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Case Report

Sigmoid volvulus in Bardet-Biedl syndrome: serendipity or a new association?

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ABSTRACT

Bardet-Biedl syndrome is a rare ciliopathy with a wide array of clinical features, including congenital blindness, obesity, neuroendocrine disturbance, hypogonadism and renal dysfunction. The association of Hirschsprung's disease with Bardet-Biedl syndrome has been published earlier, however, there is no literature regarding patients with Bardet-Biedl syndrome presenting later in life due to complications associated with Hirschsprung's disease. Bardet-Biedl Syndrome is exceedingly rare, with only around 15 case reports published from India. Here we present the case of a 55-year-old male patient with Bardet-Biedl syndrome who presented with intestinal obstruction, raising the question as to whether his emergency presentation was a complication of his flawed genotype or serendipity.

Keywords: Bardet-Biedl syndrome, Hirschprung's disease, Sigmoid volvulus

INTRODUCTION

Bardet-Biedl syndrome is a rare autosomal-recessive ciliopathy, affecting multiple organ systems, with heterogeneous features including central obesity, retinal dystrophy, mental retardation, polydactyly, hypogonadism and renal dysfunction.[1] Only around 15 cases of this rare disorder have been reported from India.[2]

Hirschsprung's disease has been documented to occur occasionally in patients with Bardet-Biedl syndrome.[1] So far, as per literature review, there has been no previous report of patient with Bardet-Biedl syndrome presenting due to complications of Hirschsprung's disease. This report is of a patient with Bardet-Biedl syndrome who presented with sigmoid volvulus, thus begging the question, if the occurrence was by serendipity, or as a complication of Hirschsprung's disease.

CASE REPORT

A 55 year old male patient was brought to the emergency department with complaints of abdominal pain and distention, associated with vomiting and constipation for the last 4 days. The symptoms were insidious in onset and gradually progressive, with rapid increase in pain over the last 12 hours. He had multiple episodes of vomiting, including while being assessed in the casualty - mildly projectile, bilious, non-feculent, non-blood-stained. He had not been given any oral feeds for over a day and was visibly dehydrated and distressed. He had not been able to pass flatus for the last 2-3 days.

He was a known hypertensive and diabetic on medication. As per his attenders history, he suffered from congenital blindness and had been progressively becoming hard of hearing. He was mentally challenged and had no formal education or occupation, and spent most of his time doing menial chores. He was unmarried and without issue.

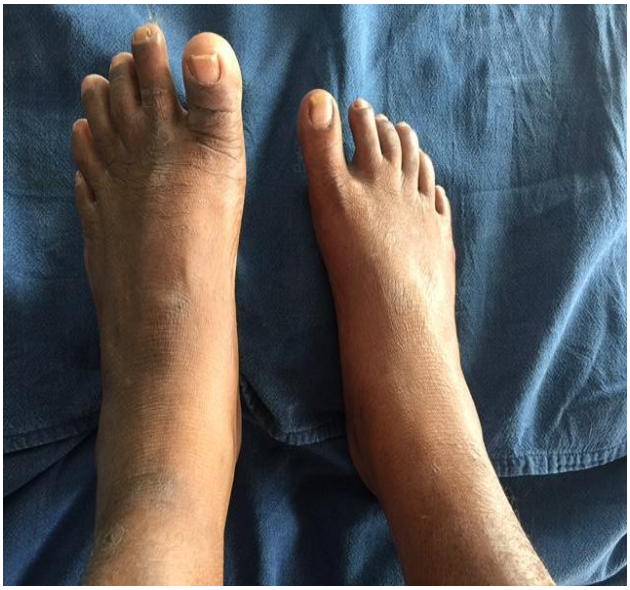

Figure 1: Polydactyly of his lower limbs.

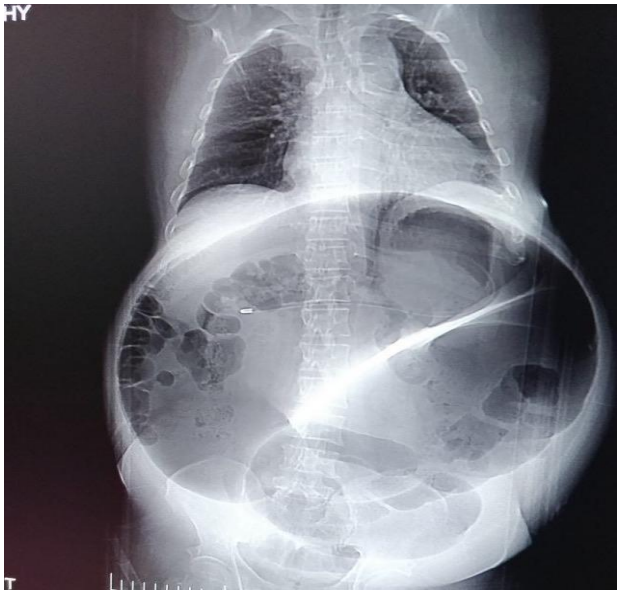

Figure 2: Coffee-bean appearance CT.

On examination, he was obese, with a predominantly truncal pattern. Both corneas appeared cloudy and there was no apparent visual acuity. He had low comprehension ability but was able to communicate. Examination also revealed postaxial polydactyly on both hands and feet (Figure 1).

Routine investigations revealed elevated total counts (16,340/cmm) with neutrophilia, mildly elevated renal parameters (creatinine 1.4 mg/dl), and hyponatremia (sodium - 128 mEq/L).

Abdominal radiograph followed by CT imaging was done which showed sigmoid volvulus, having the classical "coffee-bean" appearance (Figure 2).

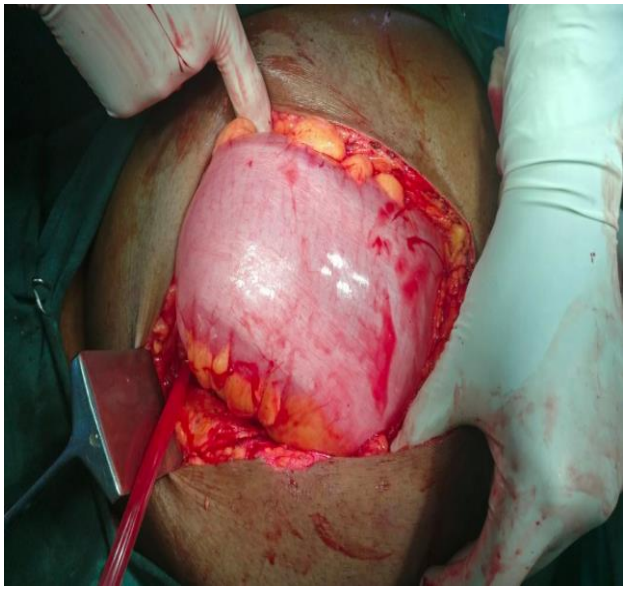

Figure 3: Bowel protruding through incision.

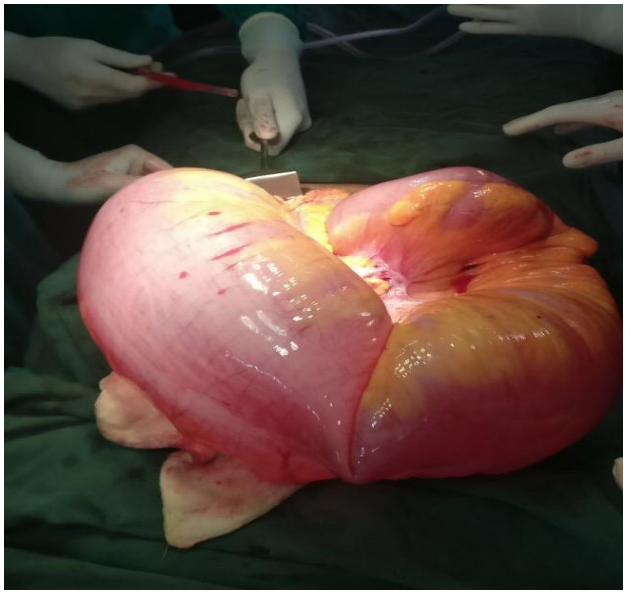

Figure 4: Grossly dilated bowel.

In view of intestinal obstruction and sepsis, the patient was taken up for emergency surgery after fluid resuscitation. Exploratory laparotomy was performed with midline incision, revealing a grossly dilated and thinned-out large bowel that "popped" out through the incision due to change in the pressure gradient (Figure 3 and 4).

The bowel appeared thinned-out to its tether at several areas, predictive of imminent perforation or rupture. Deflation of the megacolon was performed which revealed a large volume of redundant bowel (Figure 5), in view of which resection was done followed by colostomy.

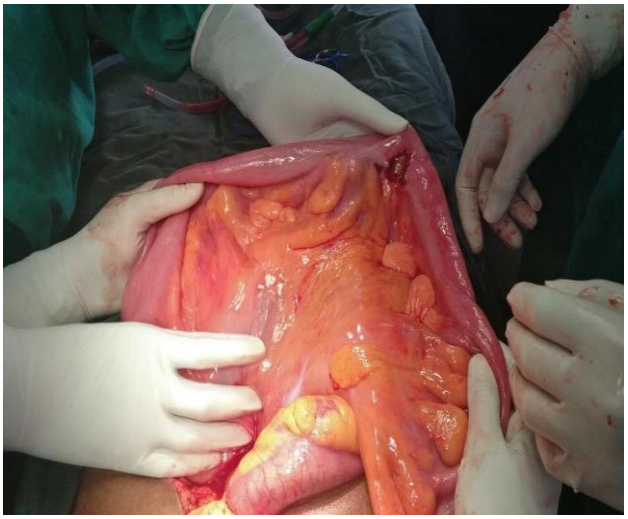

Figure 5: Redundant bowel.

The postoperative period was uneventful, and the patient was discharged with planned colostomy closure at a later date.

DISCUSSION

Bardet-Biedl Syndrome is a rare pleiotropic recessive inherited ciliopathy, with an estimated frequency of 1:160,000, with less than 15 cases being reported from India.[2,3]

The condition is characterised by a heterogenous manifestation of features, involving multiple organ systems, with the most common being cone-rod dystrophy (93%), hypogenitalism in males (89%), obesity (72%), postaxial polydactyly (69%), neurological problems including developmental delay (62%) and speech deficits (54%). Other frequent associations include renal abnormalities, asthma, dental anomalies, congenital heart disease, and diabetes mellitus.[1] Hirschsprung's disease is thought to occur in about 2% of patients with Bardet-Biedl syndrome.[1]

Bardet-Biedl syndrome belongs to the category of non-Kallmann syndromic causes of congenital hypogonadotropic hypogonadism (CHH), a category that also includes Prader-Willi syndrome and Gordon Holmes syndrome. The other two groups are isolated CHH with normal olfaction, and Kallmann syndrome.[4]

Hirschsprung's disease is characterised by the absence of enteric neurons (aganglionosis) in the distal segments of the gut. It is usually nonsyndromic, but has been associated with chromosomal aberrations and few syndromes.[5]

The association of Bardet-Biedl syndrome with Hirschsprung's disease has been reported sporadically, and highlights the role of the primary cilium in enteric nervous system (ENS) development.[5-10]

The causative mutations for Bardet-Biedl syndrome have been mapped to about 16 genes (BBS1-BBS16), without clear genotype-to-phenotype translation and the lack of gene-specific disease symptoms makes the genetic diagnosis challenging, however developments in multigene sequencing technologies have led to breakthroughs.[11] The BBS proteins are components of the centrosome and affect ciliary transport, hence, the condition falls under the umbrella of ciliopathies.[12]

The condition was previously enveloped with Laurence-Moon syndrome as Laurence-Moon-Biedl-Bardet syndrome, but later dissociated as the Laurence-Moon syndrome patients were found to have paraplegia, but lacked the polydactyly and obesity characteristic of Bardet-Biedl patients.[13]

A similar association has been made between Alström syndrome and cecal volvulus.[14] Alström syndrome, a phenotypically related condition, is a rare autosomally recessively inherited condition characterised by childhood blindness (cone-rod dystrophy), dilated cardiomyopathy, sensorineural hearing loss, fibrotic lung disease, and metabolic abnormalities including obesity and insulin resistance. The absence of certain findings (e.g., polydactyly, intellectual disability) distinguishes Alström syndrome from Bardet-Biedl syndrome.[14] Other phenotypically similar conditions include Wolfram, Cohen, Biemond II, Usher, and Joubert syndromes.[11]

In our case, the patient presented due to sigmoid volvulus. Volvulus of the intestine is a surgical emergency, with sigmoid being the most frequently involved site, and redundant colon the most common underlying cause.[15] The typical radiographic finding is described as "bent inner tube" or "coffee-bean" appearance. The preferred approach in stable patients is elective sigmoid resection after adequate bowel preparation, however in emergency settings, resection and anastomosis with colostomy as per the intraoperative findings is indicated. Minimally invasive approach is gaining popularity, however is limited by peritoneal signs, severe distention and surgeon's experience.[15,16]

In our case, large-volume resection of redundant bowel followed by colostomy preparation was performed. The resection specimen demonstrated increased fibrosis, however the picture was obscured by acute inflammatory and edematous changes so a specific histological diagnosis was not possible.

CONCLUSION

This association of sigmoid volvulus in the patient with Bardet-Biedl syndrome does not of course prove cause and effect. However given the established association of Hirschsprung's disease with Bardet-Biedl syndrome, and the usual early mortality of affected patients, it is possible that intestinal obstruction is indeed part of the wide

clinical spectrum of the syndrome that even to this day is not fully characterised.

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