

# Biofeedback therapy for chronic constipation in a patient with Prader-Willi syndrome

Juan E. Corral<sup>a</sup>, Rahul Kataria<sup>a</sup>, Dawn Vickers<sup>b</sup>, Raghad Koutouby<sup>c</sup>, Baharak Moshiree<sup>b</sup>

University of Miami Miller School of Medicine, Miami, Florida, USA

## Abstract

Constipation is a common feature of Prader-Willi syndrome. Research exploring the prevalence, cause and treatment options for constipation is limited and lacks objective measurements such as anorectal manometry. We report a case of a 16-year-old lady with Prader-Willi syndrome presenting with rectal pain and constipation for 2 years despite multiple medications and weekly enemas. She also noted passive fecal incontinence that required frequent manual disimpactions. Anorectal manometry revealed an abnormal relaxation of the puborectalis and external sphincter muscles on push maneuvers suggesting dyssynergic defecation and rectal hypersensitivity. Contraction and relaxation of her pelvic muscles were recorded with electromyography. Relaxation of the puborectalis muscle improved significantly after three biofeedback sessions. Patient was successfully tapered off laxatives and has been maintained on linaclotide only. Dyssynergic defecation may be a common finding in Prader-Willi syndrome. In selected cases we recommend anorectal manometry to identify neuromuscular dysfunction and subsequent biofeedback therapy depending on the degree of mental retardation to minimize overuse of laxatives.

**Keywords** Prader-Willi syndrome, constipation, pelvic floor disorders

*Ann Gastroenterol 2015; 28 (4): 502-505*

## Introduction

Prader-Willi syndrome (PWS) is a multi-systemic disease resulting from lack of gene expression on the paternally inherited chromosome 15 affecting 1 out of 10,000-30,000 newborns [1]. Gastrointestinal manifestations include overeating, rumination, inability to vomit and chronic constipation [2]. According to the limited evidence available, constipation is significantly more prevalent compared to the general population (40% vs. 11%) [3]. The etiology of constipation in PWS may be multifactorial including a low fiber/high fat diet, rectal evacuation dysfunction due to decreased rectal sensation, reduced muscle tone, dyssynergic

defecation (DD) and slow transit constipation [3]. The only available report however lacks use of objective testing for pelvic floor disorders or neurologic etiology such as anorectal manometry or wireless motility capsule. Furthermore, due to the low prevalence of PWS itself, studies describing treatment options for constipation have been missing.

## Case report

We present a case of a patient with PWS who was referred to our tertiary motility clinic for further evaluation of constipation. PWS was diagnosed in her second month of life after developing four major Holm criteria (neonatal hypotonia, feeding problems, facial features, and intellectual disability) and confirmatory genetic testing in an outside institution [4]. Patient was a 16-year-old female presenting with rectal pain and constipation of 2-year duration. Prior to that, she was having one bowel movement daily. She noted having one soft bowel movement every 5 days with a feeling of incomplete evacuation. Prior medications included polyethylene glycol, lubiprostone, milk of magnesia, and mineral oil leading to mild relief. Additionally, she used bisacodyl, magnesium citrate and enemas at least once weekly in order to have a bowel movement. Patient was not receiving any opioids, thyroid replacement therapy, or antipsychotics. She did not

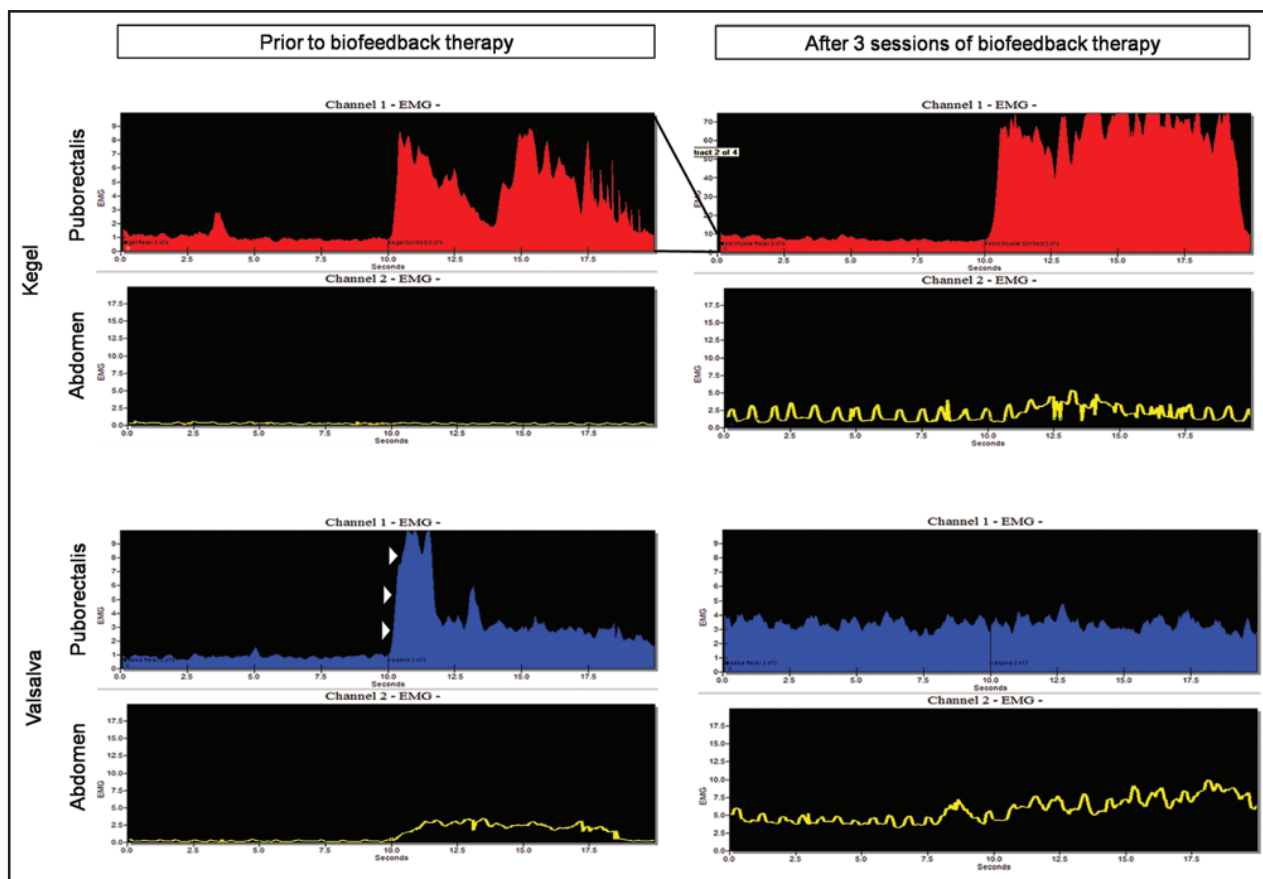
<sup>a</sup>Department of Medicine (Juan E. Corral, Rahul Kataria); <sup>b</sup>Division of Gastroenterology (Dawn Vickers, Baharak Moshiree); <sup>c</sup>Division of Pediatric Gastroenterology (Raghad Koutouby), University of Miami Miller School of Medicine, Miami, Florida, USA

Conflict of Interest: Baharak Moshiree receives financial support from Prometheus Laboratory: grant support and advisory board, and Given Imaging: Speakers bureau and grant support. The other authors declare no conflict of interest

Correspondence to: Juan E. Corral MD, 1611 NW 12th Avenue Central Building, Room 600D (R-60) Miami, FL, 33136 USA, Tel.: +305 243 8644, Fax: +305 243 3762, e-mail: je.corral@med.miami.edu

Received 8 February 2015; accepted 11 July 2015





**Figure 2** Electromyogram readings before and after biofeedback. Dyssynergic defecation seen initially (arrow heads) disappeared with biofeedback. Puborectalis pressure increased during Kegel maneuvers (different scales, 9 mmHg pre and 70 mmHg post biofeedback) and abdominal pressure increased in Valsalva (scale 17.5 mmHg)

### Discussion

The clinical manifestations of PWS are a result of hypothalamic dysfunction, multiple endocrine disorders, intellectual disability and behavioral problems among other things, leading to hyperphagia, obesity and its complications. Patients with this condition also develop gastrointestinal dysmotility including delayed gastric emptying and difficulty vomiting [1,5]. Our patient in particular did not complain of early satiety and had a normal to increased appetite. Severe cases have been reported presenting with acute gastric distention and gastric necrosis with perforation [6,7]. Constipation associated with painful defecation or sensation of anorectal obstruction are likely common in PWS (37% patients reported pain and 42% obstructive symptoms in a case series of 21 patients) [3].

The etiology of constipation in PWS is likely multifactorial, DD was the principal mechanism in our case. Even though our findings cannot be extrapolated to other patients with PWS, DD may be common in patients with PWS who frequently have compulsive behaviors like skin picking and difficulty in changing established routines [1]. This association between rigid-compulsive behavior and constipation has

been previously documented in patients with autism-spectrum disorders [8]. In patients with mild-to-moderate cognitive impairment and other selected patients, we recommend anorectal manometry to identify neuromuscular dysfunction or evaluate for pelvic floor disorders. If DD is found, we advocate for biofeedback therapy to minimize overuse of laxatives. In clinical trials enrolling adults (mean age 46-52, 85% women) that met Rome II criteria for pelvic floor dyssynergia, biofeedback was twice or three times more effective in relieving constipation than pelvic floor exercises, diazepam, or placebo at 3-month follow up [9,10]. Even though there is high-quality evidence showing biofeedback can successfully treat detrusor-sphincter dyssynergia in children, randomized studies addressing constipation have been missing [11,12]. Regardless of age, the ability of biofeedback in normalizing defecation habits will largely depend on the severity of mental retardation and behavioral challenges. Complete motility studies should be considered to rule out other causes of constipation or coexisting upper gastrointestinal motility disorders such as gastroparesis or slow transit constipation. We achieve this by using the wireless motility capsule which helps identify both gastroparesis and slow transit constipation [13].

## References

1. Cassidy SB, Schwartz S, Miller JL, Driscoll DJ. Prader-Willi syndrome. *Genet Med* 2012;**14**:10-26.
2. Alexander RC, Greenswag LR, Nowak AJ. Rumination and vomiting in Prader-Willi syndrome. *Am J Med Genet* 1987;**28**:889-895.
3. Kuhlmann L, Joensson IM, Froekjaer JB, Krogh K, Farholt S. A descriptive study of colorectal function in adults with Prader-Willi Syndrome: high prevalence of constipation. *BMC Gastroenterol* 2014;**14**:63.
4. Holm VA, Cassidy SB, Butler MG, et al. Prader-Willi syndrome: consensus diagnostic criteria. *Pediatrics* 1993;**91**:398.
5. Arenz T, Schwarzer A, Pfluger T, Koletzko S, Schmidt H. Delayed gastric emptying in patients with Prader Willi Syndrome. *J Pediatr Endocrinol Metab* 2010;**23**:867-871.
6. Wharton RH, Wang T, Graeme-Cook F, Briggs S, Cole RE. Acute idiopathic gastric dilation with gastric necrosis in individuals with Prader-Willi syndrome. *Am J Med Genet* 1997;**73**:437-441.
7. Stevenson DA, Heinemann J, Angulo M, et al. Gastric rupture and necrosis in Prader-Willi syndrome. *J Pediatr Gastroenterol Nutr* 2007;**45**:272-274.
8. Peters B, Williams KC, Gorrindo P, et al. Rigid-compulsive behaviors are associated with mixed bowel symptoms in autism spectrum disorder. *J Autism Dev Disord* 2014;**44**:1425-1432.
9. Heymen S, Scarlett Y, Jones K, Ringel Y, Drossman D, Whitehead WE. Randomized, controlled trial shows biofeedback to be superior to alternative treatments for patients with pelvic floor dyssynergia-type constipation. *Dis Colon Rectum* 2007;**50**:428-441.
10. Heymen S, Scarlett Y, Jones K, Ringel Y, Drossman D, Whitehead WE. Randomized controlled trial shows biofeedback to be superior to pelvic floor exercises for fecal incontinence. *Dis Colon Rectum* 2009;**52**:1730-1737.
11. Porena M, Costantini E, Rociola W, Mearini E. Biofeedback successfully cures detrusor-sphincter dyssynergia in pediatric patients. *J Urol* 2000;**163**:1927-1931.
12. Klijn AJ, Uiterwaal CS, Vijverberg MA, Winkler PL, Dik P, de Jong TP. Home uroflowmetry biofeedback in behavioral training for dysfunctional voiding in school-age children: a randomized controlled study. *J Urol* 2006;**175**:2263-2268; discussion 2268.
13. Kuo B, McCallum RW, Koch KL, et al. Comparison of gastric emptying of a nondigestible capsule to a radio-labelled meal in healthy and gastroparetic subjects. *Aliment Pharmacol Ther* 2008;**27**:186-196.