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ORIGINAL ARTICLE

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Double bubble, double trouble

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Abstract Duodenal atresia (DA) is not uncommon, either as an isolated anomaly or associated with trisomy 21, malrotation, or cardiac anomalies. It may be diagnosed on antenatal ultrasound by a "double-bubble" sign, which typically persists after birth on a plain abdominal radiograph. DA as a familial association is rare but has been reported with or without other associated anomalies. We report DA in two siblings of nonconsanguineous parents, one case occurring with an annular pancreas in association with gestational diabetes. These two cases suggest possible genetic and environmental components in the aetiology of this anomaly.

Keywords Duodenal atresia · Familial · Annular pancreas

Introduction

Duodenal atresia (DA) is not uncommon, with an incidence of about 1:10,000 [9]. It may be suggested on antenatal ultrasound, usually during the second trimester, with polyhydramnios in association with a dilated stomach and duodenal bulb. A plain abdominal radiograph following delivery usually shows the classic "double-bubble" sign due to air within the stomach and the proximal duodenum, with no gas in the distal bowel. Most commonly, DA involves the second part of the duodenum. Annular pancreas has been associated with duodenal atresia and sometimes is referred to as a wedge or interposed pancreas [10]. DA may be associated with other congenital abnormalities including trisomy 21, malrotation, and cardiac anomalies. The occurrence of DA as a familial association is extremely rare, especially amongst siblings. The first report of familial duodenal atresia appeared in 1959 by Pequet and Watson [11], and since then there have been six other reports in the English language literature.

We report two siblings in a family with duodenal atresia, one of which had an associated annular pancreas.

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Case report

Case 1

A female neonate weighing 3317 g was delivered vaginally at 41 weeks of gestation without complication. The child was born to a 25-year-old mother and was her second child. The mother had gestational diabetes that was controlled with diet alone. Her first pregnancy had been normal, and there was no consanguinity in the family. The mother was not on any medications or drugs during the pregnancy. Antenatal ultrasound scans performed at 13 and 19 weeks of gestation were normal. The infant developed bilious vomiting on the 1st day of life. Examination revealed no obvious congenital anomalies. A contrast study, which was performed to exclude malrotation, revealed a dilated stomach and proximal duodenum with a stenosis involving the third part of the duodenum (Fig. 1). At laparotomy a duodenal web was found. This was incised, and a standard duodenoduodenostomy was performed. Oral feeds were commenced 3 days following surgery. The infant was discharged on day 9 with a weight of 3259 g. She is now 4 years old and thriving.

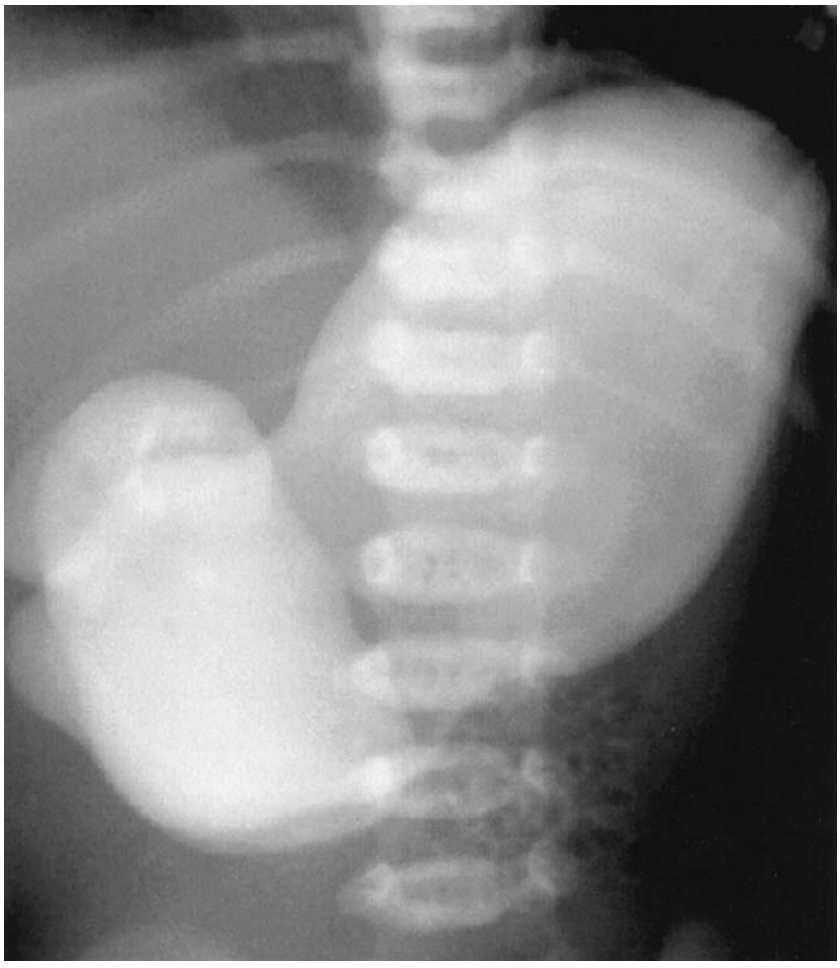

Fig. 1 Upper gastrointestinal contrast study of case 1 revealing dilated stomach and duodenum with failure of the contrast to progress beyond the third part of the duodenum, consistent with a congenital duodenal obstruction

Case 2

A male neonate weighing 2535 g was delivered vaginally at full term after induction of labour due to gestational diabetes. He was born to the same parents, the mother now 29 years old. This was her third pregnancy. The first antenatal scan at 18 weeks was normal, but it was repeated at 32 weeks because of suspected polyhydramnios. This was confirmed in addition to a double-bubble sign consistent with the diagnosis of duodenal atresia. Following delivery, a plain abdominal radiograph indicated the double-bubble sign, whilst the rest of the abdomen was gasless (Fig. 2). Clinically there were no other detectable abnormalities, and an echocardiogram was normal. At operation, duodenal atresia was confirmed and was noted to be associated with an annular pancreas at the second part of the duodenum. A duodenoduodenostomy was performed, and a transanastomotic tube was inserted for feeding. The remainder of the gastrointestinal tract was normal. Feeds were started after 2 days postoperatively via the transanastomotic tube. The infant was discharged 14 days following surgery and remains well at 4 months.

Discussion

DA with or without other congenital anomalies [5, 7] is not uncommon. A number of mechanisms have been proposed to suggest the pathogenesis of duodenal atresia, including failure of recanalisation of the duodenal lumen and vascular disruption [14]. One study has suggested abnormalities in neural crest cell migration in conjunction with vascular disruption as possible pathogenic mechanisms [6].

Familial association of duodenal atresia is very rare, although isolated cases have been reported that suggest a possible genetic association. The first reported familial case of DA was attributed to Pequet and Watson [11]. Boyden et al. [3] suggested that there must be a genetically transmissible factor, but the genetic basis and its mode of transmission remain unclear. In a study of two nonrelated families with duodenal atresia, three siblings in one family had associated Fanconi's syndrome, and all died due to overwhelming fungal sepsis [2]. In the second family, two siblings with duodenal atresia had overwhelming sepsis with T-cell dysfunction, suggesting another form of immune deficiency associated with DA

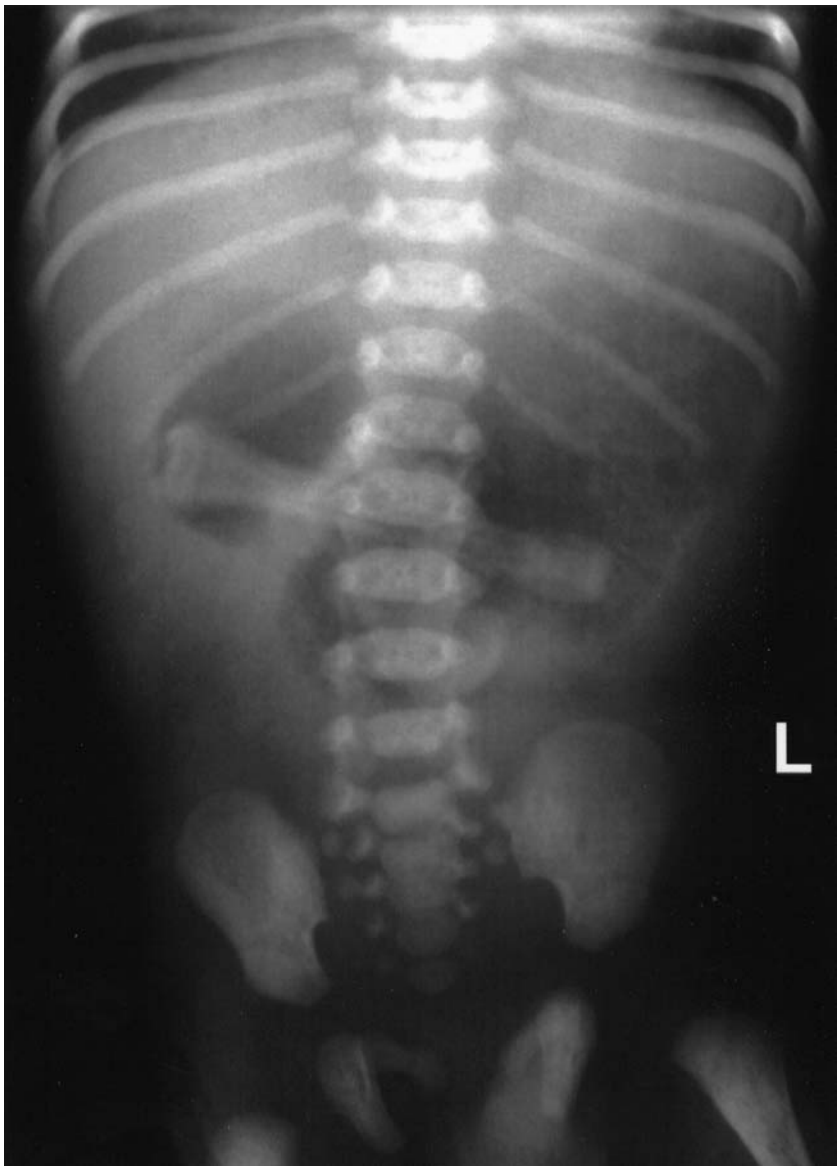

Fig. 2 Plain abdominal radiograph of case 2 revealing air-filled stomach and proximal duodenum ("double bubble") with an absence of distal bowel gas consistent with duodenal atresia

[8]. Another report described four cases of duodenal atresia in which two siblings from one family had duodenojejunal atresia associated with malrotation, volvulus, and absent parietal attachment of mesentery, suggesting a familial occurrence as an autosomal recessive inheritance condition [12].

There have been cases of other anomalies associated with duodenal atresia amongst siblings, including trisomy 21. Familial cases have been reported in association with parental consanguinity, suggesting an autosomal recessive pattern of inheritance. Isolated DA in two families, with consanguineous marriage in one family, has also been reported [8]. Another study involved four pedigrees, including a three-generation Dutch family with 11 affected members with Feingold syndrome. This is a rare autosomal dominant disorder with digital abnormalities, microcephaly, short palpebral fissures, mild learning disability, and oesophageal/duodenal atresia [4]. Genetic analysis indicated haploid insufficiency of a gene or genes in 2p23-p24.

Congenital anomalies in general can be affected by other factors, including the environment. In our cases, gestational diabetes was present and may have been a factor in both. One study showed that in mothers with preexisting diabetes, the total malformation rate was 9.5%, whereas the rate in the gestational diabetic group was similar to the normal population rate of 5.7%. There were, however, certain anomalies in the infants of gestational diabetic mothers that were found to be more frequent, suggesting that there may be a subgroup with increased risk [1]. Shaefer-Graf et al. [13] also found that the frequency of congenital anomalies was increased in both gestational and preexisting diabetic mothers.

We report two siblings with DA of unrelated parents. The first infant had an isolated duodenal web, and the second had DA in association with an annular pancreas.

This sequence has not previously been reported in the absence of consanguinity [8]. The mother had gestational diabetes in both pregnancies. This may suggest a genetic component in the aetiology of at least a proportion of cases of DA, with the phenotype perhaps triggered by gestational diabetes. Further prospective analysis, including a detailed antenatal history, of infants with DA may assist in determining the true significance of this association and the contribution of maternal diabetes.

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