

# Sandifer syndrome misdiagnosed as refractory partial seizures in an adult

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**ABSTRACT** – We report a 27 year-old man with intellectual disability and no prior history of seizures who presented with episodes of abdominal pain, head/eye version and unresponsiveness that were misdiagnosed and treated as partial seizures. Associated vomiting and haematemesis led to the correct diagnosis and treatment of reflux oesophagitis. The episodes immediately resolved and a diagnosis of Sandifer syndrome was made. This is only the second report of Sandifer syndrome in adult, a movement disorder of unknown mechanism that occurs almost exclusively in young children, often misdiagnosed as epilepsy or episodic dystonia. (Published with videosequences)

**KEY WORDS:** Sandifer syndrome, adult, partial seizure, gastro-oesophageal reflux, oesophagitis

We report on a 27 year-old man with mild-moderate intellectual disability and no prior or family history of seizures, who presented with a two-day history of epigastric pain and episodes of apparent unresponsiveness, head and eye deviation to the left, and vomiting. He was admitted to hospital where several typical attacks were witnessed by staff, and an episode was captured on video (see video). Consciousness was difficult to determine but he appeared to speak during some episodes. The events continued over the next five days, occurring 1-5 times a day and lasting 30 seconds to 20 minutes. The attacks were refractory to treatment with clonazepam, phenytoin and carbamazepine. Two scalp EEG recordings, including one following the attack captured on video, did not reveal epileptiform discharges. Magnetic resonance brain

imaging was normal. Five days after admission he vomited a small amount of blood. Omeprazole and metoclopramide were added and the response was dramatic, with no further attacks after two days of treatment. Subsequent gastroscopy showed moderate-severe reflux oesophagitis (grade 3). Sandifer syndrome was diagnosed, antiepileptic medications were ceased, and there were no further attacks at nine months follow-up.

Sandifer syndrome is an uncommon but well recognised disorder in infants and young children manifesting with abnormal postures or spasms, typically with torticollis or retrocollis, secondary to gastro-oesophageal reflux [1, 2]. It presents typically between age two months and five years, with onset between the newborn period and age 16 years described [4]. The episodic nature of attacks often results



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in misdiagnosis as paroxysmal dystonia or epilepsy. Since the original description by Kinsbourne in 1964 [5], the pathophysiology of Sandifer syndrome remains unclear. Postulated mechanisms include enhanced clearance of acid from the lower oesophagus by the dystonic posturing giving relief of symptoms [6], and triggering of brainstem release phenomena by the pain of oesophagitis [2], neither of which are proven.

There is only one report of Sandifer syndrome in an adult [3], a 58 year-old woman who presented with “episodic cervical dystonia” characterised by attacks of epigastric and retrosternal discomfort, eye deviation, facial grimacing, retrocollis and clonic neck spasms. Barium swallow revealed gastro-oesophageal reflux and the episodes resolved with cimetidine.

Our patient was misdiagnosed with a new-onset epilepsy because he had several features seen in patients with partial seizures, including epigastric discomfort, vomiting, head version and asymmetric tonic posturing. Although absence of clonic jerks, lack of response to antiepileptic medications, and normal EEG studies do not necessarily exclude partial seizures, awareness of Sandifer syndrome by paediatric members of our epilepsy team, and the presence in this patient of abdominal pain and vomiting (with blood) before and between attacks, were clues to the correct diagnosis. Sandifer syndrome should be considered in an intellectually disabled adult with suspected seizures or movement disorder, in whom there is associated abdominal pain, vomiting and haematemesis. □



### Video sequence caption

Fortuitous video recording of a typical episode, captured while setting up a routine EEG. The video shows the patient having head and eye deviation to the left, neck extension, occasional blinks, and some semi-voluntary movements. The episode lasted about 90 seconds. No sound or EEG was recorded at the time. The immediate postictal EEG showed no abnormality.

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