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Case Report

Bilateral Antrochoanal Polyps in Hurler Syndrome: A Rare ENT Presentation of Mucopolysaccharidosis Type I

Parth Sharma[1]*, Pooja Goyal[2] and Sushant Joshi[3]

[1]MS ENT Resident, Department of Otolaryngology–Head and Neck Surgery, Ananta Institute of Medical Sciences and Research Center, India

[2]Senior Resident, Department of Otolaryngology–Head and Neck Surgery, Ananta Institute of Medical Sciences and Research Center, India

[3]Professor, Department of Otolaryngology–Head and Neck Surgery, Ananta Institute of Medical Sciences and Research Center, India

***Corresponding Author:** Parth Sharma, MS ENT Resident, Department of Otolaryngology–Head and Neck Surgery, Ananta Institute of Medical Sciences and Research Center, India.

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Abstract

Hurler syndrome (mucopolysaccharidosis type I) is a rare autosomal recessive lysosomal storage disorder caused by a deficiency of the enzyme α-L-iduronidase, resulting in progressive accumulation of dermatan and heparan sulfate. Although the disorder is classically associated with skeletal dysplasia, neurodevelopmental delay, and multisystem involvement, otorhinolaryngological manifestations frequently represent the earliest presenting features. We report a rare case of a 5-year-old male child presenting with nasal obstruction due to bilateral antrochoanal polyps, subsequently diagnosed as Hurler syndrome. This case highlights the importance of early ENT recognition of systemic metabolic disorders and emphasises the role of the otorhinolaryngologist in early diagnosis and multidisciplinary management.

Keywords: Hurler Syndrome; Mucopolysaccharidosis Type I; Antrochoanal Polyp; Nasal Polyposis; ENT Manifestations; Glycosaminoglycans

Abbreviations

MPS I: Mucopolysaccharidosis Type I; GAG: Glycosaminoglycans; CECT: Contrast-Enhanced Computed Tomography; FESS: Functional Endoscopic Sinus Surgery; ERT: Enzyme Replacement Therapy

Introduction

Mucopolysaccharidosis type I (MPS I) is a rare lysosomal storage disorder caused by a deficiency of the enzyme α-L-iduronidase, leading to the accumulation of dermatan sulfate and heparan sulfate within lysosomes [9]. This results in progressive multisystem involvement affecting the skeletal, neurological, cardiac, respiratory, ophthalmic, and otorhinolaryngological systems [9]. MPS I exists as a clinical spectrum ranging from mild (Scheie syndrome) to intermediate (Hurler–Scheie syndrome) and severe (Hurler syndrome) phenotypes, with Hurler syndrome representing the most severe form [2].

The birth prevalence of MPS I varies globally, with higher incidence reported in regions with increased consanguinity [3]. Despite its rarity, MPS I remains underdiagnosed due to nonspecific early symptoms [4]. ENT manifestations such as chronic rhinitis, recurrent otitis media, upper airway obstruction, adenotonsillar hypertrophy, and hearing loss frequently present within the first year of life and may precede classical systemic features [9]. As a result, otorhinolaryngologists are often the first specialists to encounter these patients. This report describes a rare ENT presentation of Hurler syndrome with bilateral antrochoanal polyps [1].

Case Presentation

A 5-year-old male child presented to the Department of Otolaryngology–Head and Neck Surgery with complaints of right-sided nasal swelling and obstruction for the preceding three days. According to the parents, the child had a long-standing history of noisy breathing and habitual snoring since infancy. There was also a history of persistent nasal discharge requiring frequent nasal cleaning, suggesting chronic upper airway involvement.

The child was noted to have a significant developmental delay. Gross motor milestones were achieved at approximately 33% of age-appropriate expectations, while fine motor development was limited to around 25%. Speech development was also delayed, although formal assessment could not be completed at the time of presentation. There was no history of recurrent lower respiratory tract infections or seizures.

Antenatal and perinatal histories were uneventful, with no reported maternal illness, drug exposure, or birth complications. The child was delivered at term by normal vaginal delivery. Family history was non-contributory, and no consanguinity was reported. There was no known family history of genetic or metabolic disorders.

The combination of chronic upper airway symptoms, developmental delay, and progressive nasal obstruction prompted further detailed evaluation.

Clinical examination findings

General examination

The child exhibited marked short stature, with a height of 95 cm and a weight of 13.57 kg, both below the 3rd percentile for age, consistent with growth retardation seen in MPS I.

Craniofacial features

Distinct dysmorphic features were evident, including:

- Coarse “gargoyle-like” facial features characteristic of Hurler syndrome
- Macrocephaly with a head circumference of 53 cm
- Prominent frontal bossing
- Depressed nasal bridge

Ophthalmologic findings

Bilateral corneal clouding was noted on examination, a recognised manifestation of glycosaminoglycan deposition.

Abdominal findings

A prominent umbilical hernia was observed on abdominal examination.

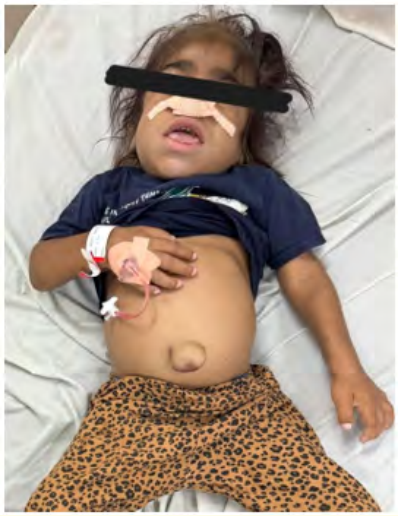

Figure 1: Clinical photograph showing protruded abdomen with umbilical hernia.

Otolaryngologic findings

- A pale, polypoidal mass was visualised in the right nasal cavity, causing complete nasal obstruction
- Bilateral tympanic membranes were dull and retracted, suggestive of middle ear effusion
- Oral cavity examination revealed a high-arched palate
- Examination of the oropharynx was limited due to severe macroglossia.

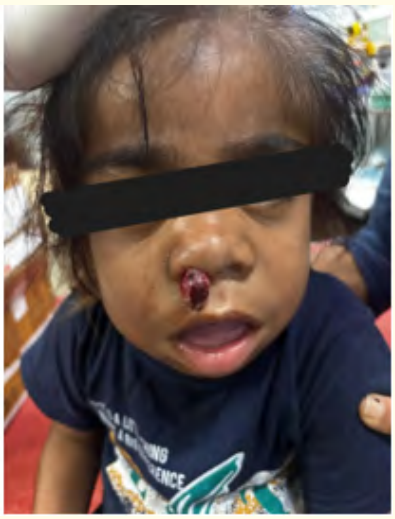

Figure 2: Clinical photograph showing a nasal mass from the right nostril.

Diagnostic investigations

Imaging studies

CECT FACIAL BONE - A polypoidal lesion is seen involving bilateral nasal cavities, maxillary sinuses and bilateral maxillary ostia. Posteriorly, the lesion is reaching upto the choana; anteriorly, the lesion is protruding outside the nasal cavity. The nasal septum is seen to be deficient anteriorly. No intracranial extension of the lesion is noted. No bony erosion or destruction is noted. Mucosal thickening is seen in bilateral ethmoidal sinuses.

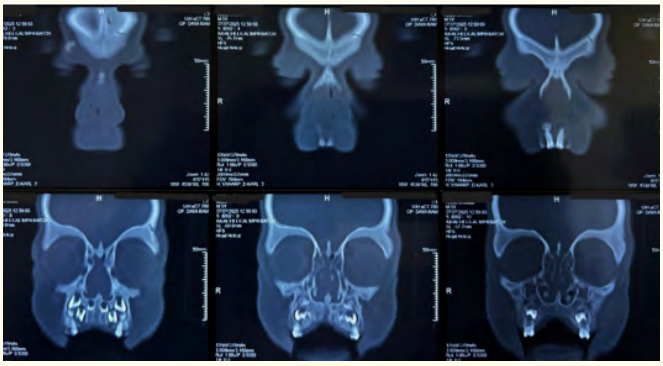

Figure 3: CECT facial bones (coronal) showing a polypoidal lesion involving bilateral nasal cavities, maxillary sinuses and bilateral maxillary ostia.

CECT – Head

Neuroimaging demonstrated multiple hypodense foci in the bilateral periventricular regions, consistent with dilated perivascular spaces. Diffuse cerebral atrophic changes were evident, characterised by ventricular dilatation and widened sulcal spaces. Features suggestive of hydrocephalus were also noted. These findings correlate with central nervous system involvement characteristic of Hurler syndrome.

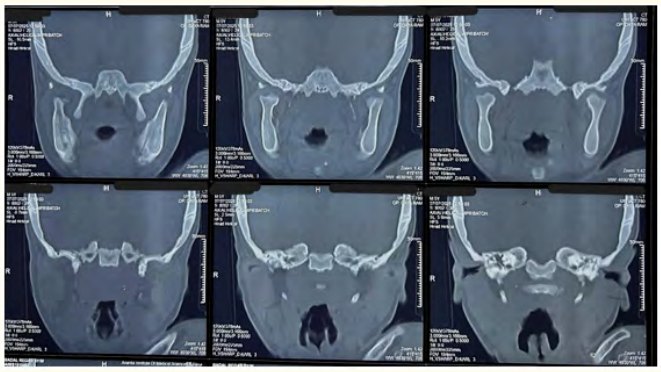

Figure 4: Adenoidal hypertrophy causing significant narrowing of the nasopharyngeal airway.

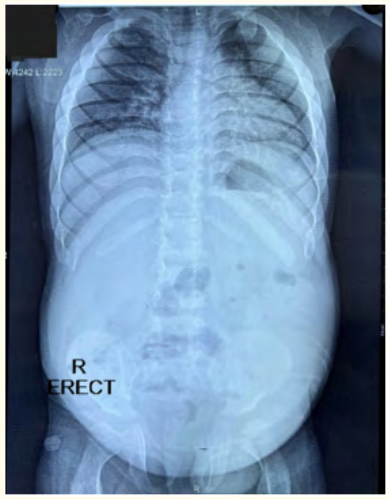

Figure 5: Chest and abdomen radiograph (erect) showing abdominal distension.

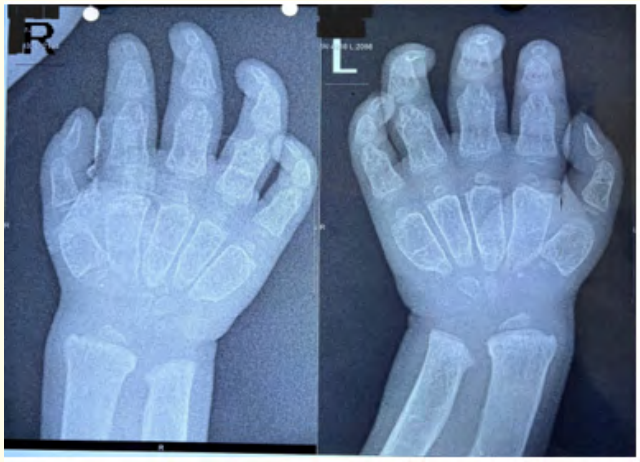

Figure 6: Hand radiographs showing dysostosis multiplex features (short, broad metacarpals/phalanges).

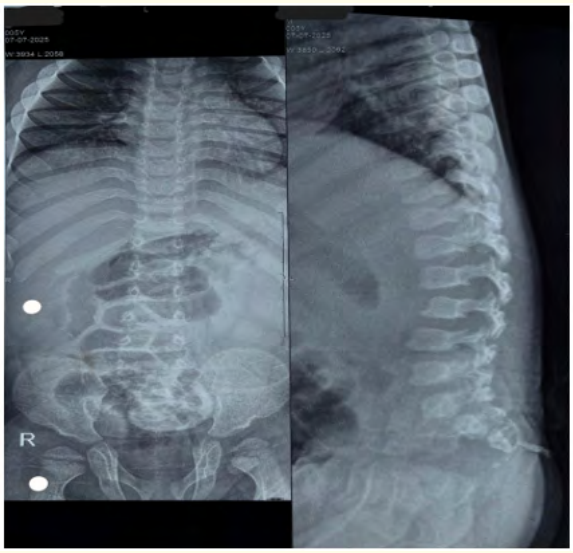

Figure 7: Spine radiographs (AP and lateral) showing kyphotic curvature/vertebral beaking (dysostosis multiplex).

Histopathological findings

Histopathological examination of the resected polypoidal tissue revealed polypoidal tissue lined by pseudostratified ciliated mucosal epithelium with a thickened basement membrane. The submucosa showed edematous stroma with inflammatory cell infiltrate predominantly comprising lymphocytes, eosinophils, and plasma cells, along with interspersed mucous glands.

Histopathological impression

Features were consistent with an antrochoanal polyp.

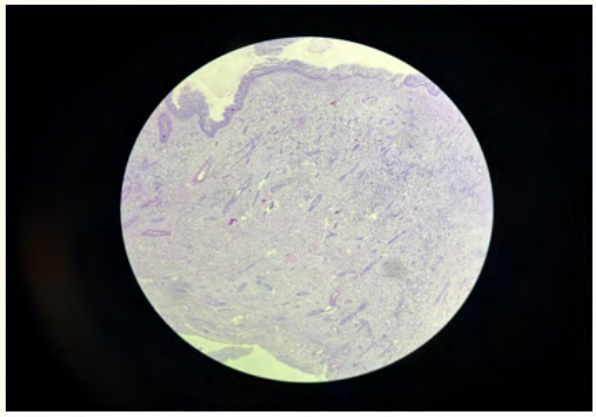

Figure 8: Histopathology (H&E) consistent with an antrochoanal polyp.

Management

The patient was planned for functional endoscopic sinus surgery (FESS) to excise the antrochoanal polyps and restore nasal patency. Anticipating a difficult airway due to severe macroglossia, short neck, and potential airway rigidity, meticulous anaesthetic planning was undertaken.

Postoperatively, the patient was referred to the pediatric department for further evaluation and initiation of enzyme replacement therapy. Genetic counselling was provided to the family.

Discussion

Hurler syndrome (mucopolysaccharidosis type I-H) represents the severe end of the MPS I spectrum and is characterised by early onset, rapid disease progression, and extensive multisystem involvement [9]. Accumulation of dermatan sulfate and heparan sulfate due to α-L-iduronidase deficiency leads to progressive organ dysfunction affecting the skeletal, neurological, cardiac, respiratory, and otorhinolaryngological systems [2].

ENT manifestations are among the earliest and most consistent clinical features of Hurler syndrome [9]. Upper airway obstruction results from a combination of adenotonsillar hypertrophy,

macroglossia, thickened upper airway mucosa, and skeletal abnormalities of the craniofacial framework [1]. Chronic rhinitis, recurrent otitis media with effusion, conductive hearing loss, and sleep-disordered breathing are frequently encountered and often precede the diagnosis of the underlying metabolic disorder [1].

Sinonasal polyposis in children is uncommon, and antrochoanal polyps are typically unilateral [1]. The presence of bilateral antrochoanal polyps, as observed in this case, is exceptionally rare and suggests an underlying systemic pathology rather than isolated sinonasal disease [1]. Glycosaminoglycan deposition within the sinonasal mucosa leads to chronic inflammation, mucosal thickening, glandular hyperplasia, and stromal oedema, creating a favourable environment for polyp formation [9].

Radiological findings such as cerebral atrophy, hydrocephalus, and dysostosis multiplex are well-documented in Hurler syndrome and correlate with neurodevelopmental delay [3,4]. Skeletal abnormalities, including vertebral beaking and widened ribs, are characteristic features resulting from abnormal bone remodelling [3].

From an anaesthetic and surgical perspective, patients with Hurler syndrome pose significant challenges due to craniofacial abnormalities, macroglossia, restricted neck mobility, and potential cardiac involvement [2]. These factors necessitate careful preoperative evaluation and multidisciplinary coordination to minimise perioperative risks [4].

Conclusion

Hurler syndrome may initially present with otorhinolaryngological manifestations before the full spectrum of systemic features becomes evident. Bilateral antrochoanal polyps in a pediatric patient, particularly when associated with developmental delay and dysmorphic features, should raise suspicion of an underlying metabolic storage disorder.

This case emphasises the importance of maintaining a high index of suspicion when evaluating unusual or bilateral nasal polyposis in children. Early diagnosis allows for appropriate multidisciplinary management, timely initiation of enzyme replacement therapy, and genetic counselling, thereby improving quality of life and potentially altering disease progression.

Otorhinolaryngologists play a pivotal role in the early detection of Hurler syndrome, underscoring the need for comprehensive evaluation beyond local sinonasal pathology.

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