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Christ-Siemens -Touraine-Syndrome : Anhidrotic Ectodermal Dysplasias Presenting in ENT as Atrophic Rhinitis With Nasal Myiasis-3 Case Reports

KEYWORDS: epistaxis,malaena,familial,telangiectasias,Osler Weber Rendu Syndrome.

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ABSTRACT *Ectodermal Dysplasia represents a complex group of pathologies containing over 170 clinical features with an incidence of 7 out of 1, 00,000 live born children. CASE 1: 3 yr, male presented with foul smelling discharge and falling of crusts from nose from 2 and half years. Case 2: 4 yr, male child with foul smelling discharge and falling of crusts from nose since 2 yrs. He also had a history of Delayed development of teeth and breathing difficulties since his first month of life. History of upper air pathway infections and recurrent otitis since he was 2 months old. CASE 3: 13 months old female child presented with complains of nasal obstruction and presence of crusts, mucopurulent rhinorrhea, nasal fetidness and breathing difficulties since her first month of life. All cases treated with regular alkaline nasal douches and glucose glycerine packs. The ears were regularly suction cleaned and ofloxacin-dexamethasone-clotrimazole ear drops was applied and adequate dietary rehabilitation with vitamin and mineral supplementation. Antibiotics in the form of ampicillin and cloxacillin for two weeks were administered. Atrophic rhinitis is a type of disease associated with Genetic and molecular alterations of ectodermal dysplasia. Anhidrotic dysplasias can present as Atrophic Rhinitis in ENT. The full-blown presentation of CST syndrome in a female patient is rare.*

INTRODUCTION:
Ectodermal Dysplasia represents a complex group of pathologies containing over 170 clinical features with an incidence of 7 out of 1, 00,000 live born children. It holds clinical alteration which affects structures coming especially from ectoderm such as skin, hair, teeth, nails and sweat glands. Atrophic Rhinitis is chronic inflammatory disease associated with Ectodermal Dysplasia. Atrophic Rhinitis is one the situations related to Ectodermal Dysplasia. It mainly affects individuals at late childhood age and young adults. Atrophic Rhinitis is a chronic inflammatory disease of the nose, producing crusts due to imperfect movement of the nose hair. Besides being related to Ectodermal Dysplasia, it can be found at granulomatous diseases such as Tuberculosis and Hansen´s disease

Case reports:
CASE 1: 3 yr, male presented with foul smelling discharge and falling of crusts from nose from 2 and half years. Bleeding from nostrils from one day. Falling of worms from nose since one day. He had history of Delayed development of teeth, and also had history of increased intake of water. Underwent full term normal vaginal delivery On examination Dry Skin with scaling , Fissured Foot, Diminished/sparse body hairs, Brittle and hyperconvexed nails were present, Upper&lower limb shows palmoplantar crease – hyperkeratosis. Face presents maxillary hypoplasia, inferior lip protusion, prominent front, periorbitary hyperpigmentation and low implanted ears Ear examination showed External auditory canal shows Impacted wax. Nose Examination revealed Live Maggots with Foul smelling discharge and crusts, Nasal mucosa was dry. Oral cavity examination showed Protuberant lips and pegged lower lateral incisors.

Diagnostic nasal endoscopy showed Greenish nasal crusts with thick purulent discharge and Atrophied nasal turbinate's and CT NOSE AND PNS revealed B/L maxillary sinusitis, Nasal biopsy showed atrophied pseudostratified columnar epithelium with sub epithelial inflammatory infiltrate, consisting of lymphocytes, plasma cells, histiocytes,eosinophils, fragment of bone and fibrocollagenous tissue and Skin biopsy was done showed keratinized stratified squamous epithelium and dermis. There is a total absence of hair follicles, sebaceous glands and eccrine glands.

Case 2: 4 yr, male child with foul smelling discharge and falling of crusts from nose since 2 yrs. He also had a history of Delayed development of teeth and breathing difficulties since his first month of life. History of upper air pathway infections and recurrent otitis since he was 2 months old. When he was four, myopia was diagnosed, presenting hypoacusis, occasional dysphonic, photophobia and a delay on psychomotor development.

In the physical exam, Dry Skin with scaling and Hypopigmented patches over chest was present. Hair becomes thin, dry and hypochromic. There is presence of hypodontia and nails are hyperconvexed. Nose is saddled-shape and face presents maxillary hypoplasia, inferior lip protrusion. In the rhinoscopic examination, presence of crusts and mucopurulent rhinorrhea with Atrophied inferior turbinate were observed. In the otoscopy there is presence of central perforation on the right tympanic membrane and retraction on the left one.

The audiometric exam showed bilateral conductive hearing loss and nasal endoscopy showed per-Eustachian tube area edema and Atrophied nasal turbinate's with plentiful formation of crusts.

Nasal biopsy showed atrophied pseudostratified columnar epithelium with sub epithelial inflammatory infiltrate, consisting of lymphocytes, plasma cells, histiocytes,eosinophils, fragment of bone and fibrocollagenous tissue and Skin biopsy was done showed keratinized stratified squamous epithelium and dermis. There is a total absence of hair follicles, sebaceous glands and eccrine glands.

CASE 3: 13 months old female child presented with complains of nasal obstruction and presence of crusts, mucopurulent rhinorrhea, nasal fetidness and breathing difficulties since her first month of life. She had suffered 2 episodes of epistaxis so far. History of Second degree consanguineous marriage in her parents. Mother had a premature labor by Caesarian (32 to 33 weeks old), and the baby was kept in neonatal ICU for 22 days. History of recurrent upper air infections and recurrent otitis since she was1month years old with developmental delay.

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In the physical exam, presence of crusts and mucopurulent rhinorrhea with nasal obstruction at rhinoscopy were observed. On examination of the right ear, live maggots with greenish yellow foul smelling discharge were found in the external auditory canal.

Skin becomes thin, dry and scaled with hypotrichosis and nipple absence. Hair becomes thin, dry and hypochromic. There is presence of hypodontia and nails are hyperconvexed. Nose is sadlled-shape and face presents maxillary hypoplasia, inferior lip protusion, prominent front, periorbitary hyperpigmentation and low implanted ears.

All cases treated with regular alkaline nasal douches and glucose glycerine packs. The ears were regularly suction cleaned and ofloxacin-dexamethasone-clotrimazole ear drops was applied and adequate dietary rehabilitation with vitamin and mineral supplementation. Antibiotics in the form of ampicillin and cloxacillin for two weeks were administered.

DISCUSSION:

Ectodermal dysplasias compromise large, heterogeneous group of inherited disorders that are defined by primary defects in development of 2 or more tissues developed from ectoderm (Skin, hair, nail, eccrine glands, and teeth). Most common X-linked Ectodermal Dysplasia (christ-siemens-touraine syndrome) Affects males inherited through females. Triad of sparse hair (atrichosis or hypotrichosis), anodontia/ hypodontia, anhidrosis / hypohidrosis. Incidence in male is 1 in 100,000 births. Most patients have normal life expectancy.

Around 94% of the cases is due to alterations on ED1 gene (2), on Xq12-q13.1 (13), which was isolate by Kere et al. in 1996 (1,14). About 53 different alterations on ED1 gene has been described so far (13). Such gene is responsible for coding 2 isoforms of ectodysplasin, a transmembrane protein of collagen which is similar to proteins that join Tumor Necrosis Factor. The alteration on homologous domain to the TNF ligand ones shows that this protein area is fundamental to its functioning.

The two isoforms of Ectodysplasin are ED1-A1 and EDA-A2, which are respectively connected to the EDAR and XEDAR receptors. When such connections occur, the cascades of nuclear factor (NF)kB and JNK/c-fos/c-jun are activated. Both are responsible for the activation of signs to Epidermal Growth Factor job, which differ epiderm from its appendages. So, one might conclude that Ectodysplasin works on cell surviving, communication and growth.

Another alteration, besides this one, is the DL gene one, which is responsible for EDAR receptor. This causes recessive or dominant Ectodermal Dysplasia.

The most common manifestation of Hypohidrotic Ectodermal Dysplasia is the reduction or complete absence of eccrine, sweat and lacrimal glands. However, apocrine glands are in normal condition . One of the consequences of gland alteration is the impossibility to sweat, what happens due to body temperature, hyperthermia crisis and fever convulsions . Skin, when usually hypo pigmented, becomes thin and dehydrated, scaled (6), and it can suffer from atopic dermatitis and xeroderma

The alteration on the lacrimal glands causes secretion in small amount and dacryocystitis .

Regarding dental formation, hypodontia and adontia are found. Teeth are small and conical shaped.

Hypotrichosis is frequent and it can many times be associated with baldness. Hair is thin, dry and hypochromic as well as the other hair of the body. Among nail abnormalities, it can be found hyperconvexed, hypertrophic dystrophic, keratinized nails and even the absence of them.

Facial abnormalities show tipical appearances, such as saddled nose prominent front, low implanted pointed ears, inferior lip protrusion periorbitary hyperpigmentation.

The other alterations found are hypoplasia of mammary glands, visual disfunction, premature birth and mental retardation.

Most of ENT manifestations are due to secretion alterations from mucosal glands, which might lead to recurrent infections. It is constant the occurrence of infections of superior air pathways and repetition otitis - medium, external and eczematous one - many times followed by sensorineural hypoacusis. Patient can present chronic conditions such as pharyngitis, laryngitis, dysphonia and odynophagy as well as atrophic rhinitis followed by epistaxis and catarrh.

Atrophic rhinitis is a chronic inflammation of the nose characterized by progressive atrophy of nasal mucosa, turbinate's, nerves & glands. It is also associated with ectodermal dysplasia. TRIAD OF ATROPHIC RHINITIS Crust formation, Atrophy of nasal structure and Foul smell. Saddled nose is related to improper blood supply during nasal and septal bone growth. This occurs due to severe inflammatory reaction in Atrophic Rhinitis.

Various tests have been employed for the detection and confirmation of HED. Non-invasive trichogram and sweat testing results can support the diagnosis of HED, but they are not sensitive or highly specific. Horizontally sectioned 4-mm punch biopsy specimens of the scalp or palms that lack eccrine structures are diagnostic of HED. Despite recent advances in the genetic basis of this disorder, the diagnosis is still established clinically in majority of patients since genetic analysis is not routinely available.

Management of patients with this disorder is a challenge and will require multidisciplinary approach that involves a general physician or pediatrician, geneticist, dermatologist, ophthalmologist oral and maxillofacial surgeon, prosthodontist, orthopedician and of course the otolaryngologist. Atrophic rhinitis is usually treated conservatively with nasal douches. Cosmetic appearance can be improved with rhinoplasty and otoplasty. Cholesteatoma in the ear requires a canal wall down mastoidectomy and hearing reconstruction. The patient should avoid hot climates and febrile episodes should be treated by cooling the body. Genetic counselling should be offered and attention should be paid to the emotional needs of the patient.

After describing signs and symptoms found in ectodermal dysplasia and atrophic rhinitis. It is possible to observe that the current patients have many of them, what characterizes a peculiar case as the disease appears in different ways in different people. But in one of our cases Anhidrotic Ectodermal Dysplasias presented in female child at very early age which is rare entity and treatment is always a symptomatic.

CONCLUSION:

Atrophic rhinitis is a type of disease associated with Genetic and molecular alterations of ectodermal dysplasia. Anhidrotic dysplasias can present as Atrophic Rhinitis in ENT. The full-blown presentation of CST syndrome in a female patient is rare. The diagnosis of HED could be missed due to its rarity and lack of clinical awareness. High index of suspicion required in these cases. Multidepartment treatment essential. Biopsy of skin and nose crucial. Early diagnosis and timely advice, counselling and treatment of this uncommon condition could subvert the complications and reduce the morbidity in these high-risk patients. The otolaryngologist needs to consider HED in the differential diagnosis of atrophic rhinitis as nasal myiasis.

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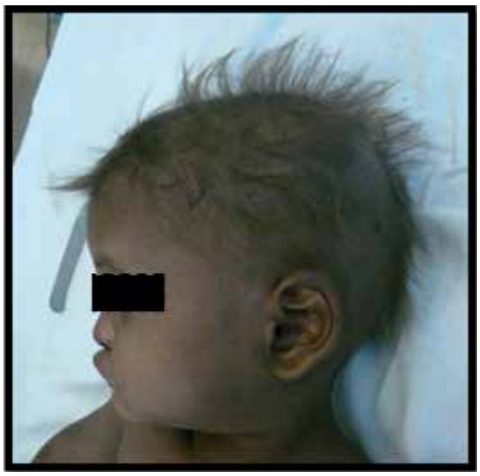

Fig 1: case 1

Fig 2: case 2

Fig3: case 3(female child).

Fig4: Teeth-peg shaped upper central incisor.

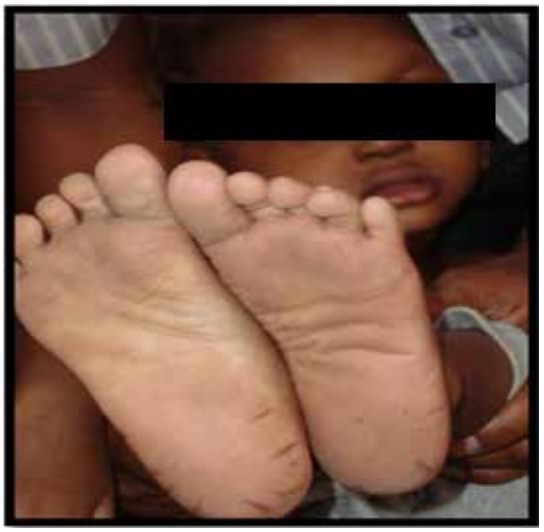

Fig 5: plantar crease- hyperkeratosis.

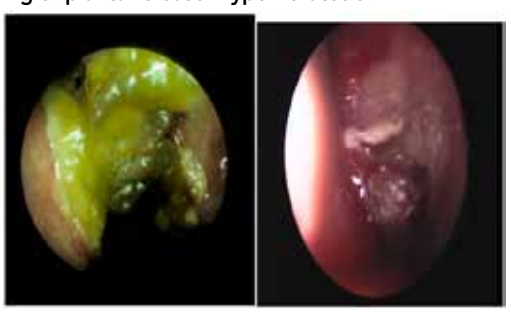

Fig 6: nasal endoscopic pics.

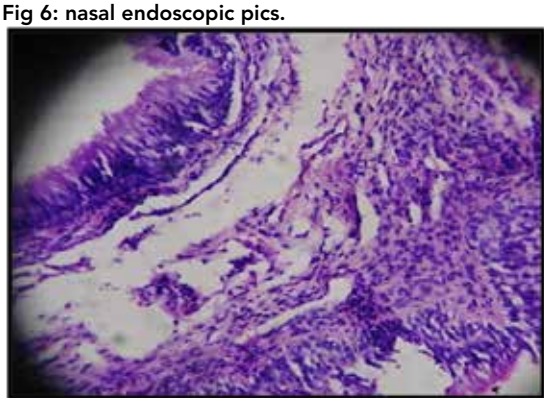

Fig 7: Nasal biopsy- atrophied pseudostratified columnar epithelium with subepithelial inflammatory infiltrate, consisting of lymphocytes, plasma cells, histiocytes, eosinophils, fragment of bone and fibrocollagenous tissue.

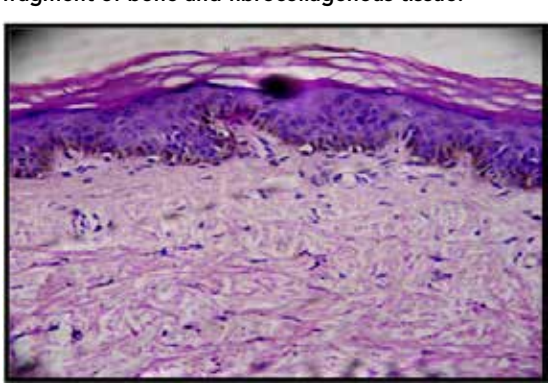

Fig 8: Fig. 3 Pictomicrograph of the skin biopsy showing atrophic eccrine glands [H&E40].

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