Case study

A rare case of Nasal polyposis in Hurler syndrome

Abstract:

Mucopolysaccharidosis (MPS) type I is a rare multisystem disorder resulting from the accumulation of breakdown products of glycosaminoglycans in the body tissues. Many patients with this disease undergo ENT surgeries as adenotonsillectomy. Nasal polyposis is a rare occurrence.

The aim of this work is to present a very rare association observed in an 8-years-old patient with Hurler syndrome treated in the ENT Head and Neck Surgery department of Ibn Rochd University Hospital Casablanca, for nasal polyposis and adenoid hypertrophy.

Keywords: Mucopolysaccharidosis (MPS) I, glycosaminoglycan (GAG), Nasal polyps

Introduction

Hurler syndrome, also known as mucopolysaccharidosis type I (MPS I), described by Gertrud Hurler and E. Schindewolf , is a rare disease with 2 to 5 per 100 000 live births for MPS disorders caused by a deficiency of the enzyme alpha-L-iduronidase[1], which is needed to break down a specific type of mucopolysaccharide called dermatan sulfate and leading to buildup and urinary excretion of high levels of glycosaminoglycans (GAGs), specifically dermatan and heparan sulfates. It shows autosomal recessive inheritance[2].

People with Hurler syndrome may be more prone to developing paranasal sinus polyposis due to the accumulation of breakdown products of glycosaminoglycans in the tissues, which can cause inflammation and lead to the growth of polyps[3]. Most frequent presentations are chronic/recurrent respiratory infections (rhinosinusitis), acute (OMA), chronic otitis media with efusion (OME), progressive hearing loss, enlarged tonsils and adenoid glands[4]. In some cases, Hurler syndrome may also cause abnormalities in the bones of the face and nasal passages, which can contribute to the development of polyps although Nasal polyposis is a rare occurrence. Treatment for paranasal sinus polyposis may include medications to reduce inflammation and shrink the polyps, or surgery to remove the polyps. Treatment for Hurler syndrome can help manage the symptoms and reduce the risk of complications, including paranasal sinus polyposis.

We present a patient with Hurler syndrome who developed nasal polyposis and adenoid hypertrophy requiring sinus endoscopic removal.

Case presentation:

This case is about an 8-years-old girl, diagnosed Hurler syndrome since 2016 and treated by Laronidase, who was admitted to the ENT Head and neck surgery department of the Academic

hospital of Casablanca in April 2022 for a bilateral nasal obstruction associated with rhinorrhea for 6 months.

The clinical examination by nasal endoscopy found polyps obstructing both nasal cavities and reaching the floor of the left cavity. She had a coarse facial features, broad nose and frontal bossing. CT scan of the paranasal sinuses (with contrast) was done and showed extensive non- enhancing soft tissue densities in bilateral maxillary, ethmoid and sphenoid sinuses extending into the nasal cavities. Associated with a voluminous soft tissue mass obstructing the cavum (fig1 & 2).

The patient underwent endoscopic polypectomy and adenoidectomy under general anaesthesia.

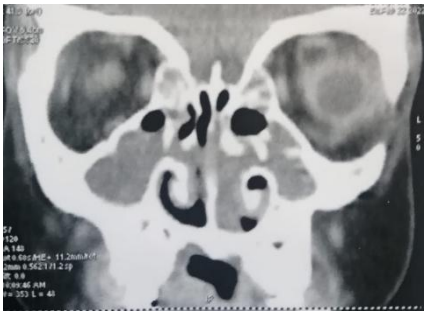

Fig 1. Coronal view of CT scan showing soft tissue in sinuses ans nasal cavities

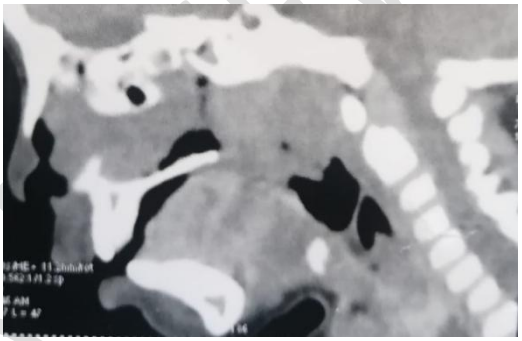

Fig 2. Sagittal view showing adenoid hypertrophy

Discussion:

MPS I is characterized by a deficiency in a-L-iduronidase enzyme activity, leading to buildup and urinary excretion of high levels of glycosaminoglycans (GAGs), specifically dermatan and heparan sulfates. The disease is genetically determined and shows autosomal recessive inheritance. It's diagnosed in several form earlier at an average 9 months of age[5].

Hurler syndrome is usually diagnosed in young children. The diagnosis may be suspected based on Common symptoms comprise atypical facies, progressive infiltration of tissues, normal intelligence or developmental delay and mental retardation, growth retardation, cardiomyopathy, valvular compromise, respiratory insufficiency, hepatosplenomegaly, recurrent respiratory infections and inflammation (otitis media, sinusitis, and tonsilitis),and head and neck manifestations include craniofacial abnormalities, depressed nasal bridge, chronic rhinitis, shortened neck, enlarged tonsils and adenoids and Upper airway resistance syndrome (UARS) has also been described in both children and adults[6].

In our case, nasal polyposis was the raison of the surgical procedures, the Recurrent and excessive rhinorrhea and enormous build-up of mucus and increase viscosity common in MPS may cause secretion accumulation and alteration in paranasal sinus drainage, causing secondary infections and chronic alterations in nasal mucosa. Although an unusual finding, formation of nasal polyps may occur as a result of chronic inflammation of nasal mucus, associated to mucus stasis.

The biological diagnosis based on a urine test to look for GAG levels that are higher than normal. The results are compared to GAG levels that are known to be normal for age-matched individuals without MPS I[3]. A clear diagnosis requires a test to measure levels of enzyme activity in the blood or skin cells. In healthy individuals, the tests show white blood cells, serum, and skin cells that contain normal levels of enzyme activity. In individuals with MPS I, the enzyme activity levels are much lower or absent, there is genetic test that fund the type of mutation of the gene responsible for making the missing enzyme.

GAG buildup in the soft tissues of the nose and throat, combined with abnormal bones, can cause the airway to become easily blocked.

Individuals with severe MPS I often have a long-term (chronic) discharge of thick mucus from the

The chronic inflammation of mucosa of the nose and paranasal sinuses, and excessive rhinorrhea and enormous build-up of mucus and increase viscosity may cause nasal polyps.

The treatment of MPS I is mainly medical, combined with physiotherapy. Surgery is needed in case of severe symptoms.

Conclusion:

MPS I is a rare, progressive disorders characterized by multisystem involvement and ENT symptoms represent the main part of their early clinical manifestation in most of patients. His management requires physicians with good knowledges of this pathology.

References:

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