RESEARCH

Imaging evaluation of the gingival fibromatosis and dental abnormalities syndrome

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Objective: The purpose of this study was to evaluate the dentomaxillofacial imaging features of one family affected by the gingival fibromatosis (GF) and dental abnormalities (DA) syndrome.

Methods: Conventional radiographs (periapical and panoramic) and cone beam CT (CBCT) were performed in nine members of this family: four were affected by the syndrome and five were not.

Results: The four affected members demonstrated mild generalized GF in association with DA, including hypoplastic amelogenesis imperfecta, intrapulpal calcifications, delay on tooth eruption and pericoronal radiolucencies in unerupted teeth. None of these oral changes were identified in the five unaffected members. All nine members presented alterations in the paranasal sinuses and mucosal thickening of the maxillary sinus was the most common finding.

Conclusion: Family members not affected by the syndrome showed similar alterations in the paranasal sinuses and CBCT was useful to characterize the dentomaxillofacial features of this new syndrome associating GF and DA.

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Introduction

Gingival fibromatosis (GF) is the overgrowth of the gingiva characterized by an expansion and accumulation of the connective tissue with occasional presence of an increased number of cells.¹ It is induced as a side effect of systemic drugs, including the antiepileptic drug phenytoin, the immune suppressor cyclosporine and the calcium channel blocker with antihypertensive activity nifedipine, or it is hereditary.² As an inheritance disorder, GF may be isolated, being referred to as hereditary gingival fibromatosis (HGF), or part of a

genetic syndrome.³ GF as a syndromic feature has been described in GF with hypertrichosis and/or mental retardation syndrome, GF with distinctive facies syndrome,⁴ Zimmermann-Laband syndrome, Cross syndrome, Rutherford syndrome,³ Murray-Puetic-Drescher syndrome,⁵ Jones syndrome⁶ and Prune Belly syndrome.⁷ Recently, we described a new association of GF and dental abnormalities (DA), including generalized thin hypoplastic amelogenesis imperfecta (AI).⁸

The clinical expression of GF is highly heterogeneous and the gingival enlargement results in both aesthetic and functional problems for affected individuals.⁹ There are few studies reporting imaging alterations associated with GF as this is a condition that does not directly affect hard tissue. However, the increase in gingival thickness

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may result in impacted teeth.¹⁰ Furthermore, the gingival swelling may increase the bacterial plaque accumulation, inducing periodontitis and bone resorption, which can be seen radiographically.²

Cone beam CT (CBCT) in the dentomaxillofacial region allows the creation in “real time” of images, not only in the axial plane, but also two-dimensional (2D) images in the coronal, sagittal and even oblique or curved image planes, a process referred to as multiplanar reformation. In addition, CBCT data also permit three-dimensional (3D) representation of the maxillofacial skeleton with minimal distortion.¹¹ The purpose of the present study is to provide an overview of the dentomaxillofacial imaging features of patients affected by GF and DA syndrome.

Materials and methods

Subjects

Recently, we described an extensive family with GF and DA syndrome.⁸ Clinical examination of the 70 family members revealed 1 member affected by isolated GF, 3 affected by GF and DA, and 1 member with GF and DA in association with mental retardation (MR). Six members presented with isolated MR. All affected members came from consanguineous marriage, except for the member with isolated GF. Pedigree analysis provides ample evidence that GF and DA were transmitted together by an autosomal recessive gene, whereas MR was superimposed with GF and DA in one patient, suggesting that the coexistence of MR and GF plus DA may represent a coincidence of two conditions in the same family with multiple consanguineous marriages. The patients included in the present study were the four individuals affected with the complete features of the syndrome and another five unaffected members of the same family.

Imaging analysis

Imaging studies included periapical and panoramic radiographs and CBCT. These images were acquired by the device I-CAT (Imaging Sciences International, Hatfield, PA), 120 kV and 36 mAs, with voxel of 0.25 mm and field of view of 22 cm. Programs for computer graphics (Xoran Technologies, Inc., Ann Arbor, MI) were used to perform multiplanar reconstructions, such as panoramic, orthoradial and 3D views, showing in detail the dental morphology and its relation to neighbouring areas. The multiplanar reconstructions were performed at 0.25 mm thick for dental evaluation, paranasal sinuses and cheek bones. Orthoradial slices were of 1 mm thickness and represented in scale 1:1. The spatial location of these slices was made by correlating the number on the upper left of the image with the horizontal rule existing in the lower portion of the reconstruction view. Images were analysed by two oral radiologists to reach consensus in

interpretation of the radiographic findings. This study was carried out with approval from the Human Research Ethics Committee of the State University of Montes Claros, Minas Gerais State, Brazil.

Results

The clinical features of the four affected members were quite similar. The gingival overgrowth partially covered the dental crowns of all teeth, and the erupted teeth presented yellow discoloration with diastemas and thin enamel of normal hardness (Figure 1). Dental findings of the patients of this study are depicted in Table 1. Periapical and panoramic radiographs showed a lack of differentiation in the density between enamel and dentine, intrapulpal calcifications and impacted teeth with hyperplastic follicles (Figures 2 and 3). After completing the CT scans it was possible to identify dental alterations with more precision, discovering that, with the exception of some third molars, all other teeth including those that were unerupted presented intrapulpal calcifications (Figure 4). Some pericoronal follicles were wide, reaching more than 2.5 cm and involving up to two dental elements (Figure 5). Beyond the hyperplastic pericoronal follicles, some unerupted teeth also had dilacerated roots and the axis of eruption modified with displacement of the root in the direction of the mandibular canal or maxillary sinus (Figures 4 and 6a). The lamina dura and the periodontal space were normal in all erupted teeth.

Both affected and unaffected members demonstrated alterations in the paranasal sinus or nasal cavity (Table 2). The most prevalent abnormality was mucosal thickening of the maxillary sinus (Figures 6a, b and 7). Opacification of the frontal sinus, discrete nasal septal deviations and turbinate hypertrophy with obstruction of the osteomeatal communication were also observed. Unaffected family members did

Figure 1 Clinical photograph of patient 1. Intraoral examination revealed a mild enlargement of the gingival tissue, and the permanent dentition demonstrated yellow discoloration and thin enamel

Table 1 Dental features of the patients of this study

<i>Patient</i>	<i>Age</i>	<i>Sex</i>	<i>NT present</i>	<i>NT unerupted</i>	<i>NT with pulp stones</i>	<i>NT with AI</i>	<i>NT with dilaceration root</i>
Affected							
1	20	Female	31	15	28	31	9
2	15	Female	31	11	28	30	2
3	15	Female	32	08	28	32	2
4	20	Male	29	14	27	29	10
Control							
5	49	Female	9	0	0	0	0
6	38	Female	26	0	0	0	0
7	28	Female	31	0	0	0	0
8	19	Male	32	4	0	0	0
9	19	Male	30	3	0	0	0

NT, number of teeth; AI, amelogenesis imperfecta

not have the dental changes found in patients affected by the syndrome, such as intrapulpal calcifications, AI, deformed roots and hyperplastic pericoronal

follicles (Figure 7). The only unerupted teeth in the control group were some third molars of patients eight and nine.

Figure 2 Periapical radiographs of patient 2. Note intrapulpal calcifications and lack of contrast between enamel and dentine. The large radiolucent area was associated with second lower molars

Figure 3 Panoramic radiograph of patient 3. Similarly to the other cases, enamel was impossible to differentiate from dentine and intrapulpal calcifications were observed in erupted and unerupted teeth. Impacted molars and large radiolucent pericoronal areas associated with molars were also noted

Discussion

Studies had confirmed the advantages of the CBCT in the dentomaxillofacial evaluation of uncommon disorders—providing a better knowledge and plan of treatment.^{12–14} Several studies had demonstrated the utility of the CBCT to evaluate similar situations as found in this study, such as impacted teeth,¹⁵ dental displacements¹⁶ and maxillary sinus membrane thickening.¹⁷ Hashimoto *et al*¹⁸ affirmed that through the CBCT it was possible to distinguish enamel, dentine,

pulp cavity and alveolar bone with better clarity, whereas Noujeim *et al*¹⁹ stated that the CBCT technique has better accuracy and diagnostic value than periapical films in the detection of periodontal bone defects. The periapical and panoramic radiographs taken initially had allowed the identification of some dental alterations. However, CBCT scans allowed verification of the alterations with more details and also quantified the tooth number with abnormalities. In the affected members, it was observed that all teeth, including the impacted ones, presented difficulty of differentiation of

Figure 4 Panoramic reconstruction of CT of patient 1. Note the intrapulpal calcification, lack of contrast between enamel and dentine in erupted and unerupted teeth and large radiolucent pericoronal areas associated with impacted molars. Note also the displacement of the premolars into the maxillary sinus and the molars into the mandibular canal

Figure 5 Cone beam CT of patient 2. Cross-section slices depicting large radiolucent pericoronal areas associated with the second lower molars. Note the intrapulpal calcifications, lack of contrast between enamel and dentine and displacement of the second lower molars into the mandibular canal

the density between enamel and dentine, suggesting hypoplastic AI.

The imaging findings of the affected members demonstrated the combination of AI with retardation in the dental eruption, deformed roots, intrapulpal calcifications and increased pericoronal follicles. The presence of the unerupted teeth may be associated with

the GF or AI, since there are studies reporting that the increase of the gingival thickness results in delay in the dental eruption,²⁰ while other studies describe patients with AI and delay in the eruption of teeth without the presence of gingival overgrowth.²¹ Feller *et al*²² and Roquebert *et al*²³ have reported single cases of patients with imaging findings very similar to the ones of this

Figure 6 (a) Sagittal view of patient 4 demonstrating mucosal thickening of the right maxillary sinus. Note the impacted premolars and molars, intrapulpal calcifications and lack of contrast between enamel. (b) Coronal view of patient 2 depicting mucosal thickening of the right maxillary sinus and hypertrophy of nasal turbinates. Note microdontia of the third right upper molar and retention of the other molars

study, including gingival overgrowth, AI, delayed eruption, intrapulpal calcifications, malformed roots and hyperplastic dental follicles.

Paranasal sinus alterations are common findings of the cranial CT, but its importance as a triggering factor to the infectious process is still undefined.²⁴ The prevalence of paranasal sinus alterations in 100% of the patients of this study was higher than that described in the literature. In asymptomatic adult patients, Havas *et al*,²⁵ evaluating CT scans of the skull, found 42% of

abnormalities in 1 or more paranasal sinus and the thickness growth of the mucosa of the ethmoidal sinus was the alteration more frequently identified. In studies with patients aged from 6 years to 22 years, Boari and Castro Júnior²⁶ found radiographic opacification of the paranasal sinus in 93% of the cases; only 20% were symptomatic, and they concluded that diagnosis of the chronic rhinosinusitis was superior in the analyses carried out through image exam in relation to anamnesis. The causes of mucosa thickness growth or

Table 2 Imaging findings of the paranasal sinuses and nasal cavity of the patients

Patient	Frontal sinus	Maxillary sinus	Sphenoid sinus	Ethmoidal sinus	Osteomeatal complex	Nasal septum	Nasal turbinates
1	Normal	Bilateral thickening	Opacification	Normal	Normal	Moderate deviation	Asymmetric
2	Atretic	Bilateral thickening	Opacification	Normal	Unilateral opacification	Normal	Asymmetric
3	Normal	Bilateral thickening	Normal	Normal	Unilateral opacification	Discrete deviation	Asymmetric
4	Normal	Bilateral thickening	Normal	Normal	Unilateral opacification	Discrete deviation	Symmetric
5	Normal	Unilateral thickening	Normal	Unilateral opacification	Normal	Moderate deviation	Asymmetric
6	Normal	Unilateral thickening	Normal	Unilateral opacification	Normal	Discrete deviation	Symmetric
7	Normal	Bilateral thickening	Normal	Bilateral opacification	Bilateral opacification	Discrete deviation	Asymmetric
8	Normal	Unilateral thickening	Normal	Normal	Unilateral opacification	Discrete deviation	Asymmetric
9	Normal	Bilateral thickening	Normal	Unilateral opacification	Unilateral opacification	Discrete deviation	Asymmetric

Figure 7 Cone beam CT of patient 8. Coronal view showing thick mucosa of the left and right maxillary sinus. Note normal contrast between enamel and dentine and absence of intrapulpar calcifications in the teeth

opacification of the paranasal sinus without clinical symptoms are not well known, but they may be caused by the physiological cycle of increased volume of the nasal mucosa²⁶ or by another factor that has led to the inflammation of the mucosa, such as asthma, rhinitis, chemical products and viral or bacterial infections.²⁷ The fact that the patients belong to the same family and coexist in one environment may justify the fact that all present alterations are in the paranasal sinuses.

In conclusion, this study describes the dentomaxillofacial imaging features of a rare syndrome characterized by gingival fibromatosis and dental abnormalities. The radiographic aspects of this syndrome are important for the early and correct diagnosis, which can be determined by an oral radiologist. CBCT was useful for the detection and characterization of those alterations.

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