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Colon atresia and frontal encephalocele: a rare association

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Abstract The association of colonic atresia with craniofacial anomalies has been well described and probably represents a malformative event that occurs in the early embryonal period. We present a case of an infant with colonic atresia and a frontal encephalocele and believe this to be a newly reported association. We review possible pathogenic mechanisms.

Colonic atresia (CA) is a rare anomaly that presents with acute intestinal obstruction in the neonatal period. It occurs less frequently than jejunoileal atresia (2%-10% of all intestinal atresias [1]), although the exact incidence has not been defined. In the northwest of England, the incidence has been reported to be 1 in 66 000 live births [2].

The incidence of other anomalies is high, and in the largest review of patients with CA [3], 106 (47%) of 224 patients had 1 or more anomalies. Midline abdominal wall defects are by far the commonest association (approximately 25%), and jejunoileal atresia may coexist in 15% to 20% [4,5]. Extraabdominal anomalies of the musculoskeletal and cardiovascular system are also relatively frequent and can contribute significantly to morbidity [4,5]. Facial malformations (facial hemiaplasia or hemiatrophy [6-9]) and ocular anomalies have been also reported [5,7,9-14], as well as central nervous system malformations [1,11]. We report a case of CA, associated with a frontal encephalocele, which is, to our knowledge, the first such case reported in the literature.

1. Case report

A 2.7-kg term male neonate was born at 38 weeks gestation. The pregnancy had been normal, and there was no history of varicella exposure or other infections. Abdominal distension, bile vomiting, the absence of meconium in a rectal washout and dilated bowel loops prompted a contrast enema, which showed a microcolon with cessation of filling in the left upper quadrant (Fig. 1).

At laparotomy, a type III cecal atresia was found. The remainder of the ascending colon and the transverse colon were absent. The distended cecum was excised, and an ileostomy and mucous colonic fistula were formed. It was noted that there was a 2-cm protruding lesion in the

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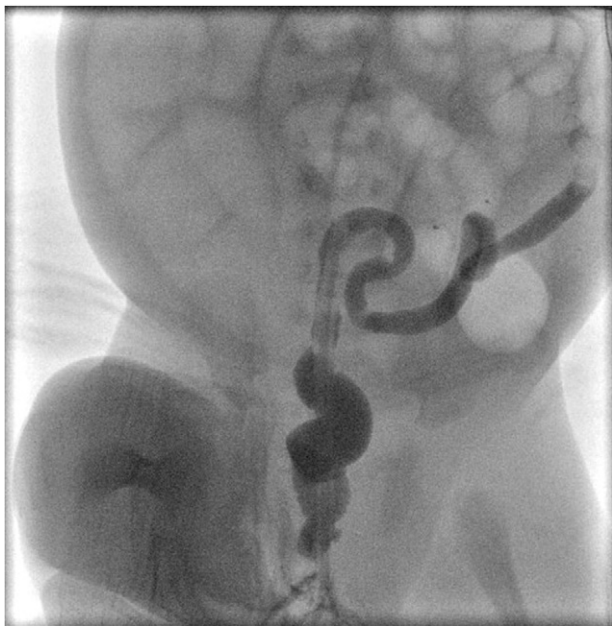

Fig. 1 Colonic atresia. Contrast enema: microcolon and cessation of filling at the level of the left upper quadrant.

left supraorbital frontal region with a palpable osseous defect (Fig. 2). Subsequent magnetic resonance imaging and 3-dimensional computed tomography reconstruction of the skull established the diagnosis of a frontal encephalocele with an osseous deficit (Fig. 3). No other congenital anomalies were evident.

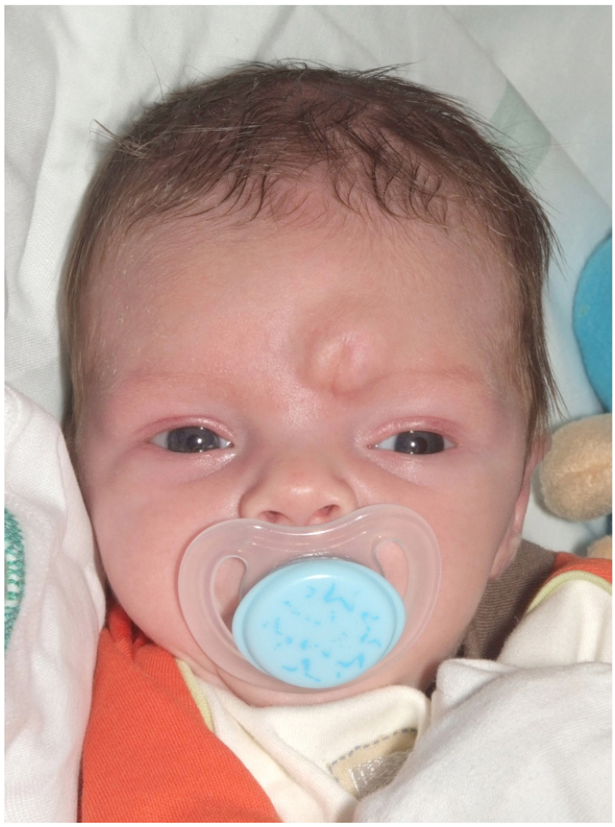

Fig. 2 Frontal facial view showing a left frontal protruding lesion proved by imaging to be a frontal encephalocele.

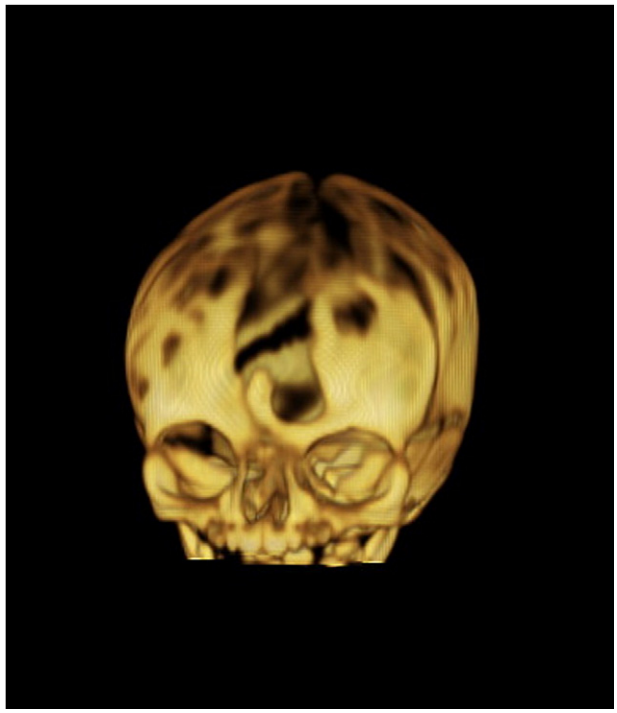

Fig. 3 Three-dimensional computed tomographic reconstruction image showing the osseous defect at the left part of the frontal bone.

Histopathology showed the absence of lanugo, bile pigments, or squamous pellets in the distal colon. Rectal suction biopsies were negative for Hirschsprung disease. The patient was discharged on the tenth postoperative day. Surgery to restore intestinal continuity is planned for approximately 6 months of age, and encephalocele repair, approximately 1 year of age.

2. Discussion

The theory of an embryologic vascular insult resulting in intestinal atresia is certainly attractive and has been reproduced in animal models [13,14]. However, it fails to provide an etiological mechanism for the frequent association of CA with other congenital anomalies, both intestinal and extraintestinal anomalies [2]. It is possible, therefore, that the pathogenesis of CA could have more than 1 mechanism, accounting for the variety of phenotypes [15,16]. The studies of Puri and Fujimoto [17], Fourcade et al [18], and Fairbanks et al [19] support the theory of disturbed morphogenesis in the embryonal period. There is also a rare association of CA with Hirschsprung disease (approximately 2%-5% of CA cases) [20,21], again illustrating a more complex etiology for CA. Finally, Baglaj et al [22] attributes the association of CA and gastroschisis to