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Chapter 2

Unveiling Complexity: Congenital Ptosis, Global Delay, and Asthma in a Paediatric Patient

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Abstract

This case report describes the clinical presentation, diagnostic evaluation, and management of an 8 years old male who presented with chronic cough, difficulty in swallowing, and a history of global developmental delay. The patient was diagnosed with congenital ptosis, global developmental delay, and bronchial asthma. The case highlights the importance of a multidisciplinary approach in managing complex cases with multisystem involvement.

Keywords: Paediatric case, Dysphagia, Congenital asthma, Global development delay, Congenital Ptosis

1. Introduction

Global developmental delay (GDD) and congenital ptosis are both rare conditions that can significantly affect a child's quality of life [1]. Additionally, the presence of bronchial asthma complicates the clinical scenario, necessitating comprehensive care [2, 3]. GDD encompasses significant delays in cognitive, motor and social milestones, often requiring extensive diagnostic and therapeutic interventions [4]. Congenital ptosis, characterized by drooping of the upper eyelid present at birth, can interfere with vision and further hinder developmental progress [5]. The coexistence of bronchial asthma, a chronic respiratory condition, adds complexity to the clinical scenario, demanding comprehensive and multidisciplinary care [6, 7]. his case report explores the rare coexistence of these conditions in an 8-year-old boy, emphasizing the challenges of multisystem involvement and the need for comprehensive, multidisciplinary management [8].

2. Case Presentation

Patient Demographics: The patient is an 8-year-old male who presented to the Paediatric ward of a tertiary care hospital with a primary complaint of a persistent cough that had been ongoing for two months. The cough was notably dry in nature, more pronounced in intensity during the night, and had gradually worsened over time. The patient's mother reported that the cough often disrupted his sleep, leading to fatigue and irritability during the day. Image is shown in Figure 1.

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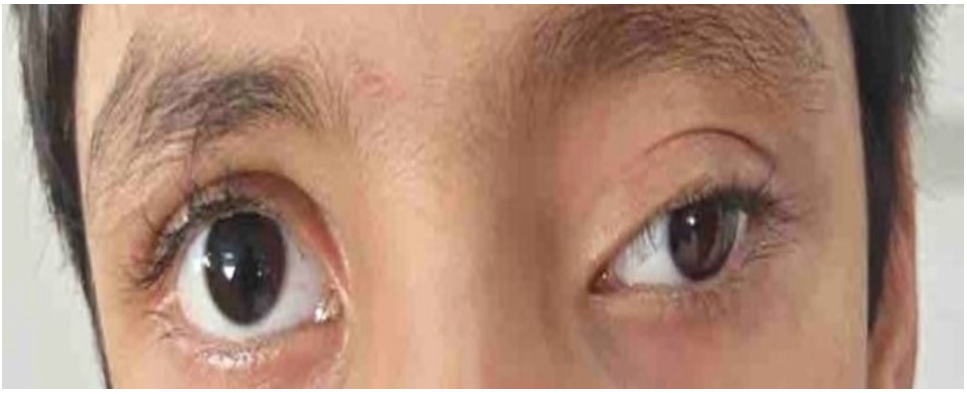

Figure 1: The above image shows Left eye ptosis with absent lid crease with Left eye hypotropia ˜30 degrees. Lid retraction observed in downward gaze of left eye.

History of Present Illness: The mother noted that the patient's cough began insidiously two months prior to admission, without any associated fever, nasal congestion, or other symptoms of upper respiratory infection. Despite trying various over-the-counter cough medications, there was no significant improvement. The cough was more severe at night, leading to multiple awakenings. There was no history of exposure to cold air, allergens, or any environmental factors that could exacerbate asthma symptoms, although the child occasionally wheezed during episodes of coughing.

Developmental History: The patient has a documented history of global developmental delay. His developmental milestones were significantly delayed compared to peers, with the patient walking at 2 years of age and speaking in short sentences only by the age of 5. His cognitive development was also delayed, as noted by difficulties in learning and social interactions at school. Despite these challenges, the patient attended a mainstream school with additional support services. His parents expressed concern over his academic performance and social integration, as he struggled with basic tasks that other children his age managed easily.

Past Medical History: Since infancy, the patient had difficulty swallowing, a condition known as dysphagia, which had been persistent since childhood. The difficulty in swallowing primarily affected solid foods, with the child often taking prolonged periods to finish meals or avoid certain foods altogether. There was no history of aspiration, but the parents noted frequent coughing during meals. The patient had not been previously evaluated for this issue in detail until the current presentation. Additionally, the patient was noted to have congenital ptosis-drooping of the upper eyelid-since birth, which had not significantly affected his vision but was cosmetically noticeable.

Family History: There was no significant family history of congenital anomalies, developmental delays, or chronic respiratory conditions. The patient's parents were non-consanguineous, and there were no siblings with similar conditions.

Social History: The patient lived with his parents and attended a regular school with special educational support. His social interactions were limited, and he often preferred solitary activities. The family was supportive and involved in his care, with both parents actively participating in managing his health needs.

Physical Examination: Upon examination, the patient was alert and cooperative, though shy and somewhat withdrawn. He appeared smaller in stature compared to his peers. His vital signs were stable, with a respiratory rate of 22 breaths per minute and no evidence of acute distress.

Respiratory Examination: Auscultation of the lungs revealed bilateral wheezing, more pronounced during forced expiration, consistent with obstructive airway disease. No crackles or other adventitious sounds were noted.

Neurological and Ophthalmological Examination: The neurological examination confirmed the presence of global developmental delay, with reduced cognitive abilities and impaired fine motor skills. The patient also exhibited congenital ptosis of the left eye, with the eyelid partially covering the pupil, but with no 2 of 8 significant impairment of eye movements or vision.

Investigations: A series of investigations were conducted to assess the underlying causes of the patient's symptoms and developmental delays. Blood Tests: Renal Function- Normal kidney function indicated by normal creatinine and urea within the acceptable range. Alkaline Phosphatase: Normal for age, likely reflecting bone activity in a growing child.

CBC Findings: Normal haemoglobin and RBC count suggest adequate haematological status.
Eosinophilia (13%) may be linked to the patient's history of bronchial asthma, hinting at an allergic component or atopy.

Chest X-Ray: The chest X-ray looks mostly normal, with no signs of serious lung problems like infections (such as pneumonia) or any unusual growths (like tumour's). However, the X-ray shows that the lungs look a bit more "puffed up" than usual, which might suggest some mild blockage or difficulty in the airways, possibly related to conditions like asthma.

The overall impression chest X-ray appears mostly unremarkable, with no evidence of acute infections (e.g., pneumonia) or any masses suggestive of neoplasia. However, there is a mild increase in lung hyperinflation, which could be indicative of airway obstruction or reactive airway disease, potentially consistent with asthma or a similar condition. Clinical correlation with pulmonary function tests and history is advised.

RFT Test	
UREA	22.5 mg/d
CREATININE	0.51 mg/dl
ALKALINE PHOSPHATE	206 U/L
CBC	
Hb	13.8 gm/dl
TLC	8.3 10^3 U/L
DIFFERENTIAL COUNT	
NEUTROPHILS	37%
LYMPHOCYTES	45%
Eosinophils	13%
Total RBC Count	5.3 millions/cu.mm

Pulmonary Function Tests (PFTs): The breathing tests show that the amount of air the person can blow out in one second (FEV1) is lower than normal, and the ratio of this amount to their total lung capacity (FEV1/FVC) is also low. This means the airways are partially blocked, making it harder to breathe out. These results match what is usually seen in conditions like asthma, where the airways become narrow and inflamed.

Doppler Test Upper Limb Impression: The test shows that there is a significant blockage at the start of the main artery in the left shoulder (called the left subclavian artery). This blockage is reducing blood flow to the arm, and the blood that does flow beyond the blockage is weaker and less steady than normal. This could lead to symptoms like weakness, pain, or numbness in the left arm.

Doppler Test Lower Limb Impression: The test shows a significant blockage in the main artery (the aorta) that supplies blood to both legs. This blockage is reducing the blood flow to the lower body, and the blood that does reach the legs is weaker and less steady than it should be. This can cause symptoms like leg pain, cramping, weakness, or fatigue, especially during activities like walking.

CT scan: The CT scan shows a problem with the aorta (the main blood vessel that carries blood from the heart). There is a missing or very narrow section of the aorta in the area between two important branches: the artery that supplies blood to the left side of the neck (left common carotid artery) and the artery that supplies blood to the left arm (left subclavian artery).

Blood is still reaching the left subclavian artery, but it's taking a longer, indirect route through smaller blood vessels (called collaterals) that have formed to bypass the blockage. This condition is known as a "pre-ductal interruption of the aorta," and it may cause problems with blood flow to the lower body and left arm. It often requires medical attention, sometimes including surgery or other interventions.

Genetic Testing and Neuroimaging: Due to the global developmental delay, genetic testing was performed, but no definitive syndromic diagnosis was established. Brain MRI was also performed, showing mild cerebral atrophy, which could be consistent with developmental delay but was non-specific.

3. Discussion

This case highlights the complexity of managing a child with multiple chronic conditions, each requiring specific interventions and long-term monitoring. The interplay between bronchial asthma, global developmental delay, and congenital ptosis necessitates a holistic approach to care, involving various specialists to optimize outcomes. Early diagnosis and intervention are critical in managing developmental delays, while effective asthma management is essential in preventing long-term respiratory complications.

About 1% to 3% of children have global developmental delay, which can have a variety of causes, such as genetics, structural brain abnormalities, metabolic diseases, and environmental variables [7]. Despite being mostly a cosmetic issue, congenital ptosis can cause strabismus or amblyopia if treatment is not received [8]. An estimated 5-10% of children worldwide suffer from bronchial asthma, which frequently makes managing concomitant illnesses more difficult by raising morbidity. It is uncommon for these three disorders to coexist in one patient, necessitating an integrated approach to treatment [6].

Comparison with Similar Cases

Isolated occurrences of congenital ptosis linked to both syndromic and non-syndromic developmental abnormalities have been reported in studies. For instance, noted that congenital ptosis can occasionally be a standalone abnormality or a component of a more comprehensive developmental condition [3]. Highlighted how ocular defects are inherited genetically, showing that congenital ptosis is sometimes associated with hereditary disorders involving GDD [5].

Patients with developmental delays have also been reported to have bronchial asthma, which may be made worse by noncompliance with therapy and trouble identifying triggers. By describing a distinct triad of circumstances that need to be managed simultaneously, the current case contributes to the body of literature [9].

Discussion Points

Neurological and Developmental Concerns: Studies looking into the genesis of GDD have found that the patient's modest brain atrophy and developmental delays on MRI are consistent with this. Although modest cerebral atrophy may not be identifiable, it may be a sign of underlying neurological dysfunction. Although a precise genetic diagnosis was not made, comparable cases have demonstrated the value of sophisticated genetic testing, such as whole-exome sequencing, in detecting subtle syndromic presentations [7].

Ophthalmological Aspects: Affected children's psychological well-being may be impacted by congenital ptosis, which is sometimes disregarded in the absence of functional disability. Early ophthalmological examination is crucial to preventing consequences like amblyopia or astigmatism, according to [1]. Even though the patient's vision was unharmed, it is standard practice to refer them for a surgical evaluation.

Asthma and Respiratory Health: As in this case, the patient's wheezing and nighttime flare-up of symptoms are typical signs of childhood asthma [6]. Guidelines encourage comprehensive care, including exposure to inhaled corticosteroids and education on avoiding triggers. Furthermore, this instance's absence of notable external stimuli emphasizes how idiopathic some asthma symptoms can be.

Multidisciplinary Management: A comprehensive approach was assured by the participation of several specialties, including pulmonology, ophthalmology, and neurology. Research has indicated that individualized education plans (IEPs) and coordinated care are highly beneficial for children with multisystem disorders [2]. The treatment sessions and school-based support services for this patient are in line with suggestions for improving developmental results.

4. Conclusions

The difficulties of caring for a young patient who has congenital ptosis, global developmental delay (GDD), and bronchial asthma all coexist are highlighted by this example. Significant obstacles are presented by each ailment alone, and their combination emphasizes the necessity of a patient-centered, multidisciplinary approach to care. While efficient asthma treatment is critical to enhancing respiratory health and averting exacerbations, early detection and intervention for developmental delays are critical for optimizing cognitive and motor outcomes.

Even in cases where there is no acute visual impairment, ophthalmological consultation for congenital ptosis improves the child's quality of life by addressing both functional and psychosocial elements. The value of combining medical, educational, and psychosocial assistance is demonstrated by the integrated treatment given in this instance, which included referrals for developmental and ophthalmological examination, customized educational planning, and inhaled corticosteroids for asthma.

This instance emphasizes the value of prompt referrals to expert services, proactive parental involvement, and attentive follow-up. By recording the uncommon presence of these three illnesses, it also adds to the body of literature, highlighting the importance of specialized, multidisciplinary care in attaining favourable long-term results for kids with complicated medical needs.

References

[1] S. Yadegari. Approach to a patient with blepharoptosis. *Neurol Sci*, 37:1589–1596, 2016. doi:10.1007/s10072-016-2633-7.

[2] P. PhDa Pavone, Sung Yoon PhDc Cho, A. D. PhDb Praticò, R. PhDa Falsaperla, M. PhDb Ruggieri, and * Jin, Dong-Kyu PhDc. Ptosis in childhood: A clinical sign of several disorders. *Medicine*, 97(36):e12124, September 2018. doi:10.1097/MD.0000000000012124. —.

[3] A. Berry-Brincat and H. Willshaw. Paediatric blepharoptosis: a 10-year review. *Eye*, 23:1554–1559, 2009. doi:10.1038/eye.2008.311.

[4] Peter J. M. D. * Sakol, Geva M. D. Mannor, Massaro Mph†, and M. M. D. Bruce. Mph‡. congenital and acquired blepharoptosis. *Current Opinion in Opthalmology*, 10(5):335–339, October 1999. doi:10.1097/00055735-199910000-00010.

[5] D. N. Hu. Prevalence and mode of inheritance of major genetic eye ; diseases in China. *Journal of Medical Genetics*, 24:584–588, 1987. doi:10.1136/jmg.24.10.584.

[6] Kenneth P. M. D. Vestal, Stuart R. M. D. Seiff, F. A. C. S., and J. Michael M. D. Lahey. Congenital ptosis in monozygotic twins. *Ophthalmic Plastic Reconstructive Surgery*, 6(4):265–268, December 1990.

[7] V. Krieger, J. A. Amador-Campos, and M. Peró-Cebollero. Interrater agreement on behavioral executive function measures in adolescents with attention deficit hyperactivity disorder. *International Journal of Clinical and Health Psychology*, 19(2):141–149, 2019. doi:10.1016/j.ijchp.2019.02.007.

[8] R. E. Essawy and M. A. Elsada. Clinical and demographic characteristics of ptosis in children: A national tertiary hospital study. *European Journal of Ophthalmology*, 23(3):356–360, 2013. doi:10.5301/ejo.5000239.

[9] P. Papoff, E. Caresta, S. Gazzanelli, R. Pinto, C. Cerasaro, C. Moretti, and F. Midulla. Sevoflurane inhalation for severe bronchial obstruction in infants with bronchiolitis. *Int J Immunopathol Pharmacol*, 25(2):493–7, April–June 2012. doi:10.1177/039463201202500219.