A 6-month-old boy was admitted to the Department of Paediatrics, University of Chieti, Italy, with a 3-month history of uncontrollable dystonic posture of the neck. During the second month of life, the child began to present with a twisting of the neck to one side that persisted for 7 days, after which the abnormal position moved to the other side. The movements were not associated with vomiting, cyanosis, loss of consciousness, tonic or clonic movements, or a post-ictal state. The child’s personal medical history was otherwise unremarkable. Many different specialists, including orthopedic surgeons, otolaryngologists, orthoptists, and ophthalmologists, examined the child and found no evidence of pathology. The investigations included electroencephalograms, magnetic resonance imaging of the head, and X-ray and ultrasound of the neck, but showed no anomalous findings.

On admission, physical examination revealed normal growth parameters, vital signs, and neurological findings. After sound stimulation, the child showed difficulty in turning his head to the opposite side and presented resistance to passive mobilization of the neck. A specialist in neurological evaluation demonstrated reduced reaction to the environment and a slight reduction of ocular motility due to torticollis.

Laboratory investigations, including a complete blood count; blood electrolytes; serum pyruvate, lactate, and ammonium levels; and urine and blood amino acids, were within normal range. An occult blood test produced a negative result.

A paroxysmal non-epileptic event was suspected and 24-hour pH metering revealed postprandial acid reflux of severe grade.

Gastroesophageal ultrasound excluded the presence of hiatal hernia. Therapy with ranitidine (8 mg/kg per day) was started, after which symptoms rapidly improved and then disappeared completely within a few days. After 8 months of treatment with ranitidine, the treatment was ended and no problems have recurred.

**For diagnosis, see page 18**

Editor’s note: Each month, this department features a discussion of an unusual diagnosis in genetics, radiology, or dermatology. A description and images are presented, followed by the diagnosis and an explanation of how the diagnosis was determined. As always, your comments are welcome via email at pedann@Healio.com.
Diagnosis:
Sandifer Syndrome

Torticollis describes a lateral head tilt with rotation of the chin toward the side opposite to the tilt. This condition is a manifestation of a variety of underlying pathologies. Sandifer syndrome (SS) is a rare cause of torticollis in infants that results from gastroesophageal reflux disease (GERD).\(^1\) SS occurs in less than 1% of children with GERD, and symptoms usually occur in early infancy or childhood, although some cases have been reported in adults.\(^2\)

The pathogenetic mechanisms are unclear, although a dysfunction of the lower esophagus is thought to be the most important precipitating factor. The abnormal posture is considered to result from a reflex mechanism that aims to protect the air passages from reflux material or to relieve the abdominal discomfort caused by acid reflux. Diagnosis may be difficult and often requires multiple evaluations by specialists. Infants are often misdiagnosed due to paroxysmal neurobehaviors and receive unnecessary medication and treatment. Analysis of the torticollis may help establish a suitable strategy for diagnosis. Symptoms of SS usually promptly respond to antireflux medications and/or fundoplication.\(^3\)

DISCUSSION

Torticollis is a clinical symptom characterized by a lateral head tilt and chin rotation toward the side opposite to the tilt. It is a very nonspecific symptom occurring in different childhood conditions (Table 1), which may be the reason so many specialists are brought in for consultation. Analysis of torticollis may help establish a suitable strategy for diagnosis.\(^4,5\)

In infants, congenital muscular torticollis associated with a contracture of the sternocleidomastoid muscle is the most common etiology. This condition generally improves with a regimen of manual cervical stretching. Unusual nonmuscular causes of torticollis in infants include ocular torticollis caused by eye muscle weakness, neural axis abnormalities, congenital anomalies of the cervical spine and of the occiput, benign paroxysmal torticollis, and SS resulting from gastroesophageal reflux.\(^4,5\) Torticollis in older children is most frequently a manifestation of atlantoaxial rotator displacement resulting from trauma or oropharyngeal inflammation (ie, Grisel’s syndrome).\(^4,5\)

SS is most commonly associated with normal examination findings, especially in infants, as occurred in the case discussed here. Irritability, crying, eye deviation with head version, and paroxysmal dystonic posture are common symptoms.\(^1\) In our patient, the absence of vomiting and other signs of GERD, such as crying, irritability, and growth failure, contributed to the diagnostic delay. Also, in the case of this child, the stiff neck posture was not intermittent (as usually occurs) but persisted for many days, which was a confounding characteristic. Moreover, symptoms normally occur during or shortly after feeding,\(^3\) whereas in our patient feeding did not interfere with neck posture. It should be noted, therefore, that children with SS with subtle esophageal disease may be more challenging to identify.

Since the description of SS in 1962,\(^3\) many cases have been reported in the literature. These reports document a preponderance of male patients, common onset of symptoms during the first years of life, and normal neurologic development in most cases. Sporadic cases present a failure to thrive. The most common symptoms reported are irritability, dysphagia, and dystonic postures. Some authors report an association with hiatal hernia,\(^1,3,6\) West syndrome, Cornelia de Lange syndrome,\(^7\) and Opitz syndrome.\(^8\) Sporadic cases associated with chronic anemia,\(^6\) mental impairment,\(^6,9\) or intestinal malrotation have also been described.

Fundoplication surgery is required for management of about 25% of patients, whereas more than 50% of infants can be successfully treated with oral antacid drugs. Most children have a good long-term prognosis, with full recovery after therapy. There is a report of one child with mental retardation who died due to an underlying disease and not because of SS.\(^9\)

The pathophysiology of SS is still not clear. One hypothesis is that the posturing might induce reflux episodes.\(^1\) However, this does not explain the resolution of symptoms with antireflux medical therapy or fundoplication. A second possibility is that the torticollis due to SS is a tic behavior caused by an unknown factor consisting of simultaneous head tilt and abdominal muscle contraction(s) that induce reflux.\(^10\) The third and most plausible hypothesis is that the reflux episodes produce the head tilting. This idea is supported by the resolution of the behavior after surgical treatment of the reflux, as described in many case reports.\(^3,10\) This hypothesis is also supported by the demonstration that head tilting induces an increase of esophageal motility and pressure.\(^10\)

The intermittent occurrence of torticollis with alternating directions, normal sternocleidomastoid muscle, and normal cervical radiographic findings make SS a probable diagnosis, which, therefore, requires upper gastrointestinal studies. pH metering, which is inexpensive and quick, plays a significant role in the diagnosis of SS and may allow the clinician to avoid other invasive and expensive
investigations. In this case, the alternating nature of the torticollis seemed to exclude virtually all other diagnoses mentioned in Table 1. This aspect might be considered an important indication of the diagnosis in our case.

REFERENCES