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

Introduction

Infantile systemic hyalinosis is a rare hereditary autosomal recessive disease, usually presents at birth or within the first few months of life; 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. the cause of this disease is the deposition of amorphous hyaline material which is very similar to collagen VI within various tissues; like skin, gastrointestinaltract, cardiac muscle, skeletal muscles, lymph nodes, spleen, thyroid, and adrenal glands. Till now the management of the disease is not well established and survival beyond 3 years of life is unfortunately rare.

However, if the patient survives until early childhood, the articular pain decreases in severity. 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. 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.

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. 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
He was first born in family, product of FT LSCS to healthy 1st degree consanguineous marriage with no PNP with no antenatal history of oligohydraminious, 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 demonstrated 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.
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

Discussion
The reported case represented an infant showed atypical clinical and biochemical features of ISH. Although the procedure was postponed because the patient's condition had deteriorated as the boy had signs of neurological impairment and malnutrition associated with intestinal protein depletion syndrome.

Diagnosis
The differential diagnosis included other congenital diseases of the connective tissue, i.e. Winchester syndrome, systemic fibromatosis, stiff skin syndrome, lipid proteinosis, and storage diseases including mucopolysaccharoses, 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.

References

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 patient medical condition may be deteriorated.