Intestinal malrotation in monozygotic twins; the asymptomatic twin should be screened: A case report and review

Daniel E. Levin, Jamil A. Matthews, Christa N. Grant, Ryan G. Spurrier, Akemi L. Kawaguchi

Division of Pediatric Surgery, Children’s Hospital Los Angeles, 4650 Sunset Blvd, Mailstop #100, Los Angeles, CA 90027, USA

A R T I C L E  I N F O

Article history:
Received 27 November 2013
Accepted 22 January 2014

Key words:
Intestinal malrotation
Volvulus
Monozygotic
Twin
Screening

A B S T R A C T

Intestinal malrotation occurs in approximately 1 per 1200 live births. However, it is not known if monozygotic twins have a differential risk for malrotation. Therefore, it is unclear whether an asymptomatic identical twin of an infant with known intestinal malrotation should undergo a formal screening evaluation. We describe a case of the diagnosis of malrotation in an asymptomatic identical twin of a baby who had been treated for malrotation with volvulus and review the relevant medical literature.

© 2014 The Authors. Published by Elsevier Inc. Open access under CC BY-NC-SA license.

Intestinal malrotation is the most significant risk factor for midgut volvulus in the pediatric population. If diagnosed prior to the onset of symptoms, the risk for midgut volvulus may be decreased with a Ladd’s procedure. There is little guidance in the medical literature regarding which asymptomatic patients should be screened and have a surgical intervention. The utility of screening the asymptomatic identical twin of a child diagnosed with intestinal malrotation has not been previously described. Prior case reports have suggested a strong concordance of intestinal malrotation between identical twins [4–7]. We report a case of identical twin males with intestinal malrotation as well as a review of the medical literature regarding twins with malrotation.

1. Case report

Monochorionic, diamniotic males were born at 32 weeks gestation to a gravida 5, para 3 mother and noted to have discordant growth at birth (twin A, 2.7 kg; twin B, 1.8 kg). Twin A was admitted to a community neonatal intensive care unit for feeding intolerance. Multiple attempts to start enteral feeds failed secondary to intermittent bilious orogastric residuals and persistent emesis. Plain radiographs were not consistent with obstruction. Initial upper gastrointestinal series (UGI) and pyloric ultrasound were interpreted as normal. Subsequently, the patient was transferred to our tertiary children’s hospital for evaluation. Repeat UGI (Fig. 1A) performed and interpreted by a pediatric radiologist demonstrated malrotation with volvulus. The patient underwent an urgent open Ladd’s procedure. Intraoperative findings included a 360-degree volvulus with venous dilatation and chylous ascites, consistent with a chronic volvulus without evidence of ischemia or infarction. Twin B was healthy and had no feeding difficulties. Because of twin A’s diagnosis, he was screened for malrotation at 4 months of age. UGI demonstrated intestinal malrotation without evidence of volvulus (Fig. 1B). He underwent an open Ladd’s procedure and was discharged home without complications. At four years follow up, both patients are thriving and have required no additional interventions.

2. Discussion

Intestinal malrotation among twins is rare and exists only as case reports in the literature. The clinical incidence of malrotation is estimated to be 1 in 6000 live births and may occur as an isolated condition or in association with other anomalies [1]. However, radiographic evidence of malrotation has been demonstrated to be 1 in 500 while autopsy studies suggest the actual incidence may be
as high as 1 in 100 [2,3]. The incidence of identical twin malrotation has been estimated to be 0.3 to 1 per million live births [4].

A search of the English language medical literature identified five prior reports of monozygotic twins with intestinal malrotation. In 1977, Kickuchi et al. first described a pair of monozygotic twins who both presented with bilious emesis in the first week of life secondary to intestinal malrotation with volvulus. Both patients underwent a successful Ladd’s procedure [4]. A later report described a set of identical twins with both intestinal malrotation and intestinal atresia with an associated type IV jejunal atresia that was thought to be associated with maternal anaphylactic shock that occurred during the first trimester [5]. Both infants underwent a jejunooileostomy and a Ladd’s procedure. Two additional descriptions of identical twins with malrotation were published in the radiology literature in the 1990s. In the first report, the twins were born at 36 weeks gestation and both infants presented with emesis on the first day of life [6]. UGI showed malrotation with volvulus in both babies and they each underwent a Ladd’s procedure. Crowley and Bawle next described a clinical scenario similar to our case. One twin developed midgut volvulus and underwent an urgent Ladd’s procedure. This prompted a UGI evaluation of the asymptomatic twin, who was diagnosed with malrotation and received a successful prophylactic Ladd’s procedure [7]. Only Smith describes an instance of monozygotic twins with discordant intestinal rotation. In that case, one of the twins presented with malrotation and midgut volvulus requiring urgent exploration and lysis of Ladd’s bands. However, the asymptomatic twin was noted to have normal rotation on a screening UGI [8]. These case reports are summarized in Table 1.

Although it is known that intestinal malrotation occurs as a result of failure of normal embryologic bowel rotation and fixation [9], the exact cause remains elusive. Some reports in the literature point to a possible genetic basis of intestinal malrotation. At least two families have been identified with a likely autosomal dominant pattern of inheritance as evidenced by the diagnosis of malrotation in multiple family members over 2–3 generations [10,11]. Additional families with non-twin siblings diagnosed with malrotation have been identified [12,13]. At times, the diagnosis of intestinal malrotation is associated with other conditions such as facial anomalies [12] or jejunal atresia [13]. Martin and Shaw-Smith recently published a comprehensive review regarding the genetic etiology of intestinal malrotation. In their review of the literature, they identified 18 separate syndromic conditions with intestinal malrotation as one of the defining anomalies [14]. Moreover, researchers have begun to identify gene mutations associated with the failure of normal midgut rotation. Heterozygous mutations in the forkhead transcription factor FOXF1 lead to the development of intestinal malrotation in addition to lethal lung abnormalities [15]. With a possible genetic etiology for intestinal malrotation, it would be expected that monozygotic twins would have a higher concordance rate than would be predicted by random association.

### 3. Conclusion

Intestinal malrotation poses a significant risk of midgut volvulus, with intestinal ischemia, short gut syndrome, and even death as potential sequela if left unrecognized and untreated. Symptoms of midgut volvulus may present at any age [9], but are most common in the first month of life when discerning symptoms is more difficult [1]. Prompt recognition of intestinal malrotation and performance of a Ladd’s procedure are the standard of care to minimize the risk of midgut volvulus [16,17]. An early diagnosis of malrotation may allow early intervention and help prevent the morbidity associated with midgut volvulus.

A possible genetic basis of malrotation, combined with several reports of concordance of intestinal malrotation in monozygotic twins, suggest that screening an asymptomatic twin is indicated.

### Table 1

Reports of identical twins with intestinal malrotation, both concordant and discordant.

<table>
<thead>
<tr>
<th>Author</th>
<th>Year</th>
<th>Twin A Malrotated</th>
<th>Twin B Malrotated</th>
<th>Diagnosed by screening UGI</th>
</tr>
</thead>
<tbody>
<tr>
<td>Kikuchi et al.</td>
<td>1977</td>
<td>Yes</td>
<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>Olson et al.</td>
<td>1987</td>
<td>Yes</td>
<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>Burton et al.</td>
<td>1993</td>
<td>Yes</td>
<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>Crowley et al.</td>
<td>1995</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
</tr>
<tr>
<td>Smith et al.</td>
<td>2006</td>
<td>Yes</td>
<td>No</td>
<td>Yes</td>
</tr>
<tr>
<td>This report</td>
<td>2013</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
</tr>
</tbody>
</table>

UGI, upper gastrointestinal.
Although we recognize the additional cost and potential risk of radiation from a screening UGI, the risk of the complications of midgut volvulus are severe and an opportunity to intervene should not be missed.

References


