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journal homepage: www.casereports.com**Case report of extreme gastric distention and perforation with pathologic *Sarcina ventriculi* colonization and Rett syndrome**

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*Sarcina ventriculi***ABSTRACT**

Here we describe a case of a 15-year-old child with Rett syndrome who presented with extreme gastric distension and fatal perforation in the setting of long-standing aerophagia and pathologic colonization with *Sarcina ventriculi*, a rare bacteria implicated in gastric perforation. This is the first report of gastric perforation associated with colonization by *Sarcina* in a patient with pathologic aerophagia. Gastric colonization with *Sarcina* should be considered in intellectually disabled children with pathologic air swallowing who present with severe gastric dilation and/or perforation.

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1. Introduction

Rett syndrome is a profound neurodevelopmental disorder associated with a number of stereotyped behaviors including excessive air swallowing, or aerophagia [1,3]. While it is not uncommon to see abdominal distension in children with aerophagia, progression to perforation of the gastrointestinal tract is rare, with fewer than a dozen cases reported in the literature [5,8–10]. Recent evidence suggests that gastric colonization with the pathogenic bacteria *Sarcina ventriculi* can be associated with a number gastrointestinal abnormalities including delayed gastric emptying, gastric outlet obstruction, acute gastric dilation, and gastric perforation [7]. Whereas pathologic aerophagia and colonization with *Sarcina* have independently been recognized as risk factors for gastric perforation, the interplay between these entities is unknown [7]. Understanding this relationship could potentially help identify at risk individuals for closer monitoring and possible elective decompressive gastrostomy placement to prevent excessive gastric dilation. The work herein has been reported in line with the SCARE criteria [11].

2. Presentation of case

A 15-year-old female with a known diagnosis of Rett syndrome presented to another hospital with several hours of progressive abdominal distension. She had been in her usual state of health until her most recent meal a few hours prior. Her parents reported that shortly after finishing dinner, the child became “fussy” and

developed a markedly protuberant abdomen. Past medical history was notable for malnutrition, esophageal dysmotility, seizures, and pathologic aerophagia—known sequela of Rett syndrome, a disabling neurological disorder characterized by intellectual disability, autistic-like behaviors, and poor growth [1]. On initial exam she was found to have a distended and rigid abdomen, along with violaceous lower extremities concerning for abdominal compartment syndrome. Urgent abdominal CT scan demonstrated extreme gastric dilation with extensive portal and splenic venous gas. She was immediately referred to a pediatric specialty hospital, however, during transportation, experienced cardiac arrest with recovery following several minutes of cardiopulmonary resuscitation. The patient remained hemodynamically unstable thereafter and labs were notable for profound acidosis (pH 6.81), lactate of 13.1 mmol/L, white blood cell count of 14×10^9 /L, creatinine of 0.3 mg/dL, abnormal liver profile (ALT 1406, AST 1272, Alkaline Phosphatase 53), and coagulopathy (INR: 1.6, PTT: 151 s). The decision was made to take the patient to the operating room for exploration of what appeared to be an abdominal catastrophe. Emergent exploratory laparotomy was performed and revealed a massively dilated stomach with a 2 cm linear, burst-like perforation along the lesser curvature (Fig. 1); significant amounts of free intraperitoneal blood and gastric contents were encountered. The stomach was not volvulated and the ligamentous attachments were all intact with no twisting. The small bowel was also dilated with evidence of hypoperfusion but no definite ischemia. A wedge resection of the perforation was performed and a temporary abdominal closure was achieved using an Open Abdomen Negative Pressure Therapy Vacuum in order to facilitate more rapid transfer to the ICU for further resuscitation and hemodynamic stabilization. The patient's condition rapidly deteriorated post-operatively and she died hours later. Histopathologic examination of the gastric

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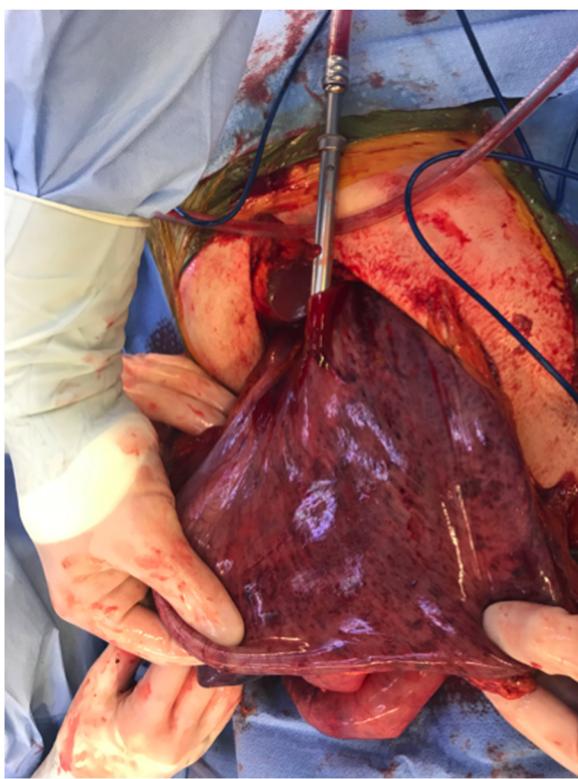


Fig. 1. Intraoperative findings: gastric necrosis with a 2 cm perforation along the lesser curvature.

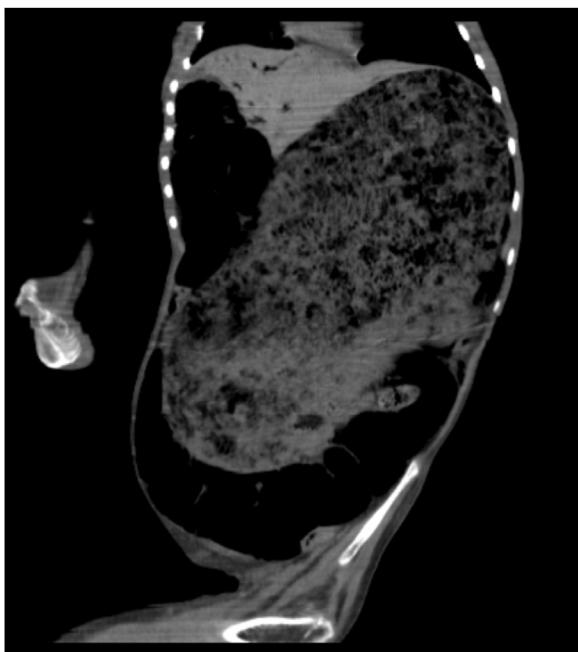


Fig. 2. Abdominal CT scan demonstrating a markedly dilated stomach with intramural air and extensive portal venous gas.

specimen revealed pathologic colonization with *Sarcina ventriculi* (Fig. 2).

3. Discussion

Rett syndrome is a rare genetic neurodevelopmental disorder affecting 1 in 9000 girls under the age of 12 [1]. Most cases are due to

spontaneous mutations in the MECP2 gene on the X-chromosome. Consequent decreases in MECP2 protein are thought to alter patterns of DNA methylation leading to impaired maturation of the central nervous system and inadequate formation of synapses [2]. The syndrome is associated with a number of pathologic behaviors including excessive air swallowing and breath holding [3]. Spontaneous gastric perforation in children with Rett syndrome, like that seen in our patient, is an uncommon event rarely reported in the literature. One proposed mechanism for the association between gastrointestinal perforation and Rett syndrome is pathologic aerophagia—the excessive and inappropriate swallowing of air often seen in intellectually disabled children [4,5]. Aerophagia in children, which has an estimated prevalence of 7.5% (range 0.1–8.8%), can be mild and transient, or can become chronic and severe enough to lead to gastric necrosis or perforation; the exact prevalence of this specific complication in the pediatric aerophagia population is limited by the small number of case reports available in the literature [5].

Additionally, *Sarcina ventriculi* is an increasingly common gram-positive, anaerobic coccus being found in the gastric biopsies of adult patients with a wide-range of gastrointestinal pathologies including delayed gastric emptying, emphysematous gastritis, and gastric perforation; its role in children remains to be elucidated [7]. Colonization can present clinically as chronic nausea, frothy spit-up, and abdominal pain, a condition known as *sarcinous vomit* [7]. Conversely, patients may be asymptomatic, and the bacteria may be an incidental finding on biopsy. The pathogenesis of the organism is debated, and it is unclear what drives colonized individuals to develop mild disease versus life-threatening gastric perforation. Similarly, it is unknown how colonization with *Sarcina* increases the risk of gastric perforation in children with other pre-existing risk factors such as pathologic aerophagia. It may be an additive risk factor or synergistic.

The diagnosis and management of gastric perforation in children with Rett syndrome is particularly challenging given the limited number of cases described in the literature; surgical repair remains the only definitive therapy. Gastric perforation is thought to occur more frequently on the lesser curvature, as was seen in this case, due to its reduced elasticity compared with other parts of the stomach [6]. In children, these gastric perforations are usually repaired primarily in order to avoid the potential feeding difficulties associated with a more extensive gastrectomy.

4. Conclusion

This is one of only a few reported cases of gastric perforation in a patient with Rett syndrome thought to be secondary to aerophagia. Moreover, this case highlights the need for additional research looking at the relationship between the *Sarcina* bacterium and the increased risk of gastric perforation in children with pre-existing conditions. By reporting this case we hope to broaden awareness of this unique surgical complication seen in Rett syndrome and children with pathologic aerophagia; increased knowledge of this potentially fatal complication might expedite diagnosis of this very rare, fatal event and avoid the unfortunate outcome that occurred in this case. For children with significant aerophagia, we encourage consideration of a venting gastrostomy tube, as has been previously reported in the literature [9,10].

Declaration of Competing Interest

None. None of the authors have any conflict of interest.

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Ethical approval

We have reached out to our IRB for expedited review of our request to be exempt from ethical approval given that this is a retrospective case study that does not contain identifying information. **We have attached a letter from our Institutional Review Board (IRB) stating that IRB approval is not required for this manuscript.**

Consent

Written informed consent was not obtained from the patient. The head of our medical team has taken responsibility that exhaustive attempts have been made to contact the family and that the paper has been sufficiently anonymised not to cause harm to the patient or their family. A copy of a signed document stating this is available for review by the Editor-in-Chief of this journal on request. **Moreover, we have gone through our Institution's IRB and obtained a letter stating that IRB approval is not required to proceed with publication of this manuscript.**

Author contribution

Carla Lopez – Writing of the manuscript; analysed the data.
Mark Kovler – Conceived and presented the idea; manuscript editing; analysed the date.

Eric Jelin – Supervised the project; analysed the data.

Registration of research studies

1. Name of the registry: Institutional Review Board (IRB). **A letter from our Institution's IRB is attached.** This is a retrospective case study.
2. Unique identifying number or registration ID: N/a. **A letter from our Institution's IRB is attached.** This is a retrospective case study.
3. Hyperlink to your specific registration: N/a. **A letter from our Institution's IRB is attached.** This is a retrospective case study.

Guarantor

Dr. Eric Jelin.

Additional information

The authors whose names are listed immediately below certify that they have NO affiliations with or involvement in any organization or entity with any financial interest (such as honoraria; educational grants; participation in speakers' bureaus; membership, employment, consultancies, stock ownership, or other equity interest; and expert testimony or patent-licensing arrangements), or non-financial interest (such as personal or professional relationships, affiliations, knowledge or beliefs) in the subject matter or materials discussed in this manuscript.

Provenance and peer review

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References

- [1] A. Anderson, K. Wong, P. Jacoby, J. Downs, H. Leonard, Twenty years of surveillance in Rett syndrome: what does this tell us? *Orphanet J. Rare Dis.* 8 (9) (2014).
- [2] S. Luijkenhuis, E. Giacometti, C. Beard, R. Jaenisch, Expression of MeCP2 in postmitotic neurons rescues Rett syndrome in mice, *PNAS* 101 (16) (2004) 6033–6038.
- [3] R. Morton, L. Pinnington, R. Ellis, Air swallowing in Rett syndrome, *Dev. Med. Child Neurol.* 42 (2000) 271–275.
- [4] C. Aldape, A. Glaser, Pathologic aerophagia in Rett syndrome, *J. Child. Dev. Disord.* (2017), <http://dx.doi.org/10.3389/fsurg.2019.00003>.
- [5] Z. Zvizdic, A. Jonuzi, A. Djuran, S. Vranic, Gastric necrosis and perforation following massive gastric dilation in an adolescent girl: a rare cause of acute abdomen, *Front. Surg.* 6 (3) (2019).
- [6] M. Alvin, N. Al Jalbout, *BMJ Case Rep.* (2018), <http://dx.doi.org/10.1136/bcr-2018-224233>.
- [7] M. Rasheed, C. Senseng, *Arch. Pathol. Lab. Med.* 140 (12) (2016) 1441–1445.
- [8] A. Akalonu, M. Yasrebi, Z. Molle Rios, Spontaneous gastric perforation in two adolescents, *Am. J. Case Rep.* 17 (2016) 694–698.
- [9] E. Baldassarre, G. Capuano, G. Valenti, P. Maggi, A. Conforti, I. Porta, A case of gastric necrosis in a young girl with Rett syndrome, *Brain Dev.* (2006), <http://dx.doi.org/10.1016/j.braindev.2005.03.010>.
- [10] A. Sezer, M. Yagci, A. Hatipoglu, The management of gastric perforation in a girl with Rett syndrome, *Brain Dev.* 33 (2011) 83–85.
- [11] R.A. Agha, M.R. Borrelli, R. Farwana, K. Koshy, A. Fowler, D.P. Orgill, For the SCARE Group, The SCARE 2018 statement: updating consensus Surgical CAsE REport (SCARE) guidelines, *Int. J. Surg.* 60 (2018) 132–136.

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