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## Synchronized occurrence of two rare genetic disorders: Amelogenesis imperfect and hereditary sensory autonomic neuropathy type II

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### KEYWORDS

Amelogenesis Imperfecta, Hypomaturation Type, Hereditary Sensory Autonomic Neuropathy (Hsan) Type Ii, Occurrence

### A B S T R A C T

Amelogenesis imperfecta (AI) is a genetic condition that consists of a group of complicated situation which in the absence of any systemic disease shows a series of developmental changes in enamel of teeth. Hereditary sensory autonomic neuropathy (HSAN) is rare genetic disease that involves peripheral nerve system. Both of them are rare disorders and to date there is no reported incidence of the two diseases. The patients were 2 brothers one 27 years old and other 37 years old that admitted for dental treatments to oral medicine clinic. According to clinical findings and consult with neurologist diagnosis of AI and HSAN II for them were confirmed. This is the first reported case of simultaneous occurrence of these two diseases in two members of a family. Additional studies are needed to investigate about genetically natures of these disorders.

## Introduction

Amelogenesis imperfecta (AI) is a complicated inherited dental condition affecting the structure and clinical appearance of the enamel of all teeth in a more or less equal manner in the absence of any systemic disease (Welbury, 2009). AI is a heterogeneous group of hereditary disorders of enamel formation it may be AD, AR, sex-linked or sporadic (Crawford *et al.*,

2007). At least 14 different inheritance types for amelogenesis imperfect and different clinical features of it are recognized (Welbury, 2009). Overall all two dental systems (permanent and deciduous) have generalized involvement (Gadhia *et al.*, 2012).

### **Clinical and radiographic features**

Amelogenesis imperfect may be inherited as autosomal dominant, autosomal recessive or x-linked. There are different reports about the prevalence of AI different studies have explained the wide variation in prevalence rates were due to the different populations studied and the genetic differences for these populations. Its approximate incidence is between 1/718 and 1/14000 (Crawford *et al.*, 2007). Overall all two dental systems (permanent and deciduous) have generalized involvement (Crawford *et al.*, 2007).

Amelogenesis imperfect is classified to hypoplastic, hypomature and hypocalcific on the basis of its clinical features and enamel deficiency state (Aldred *et al.*, 2003).

In hypoplastic type there is several changes consists of incomplete precipitation of enamel matrix. All of matrix mineralizes efficiently and in radiographic features it is distinct from dentine. This type is characterized by small crowns with thin enamel or enamel of normal thickness usually associated with pits and grooves, due to deficiencies in the amount of enamel. In generalized pattern pinpoint cavities to pinhead cavities are scattered on the surface of teeth and these cavities are not related to environmental damage pattern (Aldred *et al.*, 2002). In hypomature type enamel matrix precipitates normal with inclination to separation from its lower dentin.

Enamel normally begins to mineralization but there is a deficit in enamel maturation crystal pattern. Affected teeth have normal shape but affects mottled color change or opaque white brown-yellow color. In hypocalcified type enamel matrix precipitates normally but there is not any mineralization. In growth teeth have normal

shape but the enamel is too soft and it will lose easily (Aldred *et al.*, 2002). Occasionally, AI occurs with other features as part of a syndrome in, for example, Heimler syndrome (Tischkowitz *et al.*, 1999), Amelo onycho hypohidrotic syndrome, Kohlschutter syndrome (Christodoulou *et al.*, 1988), tricho-dento-osseous syndrome, AI with taurodontism, oculo-dento-osseous dysplasia (Crawford *et al.*, 2007), epidermolysis bullosa hereditaria, CRD (Cone Rod dystrophy), AI and nephrocalcinosis syndrome (Kirzioglu *et al.*, 2009). Regarding to the relationship between disease mentioned above and amelogenesis, amelogenesis and with consideration that the amelogenesis has occurred in two family members who had already affected with coincidence rare sensory abnormality we decided to present this case for future evaluations for finding relationship between these two disorders.

Hereditary sensory autonomic neuropathy (HSAN) is rare genetic disease that involves peripheral nerve system (Amato and Dumitru, 2002). With attention to its age of occurrence and hereditary type and clinical features, patients are classified to 5 categories. For importance of this issue and simplifying this subject we describe only type 2 of HSN. In type II HSN the inheritance is autosomal recessive and the age of affection is from newborn period to early childhood and the symptoms of this category of early patients are lack of sensation to environmental stimulates completely. The purpose of the present study is to describe a patient showing the typical features of AI and HSAN II syndrome simultaneously.

### **Case presentation**

The patients were 2 brothers one 27 years old and other 37 years old that admitted for

dental treatments to oral medicine department. In history, we found their sensation problems as partial lack of sensation to temperature and in physical examination there was a lot of severe ulceration on their fingers and toes. These symptoms began on childhood and progressed. In his familial history his parents were cousins and he had sibling first and last child of this family were known case of HSAN II. There were too many similarities between first and last child of his family in their mien.

The first child was boy and affected progressive and more severe disease because of his large chronic ulceration he underwent toes and some part of his foot amputation.

In the presented cases, the fingers were damaged and radiographic findings revealed chronic osteomyelitis, and there was shortens of fingers due to several damages (Fig.1).

Evaluation of IQ studies in these two brothers showed that their intelligence is normal they have dermal hyper sensitivity, in their feet and hands to high temperature, moisture for example after shower. These patients didn't complain of autonomic nervous system dysfunction (Gut and bladder function) but they complained of their hand excessive perspiration. After consulting with neurologist the patient admitted to electro diagnosis ward for EMG/NCV. Because of his type of inheritance and the result of electro diagnostic tools, type 2 HSAN was considered by neurologist.

Dental examination showed opaque yellowish to brown color changes, in the 1/3 middle and incise region of upper maxillary teeth. In physical examination of the region with these changes there was not any difference in consistency with other regions

enamel. General marginal gingivitis in upper and lower maxilla was seen in the absence of any considerable plaque .with consideration to this situation amelogenesis imperfect hypomature type, dental fluorosis and color Changes due to tetracycline consumption were considered, in our differential diagnosis but with farther evaluation of clinical findings, familial history, measurement of regional water fluoride and not involvement of other residents on that region, amelogenesis imperfect hypo mature type was considered (Fig. 2).

### **Discussion**

AI consists of a group of complicated situation witch in the absence of any systemic abnormality. There are some developmental changes in teeth enamel. This disease is inherited as autosomal dominant, autosomal recessive or X linked inheritance (Welbury, 2009). Two dental systems (permanent and deciduous) are involved generally (Crawford *et al.*, 2007). AI is classified based on its clinical findings and type of deficiency to hypoplastic type, hypomature type and hypocalcific type (Gadhia *et al.*, 2012).

HSAN type 2 results from mutation in *HSN2* gene on q1312 chromosome (Dyck, 1993). All of clinical findings of this disease begin from childhood with loss of sensation to all stimulators, feet degeneration, erosive arthritis, osteomyelitis, and auto amputation of toes. In these patients according to clinical findings like sensory abnormalities such as relative parasthesia to pain and temperature from childhood, fingers osteomyelitis and foot fingers amputation they are referred to neurologist for confirmatory examination like EMG/NCV and with attention to their familial history diagnosis of HSN type II was confirmed.

In reported cases dental features like generalized mottled and white – brown – yellowish color changes in teeth that looks like normally and presence of enamel that is softer than normal and presences of these common signs between two individuals in one family leads to diagnosis of amelogenesis imperfect hypo mature type in this patient. Clinical appearance of this case

was like fluorosis but according to dental involvement from the birth in these two brothers and lack of involvement in other persons in that region and normal fluoride concentration in regional water the diagnosis of fluorosis is not acceptable.

**Figure.1** Hypo maturation amelogenesis imperfect. The patient exhibiting diffuse yellow- white dentition. Generalized marginal gingivitis is seen



**Figure.2** Same patient showing Nail dystrophy .Note the fingers are shorted as a result of osteomyelitis



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