of the eyes is more exposed, making it seem as if patient is looking upward to heaven—hence the origin of the term cherubism. The association of gingival fibromatosis and cherubism with epilepsy, mental retardation, and stunted growth was first described by Ramon et al. Presently, we report a rare case of gingival fibromatosis, cherubic facial features, epilepsy, and mental retardation.

Case report

An 11-year-old boy presented with signs of gingival enlargement, bilateral cheek fullness, painless swelling of the jaw, and mental retardation in July 2004. The patient was epileptic and was on Dilantin (Parke-Davis Pharmaceuticals, India) for 9 years. Neurological examination revealed that the patient's social development and motor skills were at the level of a 5-year-old and 6-year-old, respectively. Laboratory investigations demonstrated an elevated serum alkaline phosphatase level to 262 IU/L. Chest X-ray was normal. Karyogram was found to be normal 46XY. No similar characteristics were noted in his pedigree. Likewise, the patient did not have any history of congenital or metabolic conditions.

Extraoral examination revealed bilateral painless mandibular swellings with fullness of cheeks and incompetent lips (Figure 1). Intraoral examination revealed grossly hyperplastic gingiva completely covering all the teeth in both jaws. The gingival ridges were firm and non-erythematous (Figure 2). Panoramic radiography revealed well-defined multi-ocular osteolytic lesions in the body and ramus of the mandible sparing the condylar and coronoid processes, suggestive of cherubism (Figure 3). There was no radiographic evidence of any bony lesions in the maxilla. Gingival biopsies were taken from the posterior region of mandible under local anesthesia.

Histopathologic appearance of the gingivectomy specimens revealed hyperplastic dense, fibrous connective
tissue, compatible with gingival fibromatosis (Figures 4 and 5). The clinical and histopathological findings were both suggestive of the diagnoses of gingival fibromatosis and cherubism. Laboratory findings were within normal limits and no systemic or metabolic causes were found in our case. Since the patient was the only child in the family, no other evidence of hereditary factors could be brought to light.

We decided to perform a full-mouth gingivectomy under general anesthesia in two stages. The immediate results were encouraging. There was a dramatic cosmetic improvement and the healing was uneventful. However, as it has been reported that the osteolytic lesions resolve
when the patient reaches adulthood, no specific treatment was considered for these lesions. Subsequently, the patient was lost to follow-up.

Discussion

Idiopathic gingival fibromatosis and cherubism are two rare and apparently unrelated conditions. In our case, the gingival fibromatosis, one feature of Ramon syndrome, was present since childhood. The patient had been receiving phenytoin for seizures. Gingival hyperplasia is a side-effect of phenytoin and generally is noticed 3 or more months after the drug is started. The clinical appearance of cherubism may vary from barely discernible posterior swellings of a single jaw to a grotesque expansion of both jaws, with concomitant difficulties in mastication, speech, swallowing, and respiration. Ramon et al. described two siblings with cherubism, gingival fibromatosis, epilepsy, mental deficiency, hypertrichosis, and stunted growth. Pina-Neto et al. described the same disorder in four individuals of the same Brazilian family. The features were identical to those of cases reported by Ramon et al., except that three of the four patients had juvenile rheumatoid arthritis. Progressive cherubic facies become evident after the age of 4 years. Gingival enlargement is a constant feature. It manifests after 2 years of age and ultimately prevents jaw closure. Tooth eruption is delayed and incomplete. Mild-to-moderate mental retardation becomes evident during the first few years of life. By the fourth year, major seizures develop. Stunted growth was evident in nearly all affected siblings. Hypertrichosis was present in 50% of cases. de Pina-Neto et al. reported that rheumatoid arthritis should also be considered to be part of the syndrome. It is possible that these patients did not have rheumatoid arthritis, but rather polyarticular pigmented villonodular synovitis. The finding that affected siblings had normal but consanguineous parents suggests an autosomal recessive mode of inheritance. The features of our own patient are tabulated in the Table.

In summary, Ramon syndrome, which presents as gingival fibromatosis associated with epilepsy, mental retardation, and cherubic lesions of the jaw, is rare.

References