Case Report

Andersen–Tawil Syndrome and Hypothyroidism: A Case Report with an Unusual Association

Abstract

Andersen–Tawil syndrome (ATS) is an autosomal dominant disorder, characterized by the triad of muscular paralysis, skeletal, and craniofacial anomalies and prolonged QT interval on echocardiogram with a tendency toward malignant ventricular arrhythmia. Although the patient may express one or two of the three components of triad, hypothyroidism is an endocrine disorder resulting in the delayed eruption of teeth, defective mineralization of bone and teeth, and speech and hearing deformity. Here, we report a case of ATS with hypothyroidism. To the best of authors' knowledge, no such association has been reported in the literature.

Keywords: *Andersen–Tawil syndrome, clinodactyly, enamel hypoplasia, hemivertebrae, hypothyroidism*

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Introduction

Andersen–Tawil syndrome (ATS) is an autosomal dominant disorder, characterized by the triad of muscular paralysis, skeletal, and craniofacial anomalies and prolonged QT interval with a tendency toward malignant ventricular arrhythmia. However, affected individual may express one or two of the three components.[1,2] Skeletal and craniofacial anomalies include short stature, clinodactyly of little finger, syndactyly, scoliosis, small mandible, low set ears, and widely spaced eyes; affected individuals present with cardiac symptoms (palpitation or syncope) and muscle weakness episode spontaneously followed by long rest in the first or second decade of life. Sufficient thyroid hormones are needed for proper development and growth of child. Insufficient thyroid hormones levels results in the inadequate mineralization of hard tissues including bones and teeth. Other features include delayed eruption of teeth,[3,4] speech deficit, and hearing deficit.[5,6] Here, we report a case of ATS with hypothyroidism. To the best of our knowledge, no such association has been reported in literature till date, and hence the importance of reporting this case, which presents an association of ATS and hypothyroidism. We also emphasize on the importance of checking thyroid functioning in ATS to rule out any thyroid insufficiency.

Case Report

A 14-year-old male patient reported with a chief complaint of decayed teeth. The patient gave a history of occasional pain in the decayed teeth. His past investigation records revealed that the patient was diagnosed as a case of dysmorphism with long thoracic scoliosis and hypothyroidism. The patient was undergoing treatment for hypothyroidism – tablet thyroxin 50 mg one tab per day before breakfast.

Natal events at birth reveal a history of low birth weight with normal delivery; postnatal events reveal history of failure to thrive, speech deficit, and hearing deficit since birth, started walking at 2 years, and history of 2–3 words speaking that was unclear at the age of 8 years. History of similar short stature in his paternal aunt family and history of death of father by cardiac arrest 2 years ago have been reported.

The patient had a short stature, thin built, 125 cm height, and 15 kg weight. These features did not match the features of a child with chronological age of 14 years. The patient presented with long nose and low set of ears [Figure 1a]. The right and left shoulders were at different levels. Fullness was seen on the left lateral part of abdomen, and umbilicus was normal [Figure 1b].

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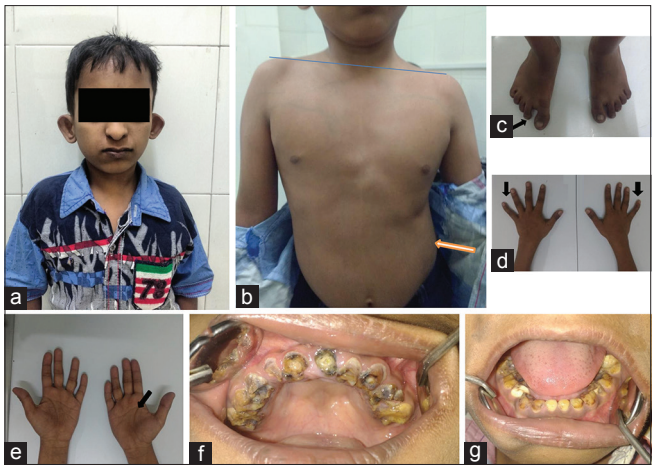

Figure 1: (a) Low sets of ears and long nose. (b) Low level of left shoulder and fullness on the left lateral side of the abdomen (shown by an arrow). (c) Widely spaced right thumb and index finger of the right foot in comparison to the left foot (shown by an arrow). (d) Clinodactyly (shows by an arrow). (e) Simian crease on palmer surface (shown by an arrow). (f) U-shaped maxillary arch with shallow vault, severely attrited and carious teeth. (g) U-shaped mandibular arch, severely attrited and carious teeth with normal tongue

Clinodactyly of distal phalanx of little fingers bilaterally was seen on hand examination [Figure 1d], and simian crease was present on palmer surface of hands [Figure 1e]. Examination of the feet revealed elongated thumb and widely spaced thumb and index finger of the right foot in comparisons to the left foot [Figure 1c].

Intraoral examination revealed U-shaped maxillary and mandibular arch with multiple carious teeth and severe attrition with yellow discoloration of the teeth; the palatal vault was shallow [Figure 1f and g]. Maxillary and mandibular canines and mandibular first premolars were missing bilaterally. Based on clinical examination, provisional diagnosis of dysmorphism with hypothyroidism was established.

Chest X-ray and orthopantogram (OPG) were advised. Chest X-ray revealed S-shaped curvature in the thoracic vertebrae region and two hemivertebrae in the thoracic region [arrows in Figure 2]. Acromion process of the left scapula was underdeveloped in comparison to the right scapula [Figure 2]. Digital OPG revealed over retained deciduous mandibular left and right first molar. Impacted teeth noted were maxillary right canine, mandibular left and right canine, and mandibular left and right first premolar. The crown of the maxillary first molar, impacted mandibular left and right canine appeared malformed. Radiographically, the crown of permanent right mandibular second premolar presented with similar radiodensities of the enamel and dentin [Figure 3a] whereas intraoral examination clearly showed whitish enamel of the erupted permanent right mandibular second premolar [Figure 1g]. This finding confirmed the hypoplastic variety of enamel hypoplasia. Radio Visio Graphy (RVG) revealed malformed crowns of impacted mandibular canine [Figure 3b].

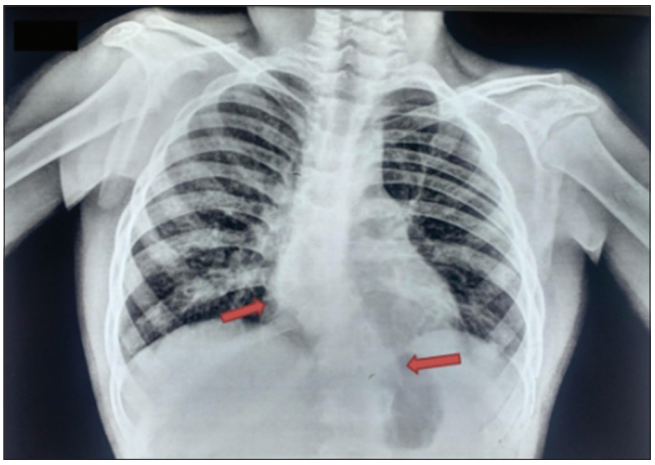

Figure 2: Chest X-ray shows S-shaped curvature in vertebral column in thoracic region with hemivertebrae (shown by an arrow) and the underdeveloped acromion process of the scapula on the left side

For impacted permanent maxillary right canine, cone-beam computed tomography was advised for better vision in different sections. In axial [Figure 4a], coronal [Figure 4b], and sagittal section [Figure 4c], crown was malformed; these features are suggestive of regional odontodysplasia in relation to 13.

ECG and echocardiography findings were normal. On the basis of history of muscle weakness and death of father due to cardiac arrest, decreased speech and hearing ability, and clinical and radiographic dentoskeletal abnormality confirmed the diagnosis of ATS with hypothyroidism.

The patient was advised for full mouth rehabilitation and T3, T4, thyroid-stimulating hormone level. These hormones levels were normal because the patient was on thyroxin 50 mg.

Discussion

ATS is a hereditary disorder comprised of a triad of episodic muscle weakness, skeletal dysmorphism, and cardiac arrhythmias.[7] However, one or two components of ATS may also represent the ATS phenotype.[2] In our case report, the patient had skeletal dysmorphic feature and history of muscle weakness. ATS is an autosomal dominant disorder. In our case report, the patient's father died due to cardiac arrest. However, records of father's previous medical history were not available. Hence, it may be presumed that his father had cardiac arrhythmia that is one of the components of triad of the ATS.

Thyroid hormone is an important element for mineralization of orofacial tissues including teeth.

Thyroid dysfunction during childhood reported a disturbance in the eruption timing of teeth, mineralization of teeth and maxillary and mandibular bones, increased risk of periodontal disease, and morphology of the tongue. Hearing loss associated with congenital hypothyroidism changes the acoustic signals to the brain, results into

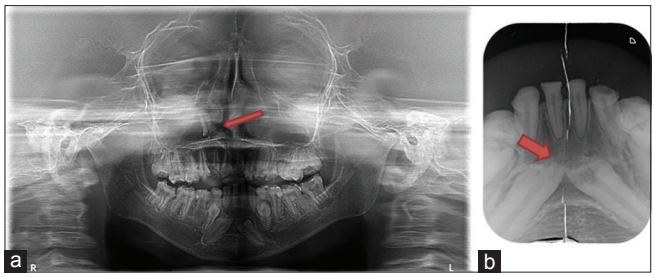

Figure 3: (a) Digital Orthopantogram shows over retained 74 and 84. Impacted 13, 33, 34, 43, and 44. Abnormally formed crown in relation to impacted 13, 26, 33, and 43 suggestive of odontodysplasia. Crown of 13 shows motheaten appearance (shown by an arrow). Generalized loss of enamel suggestive of enamel hypoplasia. (b) Intraoral periapical radiograph of mandibular anterior teeth shows malformed crown of impacted 33 and 43 suggestive of odontodysplasia (shown by an arrow)

difficulty in differentiation between sounds and associated symbols. The effect of mild hearing loss results in delayed speech acquisition, and difficulty in comprehension is reported in literature. In our case report, the patient was earlier diagnosed with hypothyroidism, and also, he had delayed teeth eruption, hypoplastic teeth, and speech and hearing deformity. The case is clinically relevant as ATS syndrome presents with dental anomalies such as impaction, enamel hypoplasia, and regional odontodysplasia. This association of ATS and hypothyroidism is novel; hence, hypothyroidism should be ruled out in every patient presenting with ATS.

Declaration of patient consent

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

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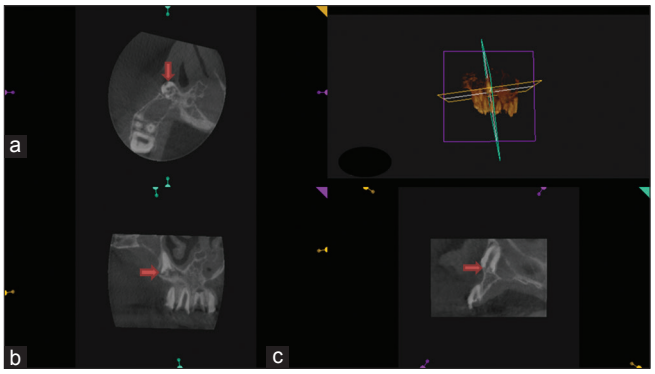

Figure 4: (a) axial section, (b) coronal section, (c) sagittal section shows odontodysplasia in relation to 13 tooth

Conflicts of interest

There are no conflicts of interest.

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