Rectal Digging Associated with Prader-Willi and Solitary Rectal Ulcer Syndrome: A Case of the Chicken or the Egg

Kelsey Soppet DO, MA and Pankaj Vohra MBBS, MD

Kelsey Soppet- ksoppet@salud.unm.edu

Introduction

Prader-Willi syndrome (PWS) is a multisystem disorder which occurs due to the absence of paternally expressed imprinted gene at 15q11.2-q13. Individuals with PWS typically display hypotonia at birth, developmental delays, a distinct behavioral phenotype, and are prone to skin picking behaviors such as rectal digging in severe instances [1]. The picking behaviors have been studied and hypothesized to be due to a behavioral phenotype and associated with OCD tendencies. Skin picking to the point of self-injury is common in individuals with PWS as they have been observed to have a higher pain threshold probable of central origin and positive autonomic nervous system phenotype. [2]

Rectal digging may also be a resultant behavior of solitary rectal ulcer syndrome (SRUS), a rare condition especially in children. SRUS commonly presents as rectal bleeding, passage of mucus, excessive straining during bowel movements, and a sense of incomplete defecation. Diagnosis is made by histopathological changes of fibromuscular obliteration of lamina propria. [3] When occurring together, in the case of this patient, it is challenging to discern which condition propelled the other.

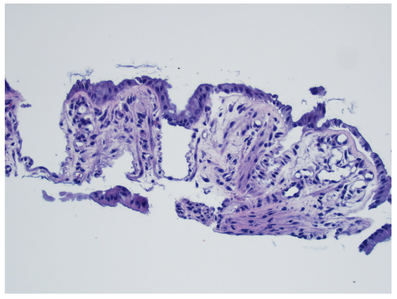


Figure 1:

Biopsy sample from Sigmoidoscopy of Rectosigmoid shows fragments of colonic mucosa with focal reactive changes and features of rectal prolapse.

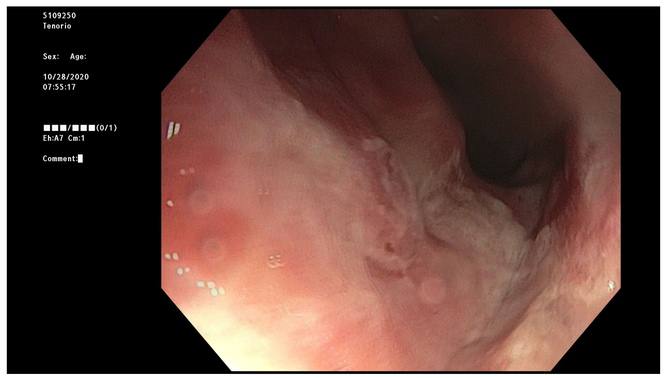
Case

A 12-year-old male with Prader-Willi Syndrome and developmental delay was admitted to the general pediatric floor for aggressive and self-injurious behaviors including rectal digging after bowel movements. His picking behaviors were defined as severe and frequently resulted in profuse bleeding. He had also displayed signs of hyperphagia and pica ingesting the blood and stool product. The initial complete blood count revealed a normocytic anemia. The source was felt to be likely behavioral and psychiatric treatment was targeted.

Upon visualization of rectal prolapse during his stay Polyethylene glycol was initiated and stool studies completed. Stool ova and parasite, Stool Giardia, and Celiac Panel were within normal limits. Liver Function Test, ESR, CRP, and lead level were also within normal limits. A complete blood count with differential was repeated with iron studies which revealed an iron deficiency anemia, and a fecal calprotectin was elevated to 2350. The patient was initiated on mineral oil daily to aid in stool passage. Sigmoidoscopy revealed a self reducing prolapse, a sample of which is shown in Figure 1, and a solitary rectal ulcer as shown in Figure 2. Biopsy of the ulcerated lesion revealed predominantly fibrinopurulent exudate and scant granulation tissue thereby confirming diagnosis of SRUS.

Medical management for microcytic anemia with iron and vitamin C supplementation begun. Medical management for the SRUS was initiated with hydrocortisone enema 100 mg administered daily . Daily administration preferred over twice daily secondary to patient compliance. Recommended course was 2 weeks however only one week was completed at our facility secondary to patient transfer to long-term facility. Subjective improvement was noted in rectal digging behavior after initiation of hydrocortisone enemas as determined by decreased frequency of behavior.

Records of objective improvement are attempting to be obtained from outlying facility.



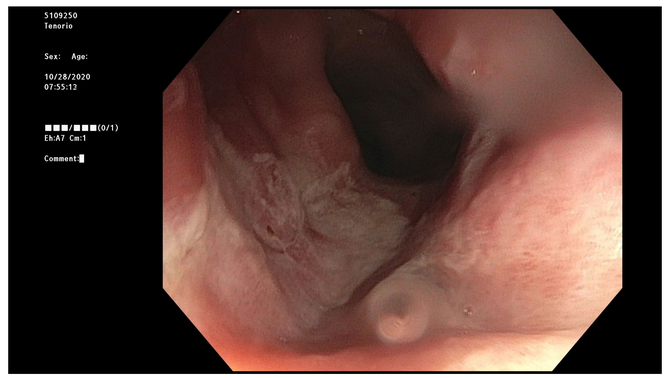


Figure 2a: Imaging from sigmoidoscopy highlighting solitary rectal ulcer best visualized at the 4 o' clock position in the rectal vault. **2b**: An alternative view of the ulcer best visualized in the 8 o' clock position

Discussion

Rectal digging with concomitant Prader-Willi behaviors and SRUS led us to ask the clinical question whether SRUS caused feelings of incomplete defecation and subsequent picking or did acute trauma from a self-injurious behavior cause the formation of a solitary rectal ulcer. In PWS the picking behaviors have been found to usually be directed at skin impurities. However, the exception to this rule is in regard to rectal picking, although it is still presumed to begin secondary to some irritation. [2] In some cases, the digging may become so severe as to lead to rectal ulceration. In discussion with family, this behavior had been ongoing for five months. Digitation and chronic trauma secondary to behavioral and autonomic response is a possibility in this patient.

Alternatively, what seems equally likely with histopathological confirmation of SRUS is that the ulceration caused irritation and feelings of incomplete defecation. This syndrome is known to occur in children with dyssynergic defecation, a likely outcome of individuals with autonomic instability such as seen in PWS. These feelings often lead to rectal digitation. Likely etiology of SRUS is felt to be related to ischemic injury due to prolapse of anterior rectal mucosa against contracted puborectalis muscle [3]. In our case, the individual was noted to have a self reducing prolapse thereby heightening suspicion for SRUS as the causative mechanism of rectal digging.

Conclusion

In the management of individuals with Prader-Willi, developmental delay, and skin picking behaviors it is important for providers moving forward to recognize SRUS as a possible causative diagnosis for rectal digging. Expanding our differential to include this diagnosis could effectively treat the source and help minimize or eliminate this behavior thereby improving morbidity and psychological stress.

References

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•**Disclosure:** Authors of this presentation have the following to disclose concerning possible financial or personal relationships with commercial entities that may have a direct or indirect interest in the subject matter of this presentation:

•Kelsey Soppet: Nothing to disclose

•Pankaj Vohra: Nothing to disclose

•Source of Funding: None