CASE REPORT

Non-familial cherubism: clinical and radiological findings

Anuradha Agrawal,¹ Saurabh Kumar Gupta,² Payal Saxena,² Pratiksha Kumar³

¹Department of Pediatric Dentistry, Government Dental College, Indore, Madhya Pradesh, India

²Department of Conservative Dentistry and Endodontics, Government College of Dentistry, Indore, Madhya Pradesh, India

³Department of Oral Pathology and Microbiology, Government College of Dentistry, Indore, Madhya Pradesh, India

Correspondence to
Dr Saurabh Kumar Gupta,
payal.endo@gmail.com

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SUMMARY

Cherubism is a rare, self-limiting, usually hereditary, benign fibro-osseous disease mostly affecting the jaws of the paediatric population. The disease is characterised by bilateral, diffuse and multilocular bony enlargement of the mandible and maxilla, and a tendency towards spontaneous remission. It has a typical radiographic and histopathological appearance. We present a case of a 9-year-old child lacking family history of this genetic disorder.

BACKGROUND

Cherubism is an extremely rare entity with a limited number of cases recorded in the literature. Owing to the rarity of this disease, determination of its frequency is found to be difficult and hence its prevalence is not known.¹ Cherubism is a benign bone dysplasia characterised by bilateral, progressive, painless, diffuse enlargement of the maxilla and/or mandible with a typical radiographic presentation due to replacement of normal bone with multilocular fibrovascular cysts. First described by Jones in 1933, the condition is usually limited to the jaws and has a tendency of regressing with the onset of puberty.²

Familial form of cherubism is inherited as an autosomal dominant disorder with mutations in the SH3-domain binding protein. In few cases, it may develop without family history and is assumed to be due to de novo mutations or possible genetic heterogeneity.² This case report presents an even rare case of non-familial cherubism in a 9-year-old boy with characteristic marked bilateral mandibular swelling affecting his dentition.

CASE PRESENTATION

A 9-year-old boy reported to the department of pedodontics and preventive dentistry with slowly expanding, painless swelling on both sides of the cheek since 3 years. No other sibling or parents had similar finding. Swelling was more prominent on the right side of the face as compared with the left side (figure 1). There was a history of difficulty in mastication due to derangement of all the teeth. Extraorally, marked diffuse swelling was present on both sides of the face. Swelling was bony, hard and non-tender on palpation. The skin over the swelling was normal. Right submandibular lymph nodes were palpable. Intraorally, the patient had both the dentition. Teeth present were normal in size but displaced and deviated from their normal position with multiple missing teeth in the lower jaw. Permanent molars were mesially migrated and had

Figure 1 Bilateral asymmetrical swelling of the maxilla and mandible.

resorption in their roots. Extension of the swelling on the right side was from right primary lateral incisor to permanent molar area (figure 2) and from left primary lateral incisor to permanent molar area (figure 3). Expansion of buccal and lingual cortical plates was observed. The overlying mucosa appeared normal.

Figure 2 Intraoral view of the right side showing expansion of jaws and irregular dentition and multiple missing teeth.

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Figure 3 Intraoral view of the left side showing expansion of jaws and irregular dentition on the left side.

INVESTIGATIONS

Orthopantomogram showed bilaterally multilocular radiolucent areas, with marked bony expansion in the mandible and maxilla (figure 4).

TREATMENT

On the basis of clinical and radiological examinations, the diagnosis of cherubism was made. Knowing the expected tendency of the lesion of becoming static and showing regression at puberty, surgical intervention was deferred and the patient was kept on regular follow-up.

DISCUSSION

Cherubism, considered a childhood disease, usually presents before the age of 5 years and usually the affected children are born normal. The lesion characteristically progresses until puberty and then its progression stops. Later on, the disease may show spontaneous partial or complete regression.^{3 4} In

Figure 4 Panoramic radiograph presents expansive multilocular cystic lesions distributed in the mandible and maxilla with loss and mesial displacement of the deciduous teeth.

majority of cases it represents symmetrical lesion but in our case it was more prominent on the right side. Earlier also, such cases have been reported in different literatures.^{5–8} The radiographic presentation of the disease is characteristic and has bilaterally symmetrical, well-defined multilocular radiolucencies in the mandible, with infrequent involvement of the maxilla.⁹ Sparing of the mandibular condyle has been considered a hallmark of the condition.^{5 9} The above case depicts a similar, classical radiographic appearance of the disease with non-involvement of the mandibular condyle. Since the lesion is known for undergoing spontaneous regression, delaying surgical intervention is considered better until after puberty. However, in patients with cosmetic and functional problems or emotional disturbances, surgical intervention may be considered.^{6 10}

Learning points

- ▶ Even though rare, cherubism creates considerable distress in involved children and their families.
- ▶ Careful clinical examination and proper radiographic interpretation is helpful in diagnosing cases of non-familial cherubism.
- ▶ During treatment planning, one should delay any surgical intervention until puberty as the disease tends to regress thereafter in most cases.

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Competing interests None.

Patient consent Obtained.

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