Letters to the Editor

The presentation of a case of Moebius and Poland syndromes associated to prenatal exposure to misoprostol has not been reported. However, this combination of two congenital defects has been reported as being associated to another vascular disruptor (such as cocaine). The presentation of these two defects in a same patient provides evidence that both pathologies present similar mechanisms, which are probably those of a vascular disruption.

Defects due to a vascular disruption are structural disturbances in development produced by vascular problems, such as severe intermittent vasoconstriction, abnormal vessel regression during the remodelling of the vascular system, arterial thrombosis or any other phenomenon that produces a lack of O₂.

The aetiology of Moebius and Poland syndromes is unknown; a possible vascular origin has been suggested for the two pathologies. Hypotheses suggest a transient ischemia, particularly in the vertebral arteries. Premature obstruction or regression of the terminal arteries of the trigeminal (V) nerve and/or delayed formation of the vertebral basilar system could lead to anomalies in cranial nerve development. In children with Moebius syndrome, cerebral necrosis has been reported together with capillary telangiectasia in the midbrain and bridge. Disruption of the subclavian artery occurs around week 6 of gestation and is related to Moebius—Poland syndrome; this same phenomenon is related to transversal limb defects and arthrogryposis. The disruption phenomenon can be secondary to a blood flow interruption secondary to an arterial spasm during the sensitive embryonic phase.

References


Oesophageal aperistalsis in a patient with myasthenia gravis with dysphagia as a symptom of onset

Aperistalsis esofágica en un paciente con miastenia gravis con disfagia como síntoma de presentación

Sir,

Dysphagia as an isolated symptom in myasthenia gravis (MG) is highly unusual. In such cases, diagnosis may be difficult. Oesophageal manometry studies in patients with MG may reveal gradual deterioration in the amplitude of the pharyngeal contractions due to involvement of the neuromuscular transmission in the upper oesophageal sphincter (UES), formed by striate muscle. Unexpectedly, there may also be dysfunction in the oesophageal motility in segments formed solely by smooth muscle. We report here on the case of a female patient diagnosed as having MG who debuted with progressive upper dysphagia and in whom the manometry study confirmed oesophageal aperistalsis with severe participation of smooth muscle and respecting the striate muscle.

Female patient, 79 years of age, without any personal or family background of note, with a history of sustained progressive dysphagia over 2 months of, essentially for swallowing liquids. She did not report any tendency to tire

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easily. She was initially admitted to the digestive department where local pathology was ruled out by means of gastroscopy, laryngoscopy, and a CT scan of the chest and abdomen. Her condition was severe as she had lost more than 10 kg since the onset of her condition. She did not present any language disorder or dyspnoea. She did not mention any other symptoms. During examination, the cranial pairs were normal. Nauseous reflex was present. Her palatal arch was normal and symmetrical. There was no double vision or drooping eyelids. Nor was there any atrophy or fasciculations on the tongue. She showed slight weakness in cervical flexion, scoring 4/5, without a clear tendency to tire easily, and the girdle musculature was normal. There were no sensorial or co-ordination disorders. Her reflexes were normal and the cutaneous planter reflexes were positive.

Biochemistry tests included thyroid hormones, lactate, creatine kinase, myoglobin, cortisol, proteinogram, tumour markers and an auto-immune study were normal.4 During the electrophysiological study, while performing repetitive stimulation of the right spinal nerve with retrocoele-dostayoideal stimulus and pickup in the trapezium muscle at 3 Hz and 20 Hz, a pathological decrease was observed after 3 min of tetanizing stimulation. The EMG of the deltoid muscles, orbicular muscles of the eyes, right masseter, and orbicular muscles in the lips showed no spontaneous activity and the motor unit potentials (MUP) were normal. An MR image of the brain was normal. Anti-AChR antibodies were positive (30.40 nmol/L, normal < 0.40 nmol/L). No thymoma was seen on the chest CT.

The oesophageal manometry study performed revealed normal pressure (11.6 mmHg), in the lower oesophageal sphincter (LES) with good relaxation on swallowing (residual pressure: −4.9 mmHg). Examination of the body revealed the absence of peristaltic waves on swallowing, with normal, not elevated, baseline pressure. Pressure in the UES was somewhat low (13.8 mmHg), with good relaxation on swallowing co-ordinated with pharyngeal contraction. The amplitude of the pharyngeal contraction was normal. In short, there was a severe motor alteration with oesophageal aperistalsis with a normal LES sphincter and without any pharyngo-oesophageal motor alteration.

With a diagnosis of MG treatment was begun with pyridostigmine (60 mg every 6 h) with evident improvement in dysphagia for both solids and liquids.

The patient is currently stable, without presenting any new relapses and controlled with pyridostigmine (240 mg/day) and prednisone (15 mg/day).

Dysphagia is a frequent symptom in patients with MG. It is commonly accompanied by other bulbar symptoms, such as a nasal tone of voice or dyspnoea, or visual symptoms. What is not so common is for it to present in isolation as the only symptom, a circumstance occurring in only 6% of patients with this illness.6,7 The study by Huang et al. showed that 96% of patients with MG and dysphagia presented alterations in oesophageal manometry, with involvement of both striate and, surprisingly, smooth muscle. The medial and lower third of the oesophagus is made up of smooth muscle, whereas the UES is under voluntary muscle control. The upper third contains the transition from striate to smooth musculature. An explanation for the present findings in smooth muscle might be as a consequence of the primary involvement of the voluntary muscles of the oesophagus, which would produce a diminished afferent feedback from the most distal portion of the oesophagus. Another hypothesis is that there could be a transmission block between the neurons on the dorsal vagus nerve and the neurons of the myenteric oesophageal plexus. This transmission is mediated by nicotinic ACh channels, but of the ganglionar type, i.e., different from the also nicotinic ACh receptors on the motor plaques.8 Crossover reactivity between these two types of receptors is a little known phenomenon. In the paper by Liabrés et al., which reported on 2 patients with dysphagia as an isolated symptom, in both of which a manometric study had been carried out, they also found data on smooth muscle involvement with low-amplitude peristaltic contractions, although not as severe as in our patient, where aperistalsis was shown. In oesophageal aperistalsis, it is important to consider a diagnosis of achalasia, which also courses with increased muscle tone in the LES or absence of relaxation in this sphincter on swallowing. In our patient, the LES tone was slightly diminished and relaxation was correct. The truly surprising aspect of this case is that the manometry did not reveal any striking pathology of the striate musculature in the pharynx but a severe involvement of smooth muscle. This result might, in principle, seem paradoxical.9 The number of swallows in the manometric study, eight, might possibly have been insufficient to trigger fatigability in the striate musculature. The findings in the manometric study may help in diagnosis and could represent a useful tool for the diagnosis of MG, particularly in cases where the response to anticholinesterases is doubtful.10

References

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