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Challenges Confront Maxillofacial Surgeons in Management of the Oral Manifestation of Infantile Systemic Hyalinosis: (A Case Report)

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Abstract

Background: Infantile Systemic Hyalinosis (ISH) (which also known as Inherited Systemic Hyalinosis) is an uncommon, progressive and fatal genetic disorder that usually affects the newborn or infants. It is an autosomal recessive syndrome of unknown etiology, caused by mutations in the anthrax toxin receptor 2 gene - ANTXR2. It is characterized by hyaline deposits in the papillary dermis and other tissues

Case Report: The phenotype characteristics of infantile systemic hyalinosis (ISH) in a two-year-old boy were present. The characteristics of flattered occiput, limited limb movements and articular abnormalities of elbows and knees. Dental findings showed excessive gingival hypertrophy multiple nodules in mucosa completely covering maxillary and mandibular teeth

Plan Of Treatment: The gingival hypertrophy was planned for surgically treated by gingivectomy and excision of nodules under general anesthesia.

Follow-Up: The patient showed a full constellation of clinical manifestations of the disease. although the surgical intervention was not able to be performed due to the unstable medical condition, oral hygiene improvement was maintained.

Conclusions: Surgical treatment of the gingival hypertrophy is the treatment of choice so the patient can perform normal feeding.

Keywords: Infantile systemic hyalinosis, Oral manifestation, Gingival hypertrophy, Surgical intervention

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Introduction

Infantile systemic hyalinosis is a rare hereditary autosomal recessive disease, usually presents at birth or within the first few months of life[1]. the clinical manifestations include; diffusely thickened and inflexible skin, papular skin lesions, hyperpigmentation over the metacarpophalangeal joints of the hands and malleoli, gingival hyperplasia, perianal nodules, limitation of joint motility, osteoporosis of bones, bone fractures, short stature, persistent diarrhea, and failure to thrive[2]. the cause of this disease is the deposition of amorphous hyaline material which is very similar to collagen VI within various tissues; like skin, gastrointestinal tract, cardiac muscle, skeletal muscles, lymph nodes, spleen, thyroid, and adrenal glands[3]. Till now the management of the disease is not well established and survival beyond 3 years of life is unfortunately rare[4].

However, if the patient survives until early childhood, the articular pain decreases in severity[5,7]. It is important to confirm that the central nervous system is not involved; misdiagnosis of muscle hypertonia at the initial stage of the disease usually leads to stubborn rehabilitation which increases the pain[8] Tissue biopsy specimens obtained from ISH patients reveal an accumulation of hyaline deposits. They are usually found in the skin, skeletal and cardiac muscles, lymph nodes suprarenal glands, gastrointestinal tract, thyroid gland and spleen[7,9]

Main concern of the disease is the involvement of the oral soft tissue with hypertrophy of gingiva and mucosal nodules that deprive the patient from normal feeding process[6]. For this reason the interference of the maxillofacial surgeon in such case is important to allow adequate feeding and swallowing The objective was to present the phenotype characteristics of infantile systemic hyalinosis with focus on oral lesions, and obstacles in the therapeutic and surgical management in the reported case.

Case presentation

A two year old boy with infantile systemic hyalinosis was referred to the Department of Oral maxillofacial surgery, Mubarak Al-kabeer Hospital, Bneid Al-Gar dental center, Kuwait. because of gingival hypertrophy that was impairing normal nutrition, tongue lesions and macroglossia. Fig 1

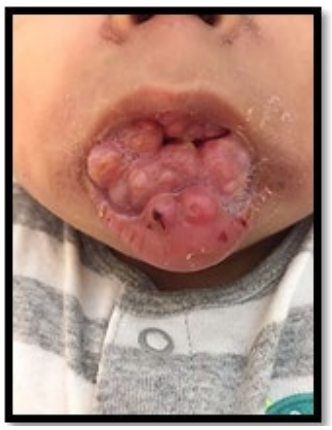

Fig 1: Oral manifestation of ISH

He was first born in family, product of FT LSCS to healthy I st degree consanguineous marriage with no PNP with no antenatal history of oligohydrominious, diminished fetal movements with dysmorphic features, malnutrition, regression of milestones, loss of joint mobility.

Medical history

At the age of 2 months, the patient developed swelling and stiffness of joints, and decreased muscle strength. At 4 months, he developed chronic diarrhea with 4-5 stools per day. In regards to development, he does not roll, sit, stand, reach or grab. He has contractures on all extremities & hands. During initial time on medical floor, patient had extensive workup with multiple disciplines. Due to family's insistence TPN was initially held but patient repeatedly demnstrated that he was unable to meet nutritional needs by feeds. And he was placed and tube feedings attempted but patient did not tolerate. At the age of 1 an EGD/Sigmoidoscopy showed evidence of nodular and edematous mucosa (non ulcerated) in stomach, duodenum and sigmoid. Noted hypertrophic gums in mouth and small oral opening. He was also found to have hypothyroidism and Endocrinology

Physical examination

Patient was diagnosed to have Chronic diarrhea, Developmental regression, Genetic syndrome, Joints contracture of the ankle, foot and interphalangeal joints Fig 2,3. Thickening of the overlying skin malnutrition, Protein losing enteropathy and Perianal irritation

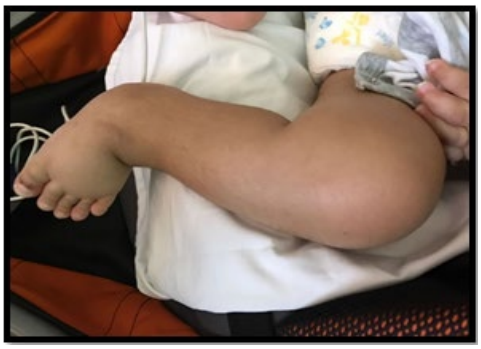

Figure 2: Joint contracture of the ankle and/or foot

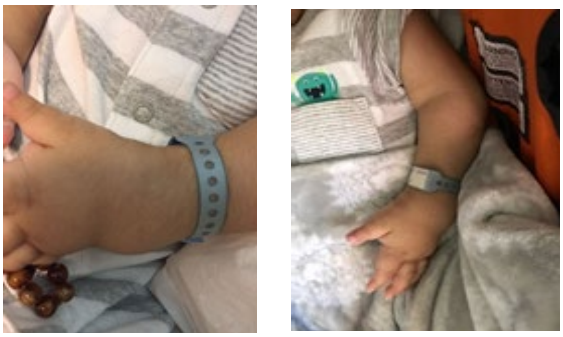

Figure 3: Contractures involving interphalangeal joints with mild thickening of the overlying skin. Skin over his digits was noted to be tight and shiny.

Intraoral examination

The pt showed most of the clinical features in patients with ISH, as gingival hypertrophy, buccal hypertrophy, macroglossia and mucosal thinking, the hypertrophy of mucosa covered the teeth leading to impair the oral hygiene process and gingivitis with teeth decay. The gingival and oral mucosa was firm, smooth and glistening

Laboratory investigation

Tissue biopsy specimens obtained from ISH patients reveal an accumulation of hyaline deposits. They are usually found in the skin, skeletal and cardiac muscles, lymph nodes suprarenal glands, gastrointestinal tract, thyroid gland and spleen

Diagnosis

The differential diagnosis included other congenital diseases of the connective tissue, i.e. Winchester syndrome, systemic fibromatosis, stiff skin syndrome, lipoid proteinosis, and storage diseases including mucopolysacharoses, sphingolipidoses and mucilipidoses

Surgical intervention

A decision was planned to be perform both maxillary and mandibular gingivectomy, although the procedure was postponed because the patient's general condition had deteriorated as the boy had signs of neurological impairment and malnutrition associated with intestinal protein depletion syndrome,

Discussion

The reported case represented an infant showed atypical clinical and biochemical features of ISH with a progressive and disabling joint pain and contractures is a life threating condition. After Landing and Nadorra described the disease in 1986, the ISH was recongnized as a separate entity.10 ISH is considered to be an autosomal recessive disorder that is caused by mutations in the anthrax toxin receptor 2 gene, ANTXR2.2,8 The ANTXR2 gene responsible for encoding the capillary morphogenic protein (CMG)2, a type 1 transmembrane protein[12]. The function of the protein in the skin and connective e tissues, is to bind to type IV collagen and laminins, and is considered to be a factor to basement membrane strength[11]. Abnormal collagen metabolism and deposition causes skin abnormalities in ISH[10] .Deficient extracellular matrix (ECM) synthesis and turnover is directly implicated in the pathogenesis of ISH.12 Skin biopsy in our patient was considered to be an extra unnecessary diagnostic tool since ANTXR2 gene mutation was already identified. Recently, It was reported that there were approximately 25 different pathogenic ANTXR2 gene mutations. Majority of patients with ANTRX2 mutations were homozygous for private mutations with consanguinity being reported in a significant proportion of patients[3] .this gene mutation with its subsequent body and organ interferences eventually lead to premature termination [10]

Conclusion

Despite the benefits of the surgical excision of the mucosal enlargement and gingival enlargement, Debate still exist behind the risk of intervention specially as it will be done under general anesthesia as the pt medical condition may be deteriorated.

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