Christ – Siemens – Touraine Syndrome: An unusual Presentation of the ectodermal disorder.

Author: Dr.B.Senthilkumar, M.D.S., D.H.M., Asst. Professor, Dept.of Oral Medicine, Diagnosis & Radiology, K.S.R.Institute of Dental Sciences, Trichengode, Mobile No.94441 39886, E-mail: balu.senthilkumar@yahoo.in

Abstract:
Ectodermal dysplasia exhibits a classic triad of hypohidrosis, hypotrichosis & hypodontia. Hair defects include longitudinal grooving, hair shaft torsion, & cuticle ruffling. Eccrine defects include sweat glands absent (or) sparse & rudimentary. Dental defects include Abnormal Morphogenesis (or) absence of teeth. In most instances dentists are the first to recognize patients with ectodermal dysplasia as they report primarily with a complaint of missing teeth. The most common type is hypohidrotic ectodermal dysplasia (Christ – Siemens – Touraine syndrome) A deepened knowledge about the disease, its clinical presentation & diagnosis, aids in treating patients systemic conditions, Masticatory function & esthetic appearance.

Keywords: Ectodermal dysplasia (ED), Christ – Siemens Touraine syndrome, hypohidrosis, hypotrichosis & hypodontia.

Introduction:
Ectodermal dysplasia is a congential disorder of one (or) more ectodermal structures. Thurman published the first report of a patient with ED in 1848, but the term ectodermal dysplasia was later coined by weec in 1929. EDs are rare with their incidence estimated as 1 in 10,000 to 1 in 1,00,000 births. The tissues primarily involved are the skin, hair, nails, eccrine glands & teeth. To date, more than 192 distinct disorders have been described. The most common is X-linked recessive hypohidrotic ectodermal dysplasia also know as Christ- Siemens - Touraine syndrome, It affects Males & inherited through female carriers. It is characterized by a triad of signs comprising of sparse hair (atrichosis (or) hypotrichosis), inability to sweat due to lack of sweat glands (anhidrosis (or) hypohidrosis) & abnormal (or) missing teeth (anodontia (or) hypodontia)

Here we present a case of hypohidrotic ectodermal dysplasia with hypodontia, hypotrichosis & hypohidrosis in a male patient of age 23 years.
Case report

A male patient aged 23 years visited the department of Oral Medicine & Radiology with a Chief complaint of Missing teeth in the upper & Lower, front & back regions of the Jaw since childhood & patient had difficulty in Mastication he feels ill of his esthetic appearance. Pt’s Medical history reveals that he frequently visited dermatologists for fever & irritation in the skin.

Family history reveals that he is the only son, his father & mother is apparently normal on examination Patient has sparseness of scalp & body hair, dry skin, frontal bossing, depressed nasal bridge, & protuberant lips.

On intra oral examination he had multiple missing teeth; the teeth were conical in shape & some with altered morphology. The Mandibular ridge was completely edentulous & appeared flat. The over all vertical dimension of face was also reduced.

* Radiographs were taken, Orthopantomogram revealed, no impacted teeth & the present teeth had altered coronal morphology & the roots were short.
Lateral Cephalogram reveals, decreased lower facial height, retruded maxilla, the nasal, alar & mouth width was smaller. Based on history, clinical examination & Radiographic findings, the diagnosis of ectodermal dysplasia was made.

Discussion:

Ectodermal dysplasias are a heterogeneous group of disorders characterized by developmental dystrophy of ectodermal structures, such as hypohidrosis, hypotrichosis, onychodysplasia & hypodontia (or) anodontia \(^1,2\).

This X-linked recessive disorder affects males and is inherited through female carriers the gene responsible for this syndrome might be either Mutated (or) deleted; some of them are Xq 12, q 13.1, PVRL, GJB6.

The recent cloning of gene had led to identification of transmembrane protein ectodysplasin (TNF family ligand & edar (TNF receptor). This TNF ligand & receptor have a developmental regulatory role & tightly associated with epithelial – Mesenchymal interactions and in signaling pathways that regulate ectodermal appendages formation and organogenesis during initiation of development \(^2,3\).
Clinically ectodermal dysplasia may be divided into two broad categories one is hypohidrotic form, which is X-linked and characterized by the classical triad of hypodontia, hypotrichosis, & hypohidrosis, which is termed as Christ- siemens–Touraine syndrome. The other category is that of the hidrotic form described by clouston, which usually spares the sweat glands & can affect teeth, nails & hairs. The Presentation of facial deformity includes frontal bossing, sunken cheeks, saddle nose, thick and everted lips, wrinkled hyperpigmented periorbital skin and large low set ears, Dental Manifestations include conical (or) pegged, shaped teeth, hypodontia (or) complete anodontia & delayed eruption of permanent teeth. Most patients have fine, sparse, lustreless hair; onchodystropy may occur but is not common 4.

Extensive scaling of the skin and unexplained pyrexia secondary to anhidrosis may occur in neonatal period. The development of chronic eczematous dermatitis is common; other common signs are short stature, eye abnormalities, decreased trearing & photophobia. Various features of ectodermal dysplasia described by various authors were found to mimic our patients. Thus a patient with ectodermal dysplasia has various features of hypotrichosis, hypohidrosis & hypodontia. So early detection is valuable, as it increases the possibility to use growth adapted measures in Multidisciplinary treatment planning 5,6.

Conclusion:
Ectodermal dysplasia has emotional consequences for affected individuals at early ages. Thus early diagnosis by a dentist can be of great importance to restore mastication, speech & esthetics.

References:

Co-authors:

Nazarji Mahabob. M.D.S Asst. Professor,
Dept. of Oral Medicine, Diagnosis & Radiology,
K.S.R.Institute of Dental Sciences,
Tiruchengode.

Case report:

Fig:1  
fig:2
Fig:1 Short stature of the patient
Fig:2 Sparseness of scalp & body hair, dry skin, frontal bossing, depressed nasal bridge, & protuberant lips.
Fig:3 Hypodontia and microdontia in maxilla
Fig:4 Mandibular ridge was completely edentulous & appeared flat
Fig:5 Frontal bossing, depressed nasal bridge, & protuberant lips.

Fig:6 Patient has sparseness of scalp & body hair

Fig:7 Orthopantomogram revealed, no impacted teeth & the present teeth had altered coronal morphology & the roots were short.

Fig:8 Lateral Cephalogram reveals, decreased lower facial height, retruded maxilla, the nasal, alar & mouth width were smaller.