

A descriptive study of colorectal function in adults with Prader-Willi Syndrome: high prevalence of constipation.

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Abstract

BACKGROUND:

Some patients with Prader-Willi Syndrome (PWS) have symptoms of constipation, but bowel function in PWS has never been systematically evaluated. The aim of the present study was to describe colorectal function in PWS by means of validated techniques.

METHODS:

Twenty-one patients with PWS (14 women, age 17-47 (median = 32)) were evaluated with the Rome III constipation criteria, stool diary, digital rectal examination, rectal diameter assessed from transabdominal ultrasound, and total gastrointestinal transit time (GITT) determined with radio-opaque markers. Results were compared with those of healthy controls.

RESULTS:

Among PWS patients able to provide information for Rome III criteria, 8/20 (40%) fulfilled the criteria for constipation. Most commonly reported symptoms were a feeling of obstructed defecation (8/19, 42%), <3 defecations per week (8/17, 47%), straining during defecation (7/19, 37%) and lumpy or hard stools (6/19, 32%). Rectal diameter did not differ between PWS (median 3.56 centimeters, range 2.24-5.36) and healthy controls (median 3.42 centimeters, range 2.67-4.72) ($p = 0.96$), but more PWS patients (13/20; 65%) than healthy controls (3/25; 12%) ($p < 0.001$) had fecal mass in the rectum. Median GITT was 2.0 days (range 0.5-4.4) in PWS versus 1.6 (range 0.7-2.5) in the control group ($p = 0.26$). However, GITT was >3 days in 5/21 (24%) of PWS and none of the controls ($p = 0.047$).

CONCLUSION:

Constipation is very common in PWS. Patients with PWS have an increased prevalence of prolonged GITT and palpable stools in the rectum at digital rectal examination.