

Development of Ileus in Edwards Syndrome with Exceptional Longevity

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ABSTRACT

Background: Edwards syndrome, or trisomy 18, is a rare condition often incompatible with life. Gastrointestinal complications of this syndrome are documented in the infant period, however, motility disorders in adult survivors have not been previously reported. We present a case of intestinal ileus in a 29 year old male with Edwards syndrome.

Case: A 29-year-old male with Edwards syndrome presented to the emergency department with gastrointestinal complications at his group home. His medical history includes hypothyroidism, severe intellectual disability, and contracted body habitus. The patient was admitted to the hospital with intestinal ileus secondary to his antipsychotic medications.

Conclusions: Documented cases of Edwards syndrome in adulthood are limited. Our case presents an unexplored manifestation of gastrointestinal complications secondary to medication. Clinicians should be aware of the side effects of antipsychotic medications when caring for patients with the condition.

Introduction

Edwards syndrome, also known as trisomy 18, is the second most common autosomal trisomy, occurring in approximately 1/6,000-1/8,000 live births^{1,2}. Existing guidelines for care are specific and limited to pediatric populations². Survival into adulthood is exceptionally rare; to date, the longest documented survivor is a 26-year-old female with non-mosaic trisomy 18³. Additionally, this condition is more prevalent in females, with a male-to-female ratio of 0.69 at birth⁴.

Gastrointestinal complications in Edwards syndrome, while less frequent (5-25%), are well-documented in the neonatal period, including omphalocele, esophageal atresia, and imperforate anus^{1,2}. However, gastrointestinal motility disorders and acute abdominal complications specifically in adult survivors have not been previously reported. We present the first documented case of drug-induced ileus in an adult with Edwards syndrome, highlighting the unique challenges of managing acute gastrointestinal complications in this rare population.

Case Presentation

A 29-year-old male with non-mosaic Edwards syndrome presented to the emergency department with abdominal distension and diarrhea for one day. The non-mosaic trisomy 18 was confirmed after birth via chromosome analysis and fluorescent in situ hybridization. The patient resided in a group home and had limited verbal communication, with baseline mental status described as alert

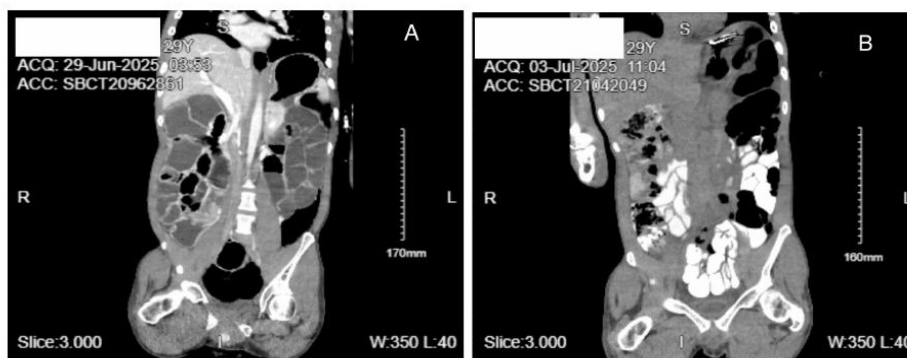


Figure 1. (A) CT A/P on admission. (B) Repeat CT A/P

and responsive but non-communicative. He was restricted to his bed due to contracted anatomical limitations.

The patient had been constipated at his group home and was given prune juice, after which he experienced at least five episodes of non-bloody diarrhea, but no reported episodes of emesis. He has a history of similar episodes, but no history of bowel obstruction. At his group home, the patient was maintained on a pureed diet with thick liquids.

Upon admission, the patient's documented weight and height were 38.8kg and 152cm (BMI 16.79). The patient had developmental and growth restriction. His medical history included microcephaly with ex vacuo dilation of the third and lateral ventricles, and hypothyroidism managed with levothyroxine 12.5 mcg daily (TSH 2.060, free T4 0.80 on admission). The patient had no documented cardiac defects, and ECG showed only sinus tachycardia. His psychiatric medications included quetiapine 50 mg twice daily and benzotropine 1 mg twice daily, both of which had been held on admission due to suspected intestinal obstruction. The patient had no prior surgical history and no known allergies.

Physical examination upon initial presentation revealed significant abdominal distension and guarding. A nasogastric tube was placed in the emergency department for decompression, in which green-colored gastric fluid was collected. Initial computed tomography of the abdomen and pelvis with oral contrast demonstrated a significantly air- and fluid-distended stomach and loops of small bowel compatible with obstruction, with swirling of the mesentery and twisting of the bowel in the mid-abdomen representing a possible transition point. The large bowel was also moderately distended, indicating potential ileus as well [Figure 1A]. Admission electrolytes were within normal limits and the patient did not have leukocytosis.

A repeat CT abdomen and pelvis with contrast on hospital day 3 ruled out intraluminal obstruction and showed decreasing distension and generalized ileus bowel gas pattern [Figure 1B]. Surgical evaluation ruled-out malrotation, however correlated the findings with ileus and dysfunctional gastrointestinal system secondary to the



Figure 2. Abdominal X-ray on Day 5

patient's underlying medical condition, and Gastroenterology attributed the diffuse bowel distension to possible drug-induced etiology, specifically the anticholinergic effects of quetiapine and benzotropine^{5,6}. No surgical intervention was deemed necessary as repeat imaging showed improved distension and resolution of a potential transition point. No bowel obstruction was found and per Gastroenterology's assessment, the patient was experiencing pseudo-obstruction secondary to anticholinergics.

The patient was managed conservatively with nasogastric decompression, bowel rest, and intravenous fluid resuscitation. Partial parenteral nutrition was initiated to address moderate malnutrition while the patient remained NPO.

The nasogastric tube was then removed and the patient was advanced to a clear liquid diet with continued electrolyte monitoring. Speech-language pathology evaluated the patient and recommended pureed solids and thin liquids. The patient required total assistance for feeding. Risk assessments indicated low risk for both aspiration and malnutrition with appropriate dietary modifications.

Abdominal x-ray on day 5 showed fecal accumulations in the entire colon with subtle improvement from prior imaging. Ileus bowel gas pattern also improved from prior imaging [Figure 2].

The patient tolerated oral intake and demonstrated return of normal bowel movements. No pressure ulcers developed during hospitalization. Electrolytes and leukocytes remained within normal limits during his hospital course. He was discharged after a 7-day hospital stay with instructions for psychiatric follow-up in one week to consider restarting quetiapine and benztropine under close monitoring for gastrointestinal side effects.

Discussion

This case represents the first documented presentation of drug-induced ileus in an adult with Edwards syndrome. The patient's survival to age 29 is exceptional, as only 12.3% of individuals with trisomy 18 survive beyond 5 years, and survival into the third decade is extraordinarily rare, especially considering the discrepancy in newborn survival based on sex^{2,3,7}.

Differential diagnoses for this patient included medication-induced ileus versus mid-abdomen malrotation versus small bowel obstruction. The patient's lack of abdominal surgical history in addition to his medication use pointed to a more functional cause of ileus rather than a mechanical one. It is possible, however, that this patient had a mechanical obstruction superimposed on a functional obstruction: malrotation of the midabdomen from drug-induced ileus, which resolved with nasogastric tube placement. There was no evidence of colonic malrotation, and so nasogastric decompression without additional rectal tube placement may have been sufficient.

The ileus was attributed to the combined anticholinergic effects of quetiapine and benztropine. Quetiapine and benztropine both exhibit anticholinergic properties, contributing to side effects such as constipation, bowel obstruction, and paralytic ileus^{5,6}. The patient's underlying Edwards syndrome likely contributed additional risk factors for gastrointestinal dysmotility. Patients with trisomy 18 commonly experience feeding problems and gastrointestinal complications, though these are typically described in infancy^{1,2}. The patient's bed-restricted status due to his anatomy further reduced mobility, a known risk factor for constipation and ileus^{4,7}. Conservative management with nasogastric decompression, bowel rest, fluid resuscitation, and discontinuation of anticholinergic medications proved successful, consistent with evidence-based approaches to drug-induced ileus^{5,7}. The decision to avoid surgical intervention was appropriate given the functional rather than mechanical nature of the obstruction and the patient's complex medical status^{2,7}. The cognitive disabilities in patients with Edwards syndrome may require pharmacologic management, exposing surviving patients with the condition to anticholinergic medications. This may further potentiate existing gastrointestinal complications associated with Edwards syndrome.

While an incredibly rare occurrence, it is important to recognize cases of Edwards syndrome and monitor patients' diets and medications in order to prevent gastrointestinal complications. An appropriate preventative measure would be to reduce the anticholinergic burden of psychiatric medications; our patient was taking both quetiapine and benztropine, both of which have anticholinergic and constipating effects. The use of medications with fewer anticholinergic effects, such as aripiprazole, has a reduced risk of ileus and constipation. As such, clinicians may consider using alternative medications for psychiatric manifestations of Edwards syndrome⁵.

In contrast to Edwards syndrome, bowel dysfunction in other neurologic conditions has been well-documented. Patients with Parkinson disease experience damage to the enteric nervous system and dorsal vagal nerve output, resulting in delayed gastric motility. Additionally, patients with Parkinson disease are often taking anticholinergic medications, which contribute to constipation. Those with neurologic conditions such as Parkinson disease, multiple sclerosis, and stroke also have reduced mobility, further contributing to constipation⁸. Immobility or reduced mobility is a complication of Edwards syndrome as well, as patients can have significantly contracted body habitus, as seen with our patient. It is suggested to incorporate digital rectal exam to assess sphincter tone and sensation into the management of neurologic bowel dysfunction, as reduced anal sensation and impaired anal sphincter tone potentiate complications of constipation⁸. Long-term management of ileus, whether secondary to Edwards syndrome or other neurologic conditions, should include both conservative and pharmacologic measures. Conservative management includes adequate fluid and fiber intake, careful monitoring of medication side effects, and abdominal massage. Pharmacologic management includes the use of stool softeners and laxative medications⁸. Ileus in these conditions as well as Edwards syndrome is multifactorial. Our patient improved with decompression and resuming a pureed diet; management upon his return to the group home should include high fiber pureed meals, sufficient fluids, and stool softeners as needed. Patients with Edwards syndrome require even more careful consideration as they may be unable to communicate with care providers.

This case highlights several important clinical considerations. Adult survivors of Edwards syndrome require ongoing multidisciplinary care with attention to medication side effects. Second, the combination of anticholinergic psychiatric medications in patients with baseline reduced gastrointestinal motility and limited mobility creates substantial risk for serious gastrointestinal complications. Third, conservative management can be successful even in medically complex patients when the underlying etiology is appropriately identified and

addressed. The decision regarding reinitiation of psychiatric medications requires careful risk-benefit analysis.

Conclusion

We report the first case of ileus in an adult with Edwards syndrome, successfully managed with conservative treatment. This case expands the limited literature on adult survivors of trisomy 18 and emphasizes the importance of recognizing medication-related gastrointestinal complications in this population. Clinicians caring for adults with Edwards syndrome should be aware of several key considerations, including overlapping medication side effects, especially anticholinergic effects, and the gastrointestinal manifestations of this syndrome, which require close monitoring. Establishing preventive strategies to minimize the risk of complications such as ileus or pseudo-obstruction is essential to avoid unnecessary surgical interventions and ensuring that conservative medical management remains the priority.

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