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Defecation reflex seizures: a case report with long-term VEEG monitoring, neuroimaging and comprehensive epilepsy evaluation

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ABSTRACT – Reflex seizures are consistently elicited by a specific afferent sensory stimulus or an activity undertaken by the patient. Among many known stimuli, defecation has rarely been reported. We describe the case of a child with reflex seizures triggered by defecation, considering the diagnostic challenge, epilepsy evaluation with video-EEG monitoring, as well as impact on neuropsychology, behaviour and quality of life. The child was a 10-year-old boy with seizure onset at age four with epilepsy diagnosis established one and a half years later. Seizures were focal with impaired awareness triggered by defecation. Video-EEG and structural and functional neuroimaging were performed and all pointed to the left temporal region. The patient became seizure-free with carbamazepine and valproic acid. Neuropsychological and quality of life assessments suggested global impairment, both before and after seizure control. This is the third case of epilepsy induced by defecation reported in the literature. The rarity of this entity may be a diagnostic challenge and postpone specific treatment. Reporting of cases of defecation reflex epilepsy may provide a better understanding of its physiopathology and optimize effective treatment, avoiding cognitive, behavioural and poor social consequences. [*Published with video sequence*]

Key words: defecation reflex seizures, reflex epilepsy, video-EEG, SPECT, PET

VIDEO ONLINE

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Reflex seizures are consistently elicited by a specific afferent sensory stimulus or an activity undertaken by the patient (Blume *et al.*, 2001). The stimulus may be somatosensorial, proprioceptive, praxic or cognitive (Gastaut and Tassinari, 1966; Striano *et al.*, 1993). It is estimated that in patients with epilepsy, reflex epilepsies have a prevalence of 5-6% (Okudan and Özkara, 2018) among which defecation-induced seizures are considered rare. Defecation is usually therefore not mentioned as a trigger for reflex seizures in reviews on reflex epilepsies. We are aware of three previous reports; in two of them, seizures were elicited not specifically by defecation but also by other somatosensorial stimuli (Schubert and Cracco, 1992; Harbord and Mitchell, 2002; Higuchi *et al.*, 2011). This study describes the case of a child with reflex seizures triggered by defecation, focusing on the diagnostic challenge, epilepsy evaluation, and impact on neuropsychology, behaviour and quality of life (QOL).

Case study

A left-handed, 10-year-old boy reached developmental milestones prior to epilepsy onset. From age four, he presented with focal seizures with impaired awareness and hypotonia during defecation, following both Valsalva manoeuvre and evacuation completeness. Seizures lasted about one minute, sometimes followed by drowsiness for one hour. This occurred three or four times a week. Seizures did not occur for every evacuation, although all seizures were associated with the process of defecation. At first, cardiologists evaluated the patient and propranolol was prescribed despite normal cardiological investigation. At the age of five and a half years, presenting with the same symptoms, a neuropediatric team evaluated the patient and no abnormalities were found either on neurological examination, EEG or brain MRI. VEEG monitoring was performed but no events were recorded. The EEG showed interictal regional slow activity and scarce sharp waves over the left temporal lobe. On the basis of seizure manifestation and interictal EEG findings, he was diagnosed as epileptic with reflex seizures and was started on antiepileptic medication (AEM). Carbamazepine (CBZ) and phenobarbital (PB) were added with no seizure control. Meanwhile, the boy had been presenting with learning disabilities besides seizures. When he turned eight, further evaluation was performed including MRI, neuropsychological testing, QOL evaluation, a second four-day period of VEEG monitoring, SPECT, and PET. No abnormalities were detected on MRI. The WISC-IV indicated mild intellectual disability, with an IQ of 58. A score of 38 was ascertained for the *Autoquestionnaire Qualité de Vie Enfant Imagé* (AUQEI), based on auto perception of QOL (the cut-off value being 48, below which qualifies as low QOL). Its main domains are autonomy, leisure, functions and family (Manificat *et al.*, 1997). For the parent-rated epilepsy-specific health-related quality of life (HRQL) instrument, QVCE-50 (Maia Filho *et al.*, 2005, 2007), a score of 123.5 was obtained. Both instruments therefore showed unfavourable results. The Zarit Burden Interview, a caregiver self-report measure, showed a score within normal limits, of 13 (Scazufca, 2002). VEEG monitoring showed slowing and repetitive sharp waves over the left temporal region, involving ipsilateral frontocentral regions during sleep recording (*figure 1A*). During properly recorded defecation, the child presented with an usual focal with impaired awareness seizure, described at home as: behavioural arrest and impaired awareness, right head deviation, left arm automatism, postictal left hand nose wiping, confusion, and speech arrest (*see video sequence*). All these data together suggest a dominant temporal lobe seizure. Ictal EEG registered a focal seizure with left temporal onset, characterized by rhythmic theta activity evolving into delta over the ipsilateral frontal region, followed by homologous contralateral areas (*figure 1B*). Postictal slowing was detected in the left cerebral hemisphere (*figure 1C*). Interictal SPECT with ^{99m}Tc-ethyl cysteinate dimer showed subtle left anterior and basal temporal lobe hypoperfusion (*figure 2A-D*). ^{18}F-fludeoxyglucose PET/CT showed subtle hypometabolism in the left temporal lobe and cerebellum (*figure 2E-G*). Defecation seizure was diagnosed as a reflex seizure with lateralizing and localizing signs pointing to the left temporal region. The patient was taking CBZ (16 mg/kg/daily) and valproic acid (VPA) (20 mg/kg/daily) was added, after which he became seizure-free. Eighteen months later, a clinical revaluation was performed. Learning disability had increased with symptoms of disruptive behaviour. EEG showed left temporal slow waves during a hyperventilation manoeuvre. WISC IV revealed a mild intellectual disability score. AUQEI and QVCE50 were unchanged. The new Zarit Burden Interview was performed showing moderate caregiver burden, with a score of 31.

Discussion

Since the presented seizures were all triggered by defecation, they were considered as a reflex epilepsy (Higuchi *et al.*, 2011). The rarity of this entity may be a diagnostic challenge and postpone specific treatment, which was the case in our patient. The scarcity of reports on somatosensorial stimuli-triggered seizures, especially defecation-related seizures, can lead to misdiagnosis, inappropriately attributing symptoms to

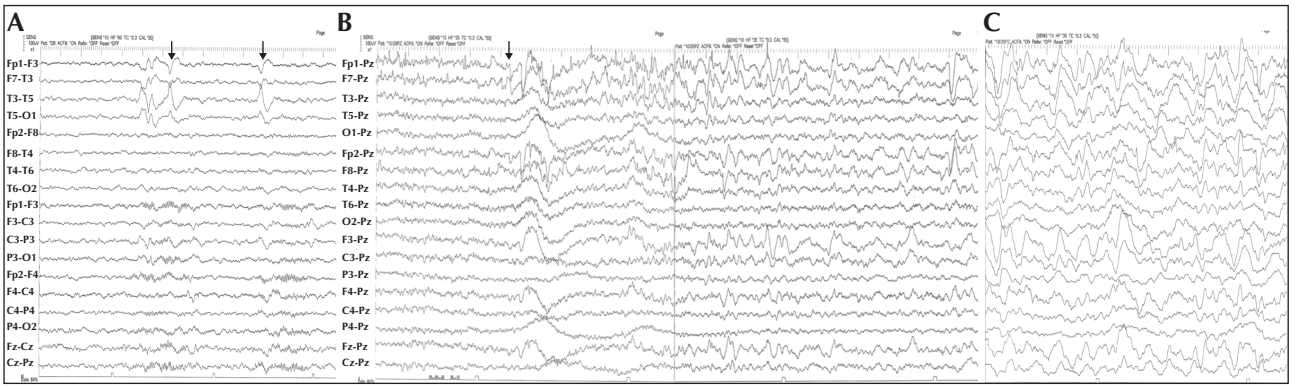

Figure 1. (A) Interictal EEG during sleep showing repetitive left temporal sharp waves. (B) Ictal EEG showing a focal seizure with left temporal onset, characterized by rhythmic theta activity evolving into delta activity in the ipsilateral frontal region, followed by homologous contralateral areas. (C) Postictal slowing, more accentuated in the left cerebral hemisphere.

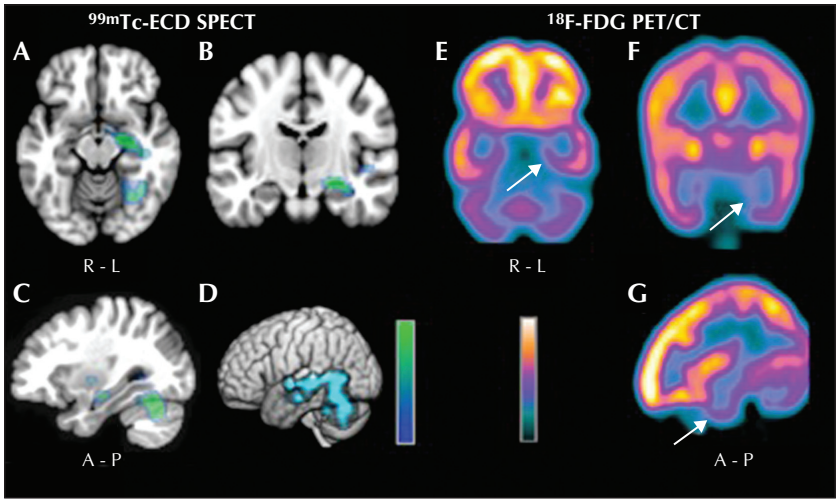

Figure 2. Interictal ^{99m}Tc-ECD SPECT and ^{18}F-FDG PET/CT of the brain. Statistical parametric mapping (SPM) of interictal SPECT showing reduced regional cerebral blood flow (rCBF; blue green colour scale) on the mesial and basal left temporal lobe and cerebellum, on transversal (A), coronal (B), and sagittal (C) slices, and 3D reconstruction (D). ^{18}F-FDG PET/CT shows glycolytic hypometabolism (white arrows) in the left temporal lobe, on transversal (E), coronal (F), and sagittal (G) slices.

syncope (Schubert and Cracco, 1992; Harbord and Mitchell, 2002).

In the only case of reflex seizures previously reported with defecation as the unique stimulus-inducing factor, a latency of one or two minutes is described between the passage of an intestinal action and the beginning of a seizure. The authors used this latency period to differentiate seizures from syncope reactions, which occur immediately, without a delay (Harbord and Mitchell, 2002). However, in our case and in the one described by Higuchi *et al*., 2011, the precipitating event and the seizure recorded on VEEG occurred almost simultaneously, suggesting that latency may not be an unequivocal argument for differential diagnosis between syncope and reflex seizure (Higuchi *et al*., 2011).

All these authors agree that this epilepsy could be considered as focal (Harbord and Mitchell, 2002; Higuchi *et al*., 2011). As in our case, there seems to be a convergence of irritative, symptomatogenic and ictal onset zones, pointing to a dominant temporal region (Harbord and Mitchell, 2002). MRI was normal in all reported cases (Harbord and Mitchell, 2002; Higuchi *et al*., 2011). As structural data showed no additional findings, neurofunctional imaging studies such as SPECT and PET were then performed, with convergent findings regarding ictal and interictal epileptiform activity. These localizing and lateralizing data were similar to those presented by Harbord and Mitchell, 2002, in describing the other case of defecation-induced seizures. The authors suggested that the most likely physiological induction pathway would be through

the proprioception of somatosensory input into the temporal lobe in response to anal dilatation or rectal evacuation (Harbord and Mitchell, 2002).

There are relatively few data regarding cerebral processing of anorectal function. Based on an anorectal fMRI study, the subjective sensation of discomfort that increased during repeated rectal distension caused activation in the anterior cingulate gyrus, insula, thalamus and secondary somatosensory cortex. Perception of rectal urge and discomfort activated the same cerebral regions with differing intensity. Voluntary contraction of the anal sphincter led to activation of the motor cortex and increased activity in the supplementary motor cortex and the insula (Bittorf *et al*., 2006). Another fMRI study has shown that rectal pain induced by pressure from an inflated balloon activated the insular cortex, as well as the extrastriate visual cortex, prefrontal cortex, and anterior cingulate gyrus (Baciu *et al*., 1999). Besides, Penfield and Faulk described a 28-year-old woman who had the feeling gas was passing through the rectum during automotor seizures; intraoperative monitoring indicated a right-sided insular seizure focus (Penfield and Faulk, 1955). Thus, the temporal or insular lobe has been associated with autonomic seizures based on EEG recordings or direct visual observation (Baumgartner *et al*., 2001; Mazzola *et al*., 2017).

Patients with ictal flatulence and others with ictal urge to defecate have been rarely reported (Koubeissi *et al*., 2005; Strzelczyk *et al*., 2010). According to the authors, the autonomic seizures that presented with a temporal ictal onset on surface EEG were associated with other autonomic signs and symptoms, pointing to insular involvement. Ictal flatulence was then suggested as an ictal localizing sign for temporal and/or insular lobe involvement and could be valuable in localizing the epileptogenic zone in non-lesional cases (Strzelczyk *et al*., 2010). The structural and functional connectivity between the temporal lobe and insula may suggest that these are symptomatogenic areas regarding the clinical presentation of defecation reflex seizures, as in our patient.

In both cases described by Strzelczyk *et al*. (2010), ictal flatulence was associated with a seizure pattern in the temporal lobe of the dominant hemisphere. According to the literature, ictal flatulence seems to have no lateralizing value (Strzelczyk *et al*., 2010). On the other hand, ictal urge to defecate has been reported to be associated with seizure onset in the non-dominant hemisphere (Koubeissi *et al*., 2005). This is concordant with other reports of central processing of visceral sensations in related somatosensory or autonomic epilepsies, such as in epileptic orgasmic auras and eating epilepsy, in which mostly interictal or ictal patterns resemble non-dominant temporal lobe epilepsy (Aguglia and Tinuper, 1983; Nagaraja and Chand, 1984; Koul *et al*., 1989; Baumgartner *et al*., 2001; Janszky *et al*., 2002).

The non-dominant hemisphere is reported to be mostly, but not always, associated with micturition-induced seizures, while there is much evidence for onset in the left hemisphere in defecation-induced seizures (Harbord and Mitchell, 2002; Glass *et al*., 2006; Okumura *et al*., 2007; Rathore *et al*., 2008; Rho, 2008; Higuchi *et al*., 2011).

Moreover, authors who reported isolated micturition reflex seizures, as well as those who described an attack associated with defecation and urination in the same patient, located ictal start to the frontocentral region, and even considered a common pathway for urination and evacuation stimuli (Glass *et al*., 2006; Okumura *et al*., 2007; Rathore *et al*., 2008; Higuchi *et al*., 2011).

In view of the rarity of these clinical features and the lack of topographic clarity, functional neuroimaging data has become more relevant, and moreover, was used for the first time for defecation reflex epilepsy in this report. Concerning micturition-induced seizures, functional studies with PET and SPECT have been previously reported with metabolism and flow imbalance pointing to the anterior cingulate gyrus, as well as the anterolateral right frontal lobe during attacks. (Glass *et al*., 2006). Physiologically, cortical control of micturition derives from the superomedial portion of the frontal lobe and anterior aspect of the cingulate gyrus (Glass *et al*., 2006). A study using PET revealed that the supplementary motor area is one of the regions physiologically activated by contraction of the pelvic floor muscles involved in micturition/defecation and showed that the right-sided micturition centres are more active than those on the left (Blok *et al*., 1997; Higuchi *et al*., 2011). These data may contribute to a better understanding of the pathophysiology of both types of seizure.

Reflex seizures may be difficult to control with AEM (Nagaraja and Chand, 1984; Dreissen and Tijssen, 2012). However, following therapeutic trials, all patients with defecation reflex seizures (including ours) achieved seizure control. Treatment included VPA plus CBZ, clobazam plus phenytoin, or lamotrigine (Harbord and Mitchell, 2002; Higuchi *et al*., 2011).

Epilepsy is known to cause neurological, cognitive, psychological, behavioural and social consequences (Fisher *et al*., 2005). The delay in establishing this unusual diagnosis, with a high frequency of seizures, may have contributed to a negative impact on the epilepsy itself in the reported case. Moreover, the physiological origin of the triggering stimulus, which is hard to deal with in everyday life, may have complicated the situation in our patient, who presented with learning disabilities evolving into intellectual deficit and moderate behavioural problems. These

psychiatric aspects were also observed in the other previously reported patient with the same diagnosis (Harbord and Mitchell, 2002). These findings had negative repercussions regarding QOL indexes and caregiver burden. Reflex seizures, especially those related to physiological stimuli that cannot be controlled by behavioural techniques, may potentially impact QOL, due to the sudden and frequent character of attacks. The literature lacks QOL data specifically for this group of epilepsies.

Finally, although seizure control was achieved, there were no changes in cognitive, behavioural or QOL data. This case emphasises the diagnostic challenge and clinical evolution of defecation reflex epilepsy, and additional cases may further our understanding of the physiopathology in order to optimize effective treatment and avoid cognitive, behavioural and socially deleterious consequences. □

Supplementary data.
Summary didactic slides are available on the www.epilepticdisorders.com website.

Disclosures.
None of the authors have any conflict of interest to declare.

Legend for video sequence

During properly recorded defecation, the child presents with typical focal seizures with impaired awareness, described at home as: behavioural arrest and impaired awareness, right head deviation, left arm automatisms and postictal left hand nose wiping, confusion, and speech arrest.

Key words for video research on www.epilepticdisorders.com

Phenomenology: head deviation, nose wiping
Localisation: temporal lobe (left)
Syndrome: reflex epilepsy (defecation)
Aetiology: not applicable

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TEST YOURSELF

(1) How are reflex seizures defined?

(2) Is defecation a common trigger for reflex seizures?

(3) In a patient with a disperceptive seizure characterized by behavioural arrest and impaired awareness, right head deviation, left arm automatisms, postictal left hand nose wiping, and speech arrest, where is the symptomatogenic zone?

Note: Reading the manuscript provides an answer to all questions. Correct answers may be accessed on the website, www.epilepticdisorders.com, under the section "The EpiCentre".