An Unusual Case of Hybrid Odontogenic Tumor in Type 1 Myotonic Dystrophy Patient

Abstract

Myotonic dystrophy, also referred myotonic muscular dystrophy, is an autosomal dominant, slowly progressive, multisystem disease characterized by skeletal muscle weakness, wasting, and myotonia. A hybrid tumor of odontogenic apparatus is a lesion showing combined histopathological characteristics of two or more previously recognized odontogenic tumors and/or cysts of different categories. We, therefore, report a case of hybrid tumor (adenomatoid odontogenic tumor associated with calcifying cystic odontogenic tumor) in a myotonic dystrophic patient.

Keywords: *Adenomatoid odontogenic tumor, hybrid tumor, myotonic dystrophy*

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Introduction

Myotonic dystrophy (DM), also referred myotonic muscular dystrophy, is an autosomal dominant, slowly progressive, multisystem disease characterized by skeletal muscle weakness, wasting, and myotonia. It can be divided into type 1 myotonic dystrophy (MD1) or Steinert's Disease and type 2 myotonic dystrophy, earlier known as proximal myotonic myopathy.[1] Hybrid tumor is a rare condition that can affect the oral maxillofacial region and usually occurs in adults as an asymptomatic swelling. There are only 26 reported cases. To date, no data are reported on the presence of hybrid odontogenic lesions in MD1. Herein, we report on a 28-year-old man affected by MD1, presenting a hybrid odontogenic tumor.

Case Report

A 28-year-old male patient came to the department of oral medicine and radiology with a chief complaint of swelling on the lower right anterior region for the past 2 months [Figure 1]. The patient noticed the swelling 2 months back, which was initially smaller in size and gradually increased in size. The increase in the size of swelling was associated with tooth mobility in the mandibular right quadrant. The swelling was associated with lingering pain with no associated aggravating and relieving factors and could not be linked to any history of previous trauma or dental infection.

He had been diagnosed with DM on clinical grounds previously (before 4 years). A positive family history for DM was also elicited. Proband has two healthy sisters. His mother was healthy till her late 40s, but started showing symptoms in her early 50s. Her two sisters were diagnosed with the condition, in which one of them, who was severely affected expired 2 years before.

Generalized weakness with apparent atrophy of the facial muscles; his neck and face were thin and elongated, gave the patient a "swan neck appearance," and there was characteristic baldness suggestive of frontoparietal alopecia. Bilateral palpebral ptosis was noticed which occasionally gave him the impression that he was dozing off. The upper lip was elevated in the form of an inverted "v," and this elevation left the mouth partly open. Atrophy of the temporalis and masseter muscles, which give a sunken appearance to this region of the face [Figure 2]. He was unable to lift his head from the dental chair. These findings led us to confirm the diagnosis of Steinert's DM.

On extraoral examination, a diffuse swelling was present on the right lower back tooth region extending superoinferiorly from the right commissure to the inferior border of the mandible and anteroposteriorly from the midline to 2 cm posteriorly. The skin over the swelling was normal in color with no

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visible pulsations. On palpation, the swelling was soft to firm in consistency and tender, no localized rise in temperature.

On intraoral examination, a solitary buccolingual swelling measuring about 4 cm × 3 cm was present on the mandibular right labial and buccal region obliterating the vestibule. The swelling extended from distal aspect of 42 to the distal aspect of 45 [Figure 3]. On palpation, swelling was soft to firm in consistency, tender, noncompressible, and without any fluctuations.

Investigations

The pulp vitality test was done using an electric pulp tester in relation to 42, 43, and 44. It showed a hyperresponse, which suggested an exaggerated response.

Orthopantomagram revealed a large ill-defined, noncorticated interradicular radiolucent lesion with 43 and 42 regions causing pathologic drifting and external root resorption of 42, 41, and 31 (knife-edged) [Figure 4].

Fine-needle aspiration cytology was done, which revealed yielded straw-colored fluid containing abundant shiny granules indicative of "cholesterol crystals" [Figure 5]. Fluid was mounted on slide and was observed under light microscope with ×10 and ×40 lens revealed "cholesterol crystals with broken corners," and the protein content of the fluid was estimated to be 6 mg/dl.

The lesional tissue was removed completely by surgical excision under local anesthesia, and the specimen was sent for histopathological examination.

Histopathology revealed a prominent basal layer with low columnar to cuboidal cells with a palisaded nucleus. Sheets of cuboidal basophilic cells extending from the lining and were noted with stellate reticulum-like cells. Some areas show the formation of duct-like structures [Figure 6]. The superficial layer showed ghost cells associated with abundant dentinoid material which is suggestive of hybrid odontogenic tumor composed of calcifying cystic odontogenic tumor (CCOT) and adenomatoid odontogenic tumor (AOT).

Linking the radiographic, clinical, and final diagnoses, ameloblastoma and its variations, central giant cell granuloma, AOT, and keratocystic odontogenic tumor in the differential diagnosis were took into account.

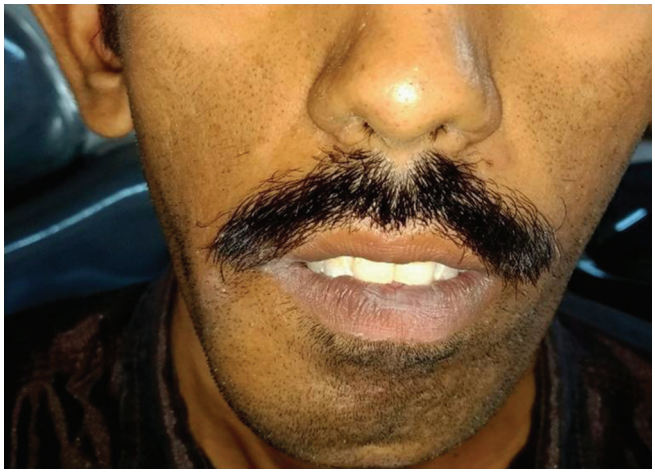

Figure 1: Swelling on the lower right anterior region

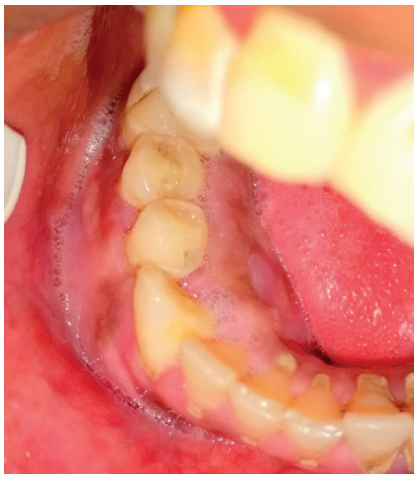

Figure 3: A solitary buccolingual swelling measuring about 4 cm × 3 cm was present on the mandibular right buccal and lingual region

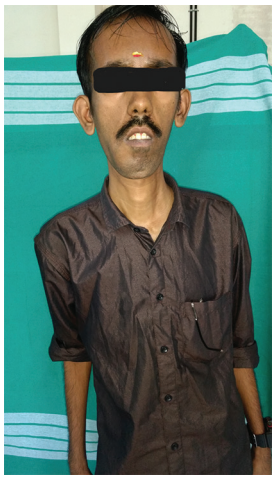

Figure 2: Generalized weakness with apparent atrophy of the facial muscles; "swan neck appearance," and there was characteristic baldness suggestive of frontoparietal alopecia

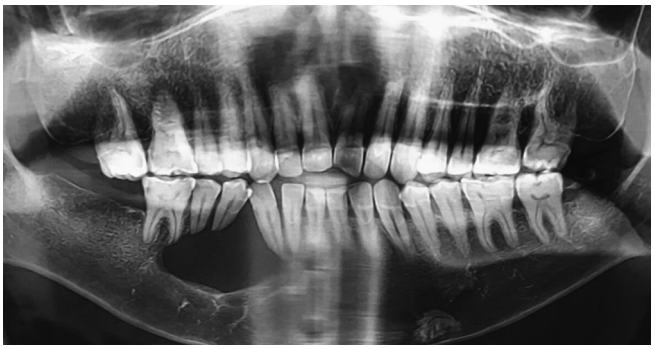

Figure 4: A large ill-defined, noncorticated interradicular radiolucent lesion with 43 and 42 regions causing pathologic drifting and external root resorption of 42, 41, 31 (knife-edged)

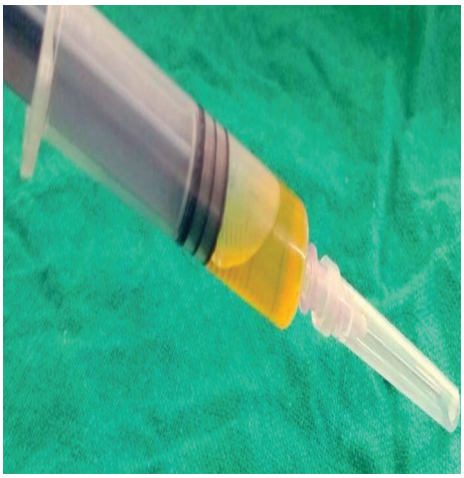

Figure 5: Fine needle aspiration cytology was done, which revealed yielded straw-colored fluid containing abundant shiny granules indicative of "cholesterol crystals"

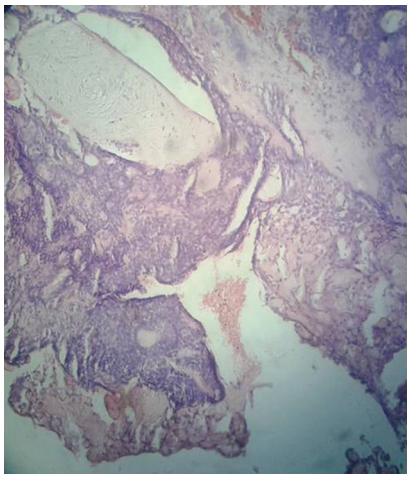

Figure 6: A prominent basal layer with low columnar to cuboidal cells with palisaded nucleus. Sheets of cuboidal basophilic cells extending from the lining and were noted with stellate reticulum-like cells

Discussion

A hybrid tumor of odontogenic apparatus is a lesion showing combined histopathological characteristics of two or more previously recognized odontogenic tumors and/or cysts of different categories.[2]

We, therefore, report a case of AOT associated with CCOT in a myotonic dystrophic patient.

There have been multiple reports of tumors in individuals with DM, most commonly benign calcifying cutaneous tumors known as pilomatricomas.[3]

Dystrophies are mainly caused by mutations in genes coding essential proteins for the integrity of muscular fibers, affecting functional muscular contraction and relaxation. These muscular dystrophies can be divided into six major types – Duchenne dystrophy, facioscapulohumeral dystrophy, limb-girdle dystrophy, MD, ocular dystrophy, and distal myopathy.[4]

In this paper, we report the first case of a hybrid odontogenic tumor composed of CCOT and AOT in a 28-year-old Indian male with Steinert's syndrome.

The radiographic findings of hybrid tumor frequently resemble other odontogenic lesions such as AOT, calcifying odontogenic tumors, ameloblastomas, and odontogenic keratocysts.[5]

Conservative surgical enucleation is the treatment modality of choice because of its low tendency to recur.

Gross sectioning of the tumor revealed more larger calcified masses, minimal yellow-brown fluid, fine, hard "gritty" granular material, and white to tan solid to crumbly tissue.[6]

Usually, hybrid tumor present with a painful jaw swelling involving different anatomic sites. The mandible was the most frequently affected site, with no preference for the anterior or the posterior region. The involvement of the maxilla has been reported in only one case.

In our case, adjacent teeth with resorbed roots were removed simultaneously with enucleation of the lesion. Although enucleation and excision appeared to cure the hybrid lesion, long-term follow-up data and additional cases are still needed to clarify the clinical significance of these lesions.

Rare conditions that can affect the oral maxillofacial region and usually occur in adults as an asymptomatic swelling with only 26 reported cases.

The common symptom of these cases was a painful jaw swelling involving different anatomic sites. The mandible was the most frequently affected site, with no preference for the anterior or the posterior region. Involvement of the maxilla has been reported in only one case.

There is no effective treatment to prevent or delay DM1. In genetic counseling, it is important to be aware of the following factors: DM1 is a disorder affecting the whole family, in particular partners and close relatives in addition to the affected person.[7]

Considering the frequency of hybrid tumors in the general population, it is difficult to establish if the association between DM1 and hybrid tumor is casual or part of the multisystem involvement with a complex pathogenic mechanism involving a molecular dysregulation of cell proliferation that DM1 disease implies.[8]

Conclusion

We aim to highlight two key points from this report. The first is we can hypothesize that DM1 may predispose to hybrid odontogenic lesions compared with the general population.

Second, further reports are needed to deeply investigate and better evaluate such coexistence in a larger sample of DM1 patients.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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