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## Non epileptic paroxysmal disorder of infancy: Sandifer syndrome

### To the Editor,

Non-epileptic paroxysmal disorders (NEPD) are repetitive conditions which share similar characteristics with epileptic seizures including change in consciousness, cyanosis and tonic and clonic convulsions. NEPD is found in 20-25% of the patients who are planned to be evaluated further with a suspicion of seizure (1). Age, history and assistive examinations including video electroencephalography (EEG) are helpful in differentiation from epilepsy (2). Patients who can not be diagnosed accurately may be exposed to unnecessary tests and treatment. Sandifer syndrome is a NEPD which is classically characterized with extension in the neck, bending of the head to one side and accompanying opisthotonus. A 9-month-old male patient who was diagnosed with Sandifer syndrome was presented to draw attention to the differential diagnosis of epilepsy and emphasize the clinical properties of the disease.

A 9-month-old male patient presented to our outpatient clinic with a complaint of contraction of the head and neck to the left side. The contractions occurred when the patient was awake, each contraction lasted for 1-2 seconds and in clusters which lasted 10-15 seconds. During this time, his looks became dull and his facial expression changed. It was learned that his contractions started when he was 4,5 months old for the first time and increased in the last 15 days. He was internalized with a prediagnosis of benign infantile myoclonic epilepsy. He had been using syrups containing the active ingredient including sodium alginate, magnesium alginate and domperidone for four months with a diagnosis of gastroesophageal reflux disease (GER). No other significant finding was present in his personal history. He had no familial history of epilepsy and her parents had no consanguineous marriage.

On physical examination, his body weight was between the 75<sup>th</sup> and 90<sup>th</sup> percentiles and his height and head circumference were between the 25<sup>th</sup> and 50<sup>th</sup> percentile. His neurological examination and examination of the other systems were found to be normal. His complete blood count, hepatic and renal function tests, serum electrolytes and complete urinalysis were found to be normal. Acylcarnitine analysis by TANDEM mass spectrometry, amino acid profile and screening of metabolic diseases in the urine were found to be normal. Sleep and wake EEG recordings and brain magnetic resonance imaging were found to be normal.

For differential diagnosis of epilepsy 24-hour video EEG was performed. On video recording, contractions of the head and neck to the left side were observed as described and epileptic finding could not be found on simultaneous EEG. When the patient's history was deepened further, it was learned that the contractions mostly occurred during feeding or just after feeding. When the patient was consulted to the division of Pediatric Gastroenterology-Hepatology, it was observed from the records that feeding was done rapidly and the amounts of meals were excessive. Upper gastrointestinal endoscopy was performed for the diagnosis of gastroesophageal reflux. It was observed that the gastric cardia sphincter was open throughout the procedure. Histopathological examination of the esophageal biopsy was compatible with reflux esophagitis. A diagnosis of Sandifer syndrome was made with the present history and clinical and laboratory findings. The findings disappeared in one week in the patient who was started on proton pump inhibitor treatment. The patient who is being followed up for 6 months has no contractions.

Sandifer syndrome was described by Kinsbourne in 1964 for the first time in five children in whom different

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head and neck movements related with hiatus hernia were observed. The disease was named after the neurologist Paul Sandifer who was responsible of supervision of two of these patients (3). Although Sandifer syndrome was described in association with hiatus hernia initially, it is more frequently related with GER. During the episodes, tonic extension in the neck, spastic torticollis and accompanying truncal dystonia are observed. Torticollis may be observed without truncal dystonia (4). Our patient also had intermittent spasmodic torticollis without truncal dystonia.

The onset usually occurs during the infancy or early childhood, but adult cases have also been reported (5). Although the prevalence is not known, it has been reported to occur in 1-8% of GER cases (6,7). In a study in which 134 NEPD cases were evaluated, Sandifer syndrome was found in the etiology in four of a total of 26 infants (8).

Initially, the disturbance caused by the transfer of the gastric acid into the esophagus was blamed, but studies could not find a relation between the grade of reflux and the severity of Sandifer syndrome (9). According to an assumption, it is thought that a vagal reflex which is named vagocervical or esophagocervical reflex and the center of which is the nucleus tractus solitarius is responsible of the clinical findings (10).

In the neonatal period and early infancy, apnea, tonic contraction of the trunk and circular movements in the extremities may be observed (9). Sandifer syndrome is frequently associated with normal neurological examination especially in infants. Reflux of the ingested food or vomiting may not always be present. Absence of change in consciousness, occurrence of the episode during feeding or just after feeding, torticollis being intermittent and absence of epileptic findings on EEG are helpful in the differential diagnosis (4). No improvement in the findings of the patients who are started on epilepsy treatment despite all these is one of the significant points. Rapid feeding, excessive amount of feeding, relation of the contractions with feeding, intermittent occurrence of the contractions and normal video EEG were helpful in the diagnosis in our patient. On upper gastrointestinal endoscopy, findings including an open cardia sphincter and an esophageal biopsy result compatible with reflux esophagitis supported the diagnosis.

Sandifer syndrome may also be observed in individuals with metabolic disease or central nervous system disease. Here, GER is responsible of the picture, but not the brain damage in the child. Since the symptoms are associated with the underlying central nervous system disease, the diagnosis may be delayed (11,12).

Medical or surgical treatment of GER which is the actual cause of the syndrome is sufficient to eliminate

the symptoms. Especially in presence of hiatus hernia, Nissen funduplication may be needed (7). In our patient, domperidone and anti-acid treatments which prevent reflux from the gastroesophageal junction could not provide clinical improvement and the findings were eliminated with proton pump inhibitor treatment.

Conclusively, Sandifer syndrome in which detailed history is helpful in the diagnosis is a NEPD with good prognosis (13). Early diagnosis in this syndrome increases the quality of life in children and allows efficient response by preventing unnecessary tests and treatment.

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## References

- Desai P, Talwar D. Nonepileptic events in normal and neurologically handicapped children: a video-EEG study. *Pediatr Neurol* 1992; 8(2): 127-129.
- Paolicchi JM. The spectrum of nonepileptic events in children. *Epilepsia* 2002; 43(Suppl 3): 60-64.
- Kinsbourne M. Hiatus hernia with contortions of the neck. *Lancet* 1964; 1(7342): 1058-1061.
- Köse G. Epilepsi ile karışabilecek paroksizmal durumlar. *Turkiye Klinikleri J Pediatr Sci* 2008; 4(4): 49-54.
- Rana AQ, Yousuf MS, Joian S. A chronic case of adult-onset Sandifer syndrome. *Neurol Sci* 2013; 34(3): 405-406.
- Shepherd RW, Wren J, Evans S, Lander M, Ong TH. Gastroesophageal reflux in children. Clinical profile, course and outcome with active therapy in 126 cases. *Clin Pediatr (Phila)* 1987; 26(2): 55-60.
- Lehwald N, Krausch M, Franke C, Assmann B, Adam R, Knoefel WT. Sandifer syndrome--a multidisciplinary diagnostic and therapeutic challenge. *Eur J Pediatr Surg* 2007; 17(3): 203-206.
- Kotagal P, Costa M, Wyllie E, Wolgamuth B. Paroxysmal nonepileptic events in children and adolescents. *Pediatrics* 2002; 110(4): 46.
- Mandel H, Tirosh E, Berant M. Sandifer syndrome reconsidered. *Acta Paediatr Scand* 1989; 78(5): 797-799.
- Cerimagic D, Ivkic G, Bilic E. Neuroanatomical basis of Sandifer's syndrome: a new vagal reflex? *Med Hypotheses* 2008; 70(5): 957-961.
- Sommer A. Occurrence of the Sandifer complex in the Brachmann-de Lange syndrome. *Am J Med Genet* 1993; 47(7): 1026-1028.
- Sondheimer JM, Morris BA. Gastroesophageal reflux among severely retarded children. *J Pediatr* 1979; 94(5): 710-714.
- Smallpiece CJ, Deverall PB. Sandifer's syndrome: a new cause. *Thorax* 1982; 37(8): 634-635.