Case in Point: Sandifer Syndrome

March 01, 2007
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A previously healthy full-term, 30-week-old infant presents to the emergency department after a 30-minute episode in which he turned blue after a feeding. His mother states that the infant was also stiff, that he "arched his back," and that his eyes "had a blank look." There was no twitching associated with this episode, and the infant had no fever, cold symptoms, or any sick contact.

On examination, the child is comfortable and well-nourished. Examination of CNS function, the heart, and the GI tract reveals normal findings. Blood tests to rule out infection, seizures, and electrolyte abnormalities yield normal results. Referral to a neurologist is recommended to evaluate the cause of the "stiffening" episode.

This child was subsequently found to have Sandifer syndrome. This uncommon, but well-documented condition affects children and young adults with gastroesophageal reflux disease (GERD).[1] Clinical Picture

Following observations by Drs Sandifer and Kinsbourne in 1964, the symptom complex of dystonic movements in association with GERD was named "Sandifer syndrome" by Sutcliffe. Sandifer syndrome is a sudden-onset dystonia that can involve various body parts: the neck and head are most commonly affected. This syndrome is more common in infants and children who have GERD or hiatal hernias. There are a few reports in adults. Patients may present with episodes of torticollis, laterocollis, or retrocollis. Occasionally, arching of the back, opisthotonus-like positions, and eye deviations may be concurrently seen.

Dystonic episodes have been shown to be temporally associated with food intake, although intermittent episodes are frequently present. Characteristically absent during sleep and paroxysmal in nature, these episodes are frequently mistaken for seizures. Pathophysiology The mechanisms that underlie Sandifer syndrome still need to be defined. The etiological association with GERD is confirmed by the resolution of dystonia with anti-reflux therapy. Corrado and colleagues found a significant correlation between low pH and dystonia. Bruchheimer's group found evidence to the contrary, however. Various authors speculate that the involuntary dystonia is a physiological response that attempts to alleviate symptoms of GERD. However, Puntis and coworkers showed that posturing actually increases the amplitude and velocity of esophageal peristalsis. This may, in turn, promote acid clearance, thereby providing symptom relief.

Other explanations, such as neurological innervations, have been offered. One hypothesis is that the neck muscles and diaphragm share a common innervation. In patients with a hiatal hernia and coexisting acid reflux, local diaphragmatic irritation occurs, resulting in a referred dystonic spasm in the neck. However, this does not seem to hold true in patients with only GERD without a hiatal hernia.

Another possible explanation is that there is brain stem involvement secondary to pain elicited from acid reflux, which causes the dystonic reaction, eyes rolling upward, and excessive lacrimation. Differential Diagnosis Sandifer syndrome has been mistaken for such neurological disorders as status epilepticus, complex partial seizures, and refractory seizures. The differential also includes psychiatric disorders, cervical anomalies, congenital muscular torticollis, trauma, and inflammatory conditions of the neck and head. Diagnosis and Treatment Demonstration of GERD on pHmetry, impedance testing, or nuclear technetium scan or the presence
of esophagitis on endoscopy may be helpful, even though a direct correlation with dystonic episodes may be difficult to prove. A neurological workup may help elicit the cause. Resolution--either gradual or dramatic--following anti-reflux therapy confirms the diagnosis. Medical therapy is usually successful, although a few cases have responded well to fundoplication. Clinical Take-Home Message
Pediatricians need to be aware of Sandifer syndrome when faced with an infant or child with a suspected seizure disorder, developmental spasticity, or neuropsychiatric disorder. Once identified, children with Sandifer syndrome can be medically or, in some cases, surgically treated with good results.

References: REFERENCES:

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