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A Sandifer Syndrome Case with 6 Year of Evolution

ABSTRACT

We describe a male pediatric patient showing dystonic head posture and occasional dystonic movements on the upper limbs since his 24 month of age. Those movements are associated with "burning sensation in the throat", vomiting without nausea and dental erosion. He was treated with antiepileptics, neuroleptics, dopaminergics and proton pump inhibitors, without any evident results. Six years later a proper diagnosis was made and the proper treatment was indicated. In that period of time the patient was recurrently admitted on the hospital and submitted to many lab and other diagnostics tests. The disorder in question is very uncommon and as this particular case was formerly misdiagnosed and hence mistreated, it might be on the interest to the scientific community, health care professionals and medical science students.

Keywords: Gastroesophageal Reflux Disease; Focal Dystonia; Sandifer Syndrome; Long Evolution Time.

INTRODUCTION

The patient is male, 13 years old and the second sibling of a two member offspring from young parents not related by blood and native from different geographic areas of the country, no prenatal or perinatal facts was remarkable and he had an apparently normal psychomotor development.

He start having episodes of prolonged fast in 2007 with 22 months old, after that he was presented with repeating movements of the neck to both sides (with preference to the left) with frequent flexion and extension and also occasional involvement on the upper limbs. These movements indeed impair walking and standing. same clinical findings prevailed since 24 month of age, characterized by involuntary movements, unnoticed at wake up and shown with increasing intensity immediately upon eating (with a major increase by liquid intake than solids). At the beginning of the day, the duration of the movements is usually 30 s-1 min, followed by vomiting (without nausea) and cessation of all symptoms, increasing the duration of the onsets with the day and having a peak at night with upper limb involvement. At every onset, the patient referred a "burning sensation on the throat", with frequent burping and heartburn sensation. He presents frequents dental cavities and erosion.

The patient did not presented alterations at general physical examination but in neurological examination we reported a right handed patient, mindful, oriented on time, space and person, who answers the questionnaire with clear and coherent language. Immediate, recent and late memories not seem affected. The involuntary

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lateralization movements of the neck may affect the ability to stand, depending of the intensity. Increased support surface on standing and walking. Simple Romberg maneuver and sensitize Romberg maneuver were impossible to perform because the affected standing. Movement disorder characterized sustained, patterned, and repetitive muscle contractions of opposing muscles of the neck and occasional the upper limbs always in relation with eating.

Several superior airway endoscopy studies showed chronic superficial gastritis, esophagitis and small sized hiatal hernia. 24-hour esophageal-pH monitoring shows that the presence of moderated acidity GERD matching with the paroxystic neck movement's disorder.

EEG, head CT, and MRI were unremarkable. No Kayser-Fleischer rings were noted, and a 24-hour urinary copper was normal, mitochondrial disease studies was normal. Gluten free diet was indicated to treat a suspected Celiac disease or gluten-sensitive enteropathy rules out as the symptoms did not stop after 6 month of the diet.

BACKGROUND

Dystonia is currently defined as a neurological syndrome characterized by involuntary, sustained, patterned, and often repetitive muscle contractions of opposing muscles, causing twisting movements or abnormal postures [1]. One of the earliest descriptions of dystonia was provided by Gowers in 1888, who coined the term *tetanoid chorea* to describe the movement disorder in two siblings later found to have Wilson's disease [2]. The term *dystonia musculorum deformans*, coined by Oppenheim in 1911, was criticized because fluctuating muscle tone was not necessarily characteristic of the disorder, the term *musculorum* incorrectly implied that the involuntary movement was due to a muscle disorder, and not all patients became deformed. Until recently, the term *torsion dystonia* has been used in the literature, but since *torsion* is part of the definition of dystonia, the term torsion dystonia seems redundant and, hence, the simple term dystonia is preferred [1]. Cervical dystonia is the most common form of focal dystonia encountered in a movement disorders clinic [2]. In addition to turning (torticollis), flexing (anterocollis), or extending (retrocollis) of the neck, the head might be shifted forward or off the midline to either side or tilted toward one shoulder [3].

Sandifer syndrome (SS) named after Dr. Paul Sandifer, was first described by him in 1964 and is a rare complication of gastroesophageal reflux disease (GERD) [4], most commonly seen in infants and children [5]. Very rarely SS has been recognized in adults [1-4,6,7].

This syndrome is a GERD, with or without a hiatal hernia, associated with dystonic movements of the head and neck and, at times, abnormal posturing of the body, including opisthotonos. Recurrent paroxysmal posturing are typically associated with feedings, although they may persist postprandial. Barium swallow studies, esophagoscopy, and pH probes are used to document the reflux and hernia. Intermittent stiff tonic postures and episodic crying and discomfort may suggest seizures. Symptoms beginning shortly after feeding would suggest the diagnoses. Symptoms usually resolve after appropriate gastrointestinal intervention [8]. It is probably misdiagnosed as epileptic seizures or paroxysmal dyskinesia [7].

The pathogenic mechanisms are unclear, although a dysfunction of the lower esophagus is thought to be the most important precipitating factor [1-8]. The role of GERD in SS is well established in the literature. Among the mechanisms for neck muscle contraction in SS, it was postulated that neck posturing might favor clearance of acid from the lower esophagus providing relief from discomfort caused by acid reflux [9]. Other authors consider the abnormal posture as a result from a reflex mechanism that aims to protect the air passages from reflux material [7].

Diagnosis may be difficult and often requires multiple evaluations by specialists. Infants are often misdiagnosed due to paroxysmal neurobehavior or focal epilepsy and receive unnecessary medication and treatment [10,11]. Analysis of the torticollis may help establish a suitable strategy for diagnosis [12]. (3) (4) (5)

CASE DISCUSSION

Diagnostics

The diagnoses on this case was made based on the patient background and collected clinical data horugh the years of evolution.

- **Background:**
- The age of onset is ± 2 years old
- The preceding symptoms of the Movement disorder onset match with GERD.
- **Clinical findings:**
- Presence of a movement disorder characterized by

sustained, patterned, and repetitive muscle contractions of opposing muscles of the neck and occasional involvement on the upper limbs associated with GERD episodes upon eating (with a major increase by liquid intake than solids).

- Symptoms resolve after appropriate GERD management (see treatment).

- **Complementary tests:**

- Esophagus-stomach-duodenum barium contrasted Rx study who shows IV Grade GERD

- Upper GI endoscopy showing II grade esophagitis (possibly due to GERD) and small size hiatal hernia (alteration also on the physiopathology of GERD)

- 24-h esophageal pH monitoring: Presence of moderated acidity GERD matching with the paroxystic neck movement's disorder.

Treatment

The patient was treated for the movement disorder with clonazepam, valproic acid, carbamazepine, L-dopa, bromocriptine, trihexifenidil, risperidone, thioridazine and also with non-drug treatment such as cognitive behavioral therapy, physiotherapy, acupuncture, ozone therapy. In all cases without results. Even a gluten free diet was indicated to treat a suspected Celiac disease or gluten-sensitive enteropathy.

The medial management of GERD include several cycles of domperidone, simethicone and omeprazole. In all cases without result.

In December 20 of 2013 our patient had an anti-reflux surgical intervention (fundoplication surgery). After that the movement disorder and the GERD had completely stopped and no problems have recurred

Concluding Remarks

These is an atypical case because medical GERD management is usually successful with good prognosis and our patient required surgical intervention. Precisely for that fact many medical doctors hesitated and rejected SS as Nosological Diagnosis and delayed the proper treatment.

A remarkable fact about this case is that six years after the onset of the symptoms a proper diagnosis was made. In that period of time the patient was recurrently admitted on the hospital and submitted to many lab and other expensive diagnostics tests increasing hospital costs, on the oher hand social rejection and school absence affect the parent's psychology and their jobs, like it has also been described in the literature.

The misdiagnosis could imply medical malpractice. Our patient at early ages was submitted to treatment with trihexifenidil, risperidone, thioridazine, drugs having severe side effects.

The disorder in question is very uncommon and as this particular case was formerly misdiagnosed and hence mistreated, it might be on the interest to the scientific community, health care professionals and medical science students. Sandifer Syndrome do not pose a severe risk for the patient's live but indeed heavily affect its quality.

Authors' Contributions Statement

Carlos Maragoto-Rizo was involved in directing the research project, collection of the main data and writing of the definitive manuscript. Also he is the head of the multidisciplinary team who attended the patient since 2012. Arturo Rodríguez-López was involved in the conception and execution of the research project and writing of the first draft of the Manuscript. Irene González-González and Carlos A. Dávla-Gómez were involved in execution of the bibliography research and organization of the supplementary data. Hector Vera-Cuesta, Lazaro Gómez-Fernández, Maria de los Ángeles Ortega, Reinaldo Galvizo-Sánchez and Nancy Pavón Fuentes were involved on the review of the data and the manuscript and also are part of the multidisciplinary team who attends the patient since 2012.

Conflicts of Interest Statement

None to be reported

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