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Case Report

Combination of Hurler Syndrome and Nasosinus Polyposis: Case Report and Review of the Literature

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ABSTRACT

Mucopolysaccharidosis (MPS) type 1 is characterized by a heterogeneous clinical spectrum, a progressive evolution and multisystemic manifestations including ENT. Early recognition of MPS by otolaryngologists play an increasingly important role in the multidisciplinary approach to diagnosis and management of many children with MPS. In addition to symptomatic measures, current treatments for MPS I include enzyme replacement therapy and hematopoietic stem cell transplantation alone or in combination.

In this context, we report the case of a child suffering from Haller's syndrome who underwent surgery in our department for nasosinus polyposis and adenoid vegetations, thus improving respiratory comfort and guiding the diagnosis of Hurler syndrome.

Keywords: Hurler Syndrome; ENT symptoms; Case Report; Surgery

Introduction

Head and neck disorders affect the majority of mucopolysaccharidosis patients. Symptoms such as sleep apnea, frequent respiratory and ear infections, chronic nasal discharge and enlarged tonsils and adenoids, may be indicative of MPS disease. Therefore, a more complete diagnostic search will enable us to establish an early diagnosis and, consequently, provide adequate care, enabling these patients to enjoy a better quality of life and longer life expectancy[1,2].

In this perspective, we report the case of a 3-year-old child who consulted our department for chronic nasal obstruction, bulging and sleep apnea. The patient underwent a CT scan of the nasal cavity showing nasosinus polyposis and hypertrophy of the adenoid vegetations, before deciding to undergo surgery.

Case Report

We report the case of a 3-year-old child with no particular medical history, admitted to our ENT department for bilateral nasal obstruction.

The history of the disease dates back 2 years, with the onset of bilateral nasal obstruction associated with rhinorrhea, bulging and sleep apnea. The patient's general condition was good.

On ENT physical examination, anterior rhinoscopy revealed complete filling of both nasal cavities by polyps extending to the floor of the nasal cavities. Endobuccal examination revealed bilateral tonsillar hypertrophy.

A CT scan of the nasal cavities showed a hypodense, homogeneous, confluent process filling the maxillary sinuses,

frontal sinuses, ethmoidal cells, sphenoidal sinus and nasal cavities, in favor of bilateral nasosinus polyposis. There is also a significant hypertrophy of the posterior soft tissues of the cavum, leading to obstruction of the upper airway **(Figures1 and 2)**.

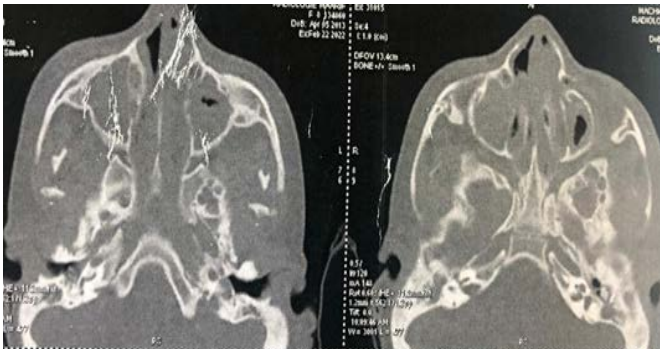

Figure 1: Axial- section CT scan of the nasal cavities showing total filling of the maxillary, ethmoidal, frontal and sphenoidal sinuses and nasal cavities.

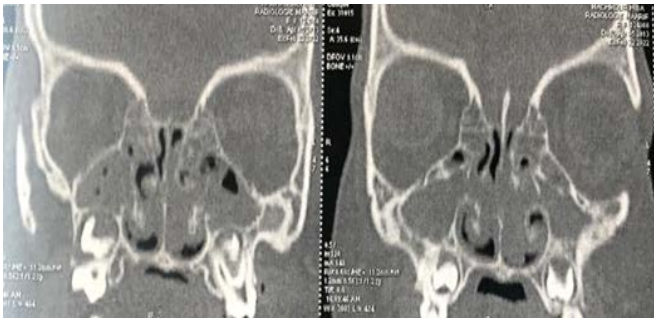

Figure 2: CT scan of the nasal cavities in coronal section, showing complete filling of the maxillary, ethmoidal, frontal and sphenoidal sinuses and nasal cavities.

On this basis, the therapeutic decision taken at the staff meeting was to perform a bilateral polypectomy, a minimal inferior turbinectomy (just the proximal part of the inferior turbinate), a bilateral middle meatotomy, a tonsillectomy and an endobuccal removal of adenoid vegetation.

The patient was then referred to the pediatrics department for further follow-up. On specialized pediatric examination, based on our ENT observations, the diagnosis of Hurler syndrome was made. In terms of ENT, during post-operative consultations, respiratory progress was excellent, with improved respiratory comfort without swelling or sleep apnea.

Discussion

A group of hereditary metabolic diseases known as mucopolysaccharidosis is caused by a deficiency in the specific enzymes involved in the lysosomal degradation of glycosaminoglycans (GAGs). The type I mucopolysaccharidoses (MPS 1) are due to a lack of alpha-L-iduronidase, the enzyme responsible for the hydrolysis of heparan sulfate and dermatan sulfate. A German pediatrician, Hurler, reported the first cases of MPS I in 1919[1,2].

Type 1 mucopolysaccharidosis can be classified into three different forms depending on the mutations, Hurler's disease and Scheie's disease, representing the two extremes of the spectrum of severity, and Hurler-Scheie disease which is the intermediate phenotype[1].

Hurler syndrome is the most severe form of MPS, leading to death in childhood. The child appears normal at birth. Diagnosis is usually made between 4 and 18 months of age, based on the association of macroglossia, skeletal deformities, hepatomegaly accompanied by umbilical and inguinal hernias, and recurrent infections of the ears and upper and lower airways[3].

If there is a strong clinical suspicion, a quantitative and qualitative study of urinary GAGs is often carried out as a first line of investigation, and if positive, can help to orientate the diagnosis of MPS I. This is confirmed by measuring alpha-L-iduronidase enzyme activity in leukocytes and fibroblasts[4].

A multidisciplinary team is required to manage the disease, including orthopedic surgeons, neurosurgeons, cardiologists, otorhinolaryngologists, physiotherapists and others. Early diagnosis of MPS is essential to enable patients to benefit from rapid therapeutic intervention[5].

A genetically-engineered analogue of human alpha-L-iduronidase, laronidase, is the enzyme replacement therapy used in the treatment of MPS I, with a dosage of 100 IU/kg[6].

Respiratory infections, facial dysmorphia and hernia were the most frequent reasons for consultation, according to[7]. All MPS I patients, and particularly those with the Hurler phenotype, are at risk of developing severe respiratory failure as a result of pulmonary restrictive disease, sleep apnea and/or asthma[8].

ENT doctors are frequently in contact with patients referred for ENT symptoms before the diagnosis of MPS is made, which enables them to make an early diagnosis of this disease. Symptoms such as sleep apnea, frequent ENT and respiratory infections, macroglossia, hypertrophy of the adenoids and tonsils often, irregular nasal septum and turbinate hypertrophy appear several years before the definitive diagnosis of MPS. Obstructive symptoms are at first more pronounced in the upper airways, with tracheobronchial manifestations occurring later. Because of the small number of patients, however, no conclusions can be drawn as to the prevalence and severity of respiratory problems for MPS[9,10].

The resulting hypertrophy and accumulation of GAGs in the adenoids and tonsils have made these structures common targets for surgical intervention. Adenoidectomy and tonsillectomy provide only temporary relief of upper airway obstruction, also risks are generally higher in a child with MPS, including postoperative hemorrhage, airway edema and extubating failure. Both chronic rhinosinusitis and chronic otitis media may develop[11,12].

It should also be noted that, in addition to the poor prognosis associated with delayed diagnosis, the fact that MPS patients undergo surgical procedures before being diagnosed is a major worry, given that the anesthetic risk is extremely high in these patients, due to deformities of the larynx, trachea and lower respiratory tract[13].

In terms of outcome, Lin et al. found that enzyme replacement therapy in MPS helped to reduce cardiac hypertrophy, particularly when administered at an early age, but has little effect on valvular damage. Without replacement therapy, Bousssof's results show that the general condition of most patients deteriorates, with progressive physical deterioration and loss of quality of life[14,15].

Conclusion

Mucopolysaccharidosis type 1 is responsible for multisystem damage that progressively worsens gradually with age. This

calls for multidisciplinary management based on specific treatment and symptomatic measures. A wide range of ENT symptoms appear in the early stages of MPS, including rhinosinusitis, macroglossia, adeno-tonsillar hypertrophy, nasal obstruction, OSA, progressive respiratory disorders and hearing loss. Otorhinolaryngologists should be aware of MPS, particularly in young children (2-3 years) with an indication for adenotonsillectomy.

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