Familial Duodenal Atresia : A Report of Two Siblings†

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Abstract

The familial occurrence of duodenal atresia is extremely uncommon. The author reports duodenal atresia occurring in two siblings who underwent successful surgical repair at the Pediatric Surgical Unit, Department of Surgery, Ratchaburi Hospital. To the author's knowledge, this is the first report of familial duodenal atresia in Thailand.

Key word : Familial, Duodenal Atresia

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Duodenal atresia is an uncommon congenital malformation, and its occurrence in the same family is extremely rare. Very few reports have been published on familial duodenal atresia (Hyde, 1965 ; Berant and Kahana, 1970 ; Best, Wiseman and Chudley, 1989 ; Gahukamble, Khamage and Shaheen, 1994)[1-3]. To the author's knowledge no instance of familial duodenal atresia has been reported in Thailand. The author wishes to report the occurrence of duodenal atresia in two siblings.

CASE REPORTS

Case 1

The patient was a 2,600 g normal female infant delivered after a 40–week uncomplicated gestation, labor, and delivery at Ratchaburi Hospital in January 3, 1999. She appeared normal at birth, but her mother had polyhydramnios during the last trimester of pregnancy. The mother was a healthy 32–year–old secondpara and the father was 32 years old. No consanguinity was identified.

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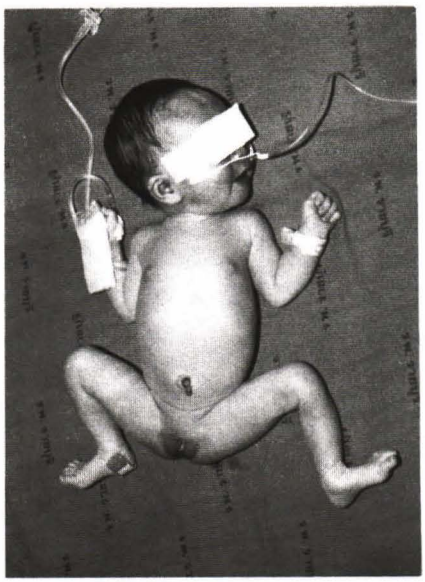

Fig. 1. Case 1 at the age of 6 hours.

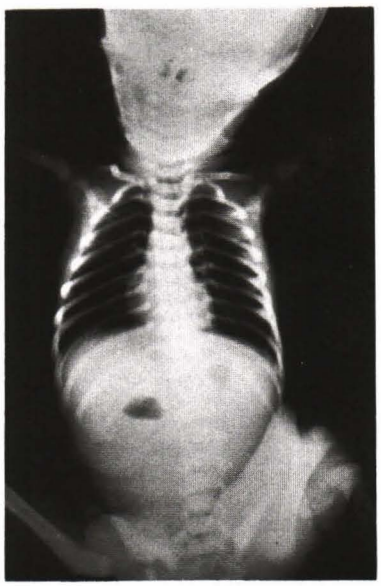

Fig. 2. Plain upright radiograph of the infant in case 1 shows double bubble appearance.

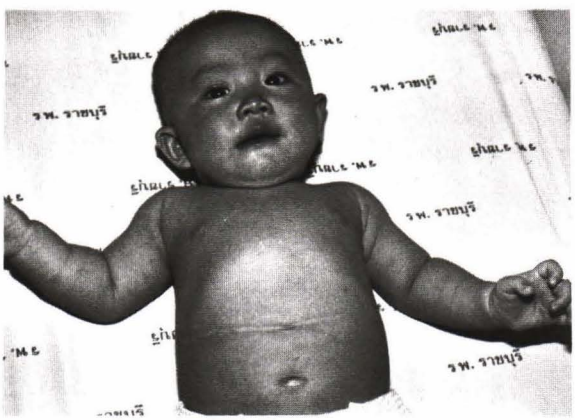

Fig. 3. Case 1 at the age of 6 months.

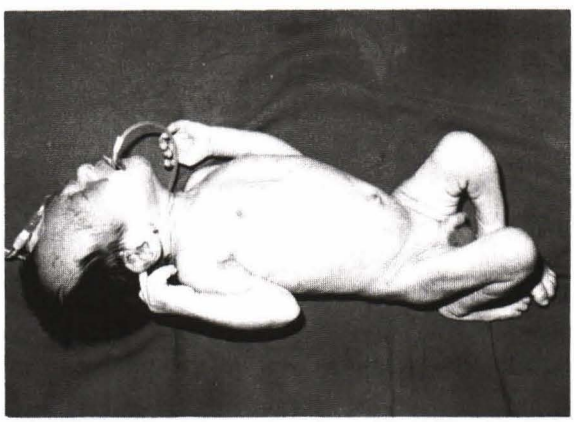

Fig. 4. Case 2 at the age of 16 hours.

At the age of 6 hours the infant had bilious emesis and was transferred to the Neonatal Special Care Unit (Fig. 1). Mild jaundice was noted and the abdominal examination showed upper abdominal fullness. Plain X–rays of the abdomen showed a typical "double bubble" appearance (Fig. 2).

During laparotomy, the duodenal atresia was found to be situated just distal to the Ampulla of Vater. The atresia was diaphragmatic in type. After duodenotomy, duodenal diaphragm was excised and duodenoplasty was performed. Oral feeding was tried in the fourth postoperative day and she was discharged at the age of 15 days.

Fig. 5. Plain upright radiograph of the infant in case 2 shows double bubble appearance.

Follow–up at 6 months showed a normal healthy child (Fig. 3, 7).

Case 2

The patient was a 2,200 g normal male infant delivered after a 35 – week uncomplicated gestation. He was delivered by vacuum extraction at Ratchaburi Hospital in January 1, 1998. He is the brother of the first case. This newborn had problems similar to those of case 1. The mother was a healthy 31–year–old primipara but she had polyhydramnios during the last trimester of pregnancy.

At the age of 16 hours the infant had bilious emesis and was transferred to the Neonatal Special Care Unit (Fig. 4). Plain X–ray of the abdomen showed a typical "double bubble" appearance (Fig. 5).

During laparotomy, atresia of the second and third portions of the duodenum was confirmed with normal rotation of the bowel. A diamond–shaped duodenoduodenostomy was carried out without complications. He was discharged at the age of 23 days. Follow–up at 18 months showed a normal healthy child (Fig. 6, 7).

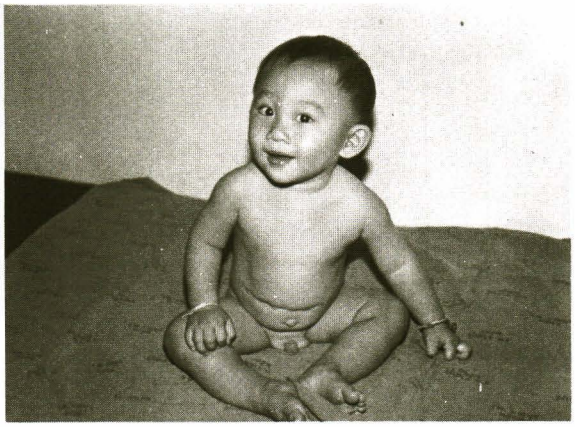

Fig. 6. Case 2 at the age of 18 months.

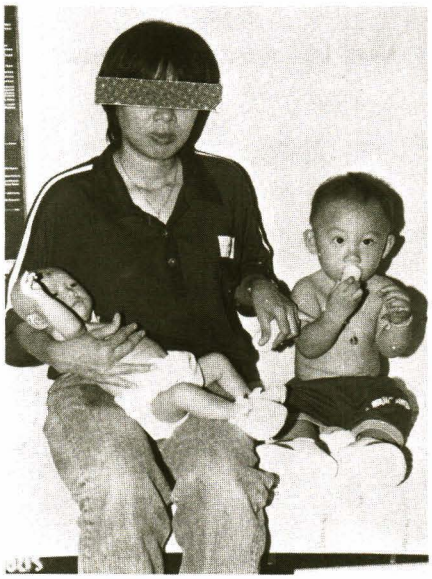

Fig. 7. Case 1 and case 2 with their mother at the age of 6 and 18 months.

DISCUSSION

Hyde (1965) reported the first cases of familial duodenal atresia in a sibship of 4 affected sibs of non-consanguineous Greek immigrant parents in the United States[2]. Berant and Kahana (1970) described 3 affected sibs, a boy and 2 girls, born to first-cousin parents from Israel[1].

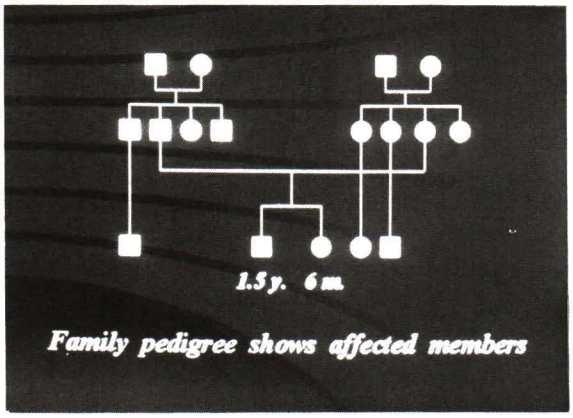

Fig. 8. Family pedigree shows affected members.

Best, Wiseman and Chudley (1989) reported 2 families of familial duodenal atresia[2]. Gahukamble, Khamage and Shaheen (1994) described a family in which two sisters had proven duodenal atresia and one brother possibly died of complications from it[3].

The pathogenesis of duodenal atresia remains conjectural. Atresia is distinguished as being 1) primary (in which there is failure of recanalization of the solid embryonic duodenum, which occurs normally between the 8th to 10th week (Tandler, 1900)) or 2) secondary or extrinsic (as a result of compromise of vascular supply to a bowel segment, or due to other intestinal pathology (Louw and Bernard, 1955)). The former mechanism is the more likely pathogenetic mechanism, whereas secondary atresia likely explains bowel atresia distal to the duodenum[4-20]. In the cases of familial duodenal atresia in the literature, including the two siblings of this report, may support the hypothesis that an autosomal-recessive gene is responsible for the incomplete recanalization of the duodenum, which can lead to duodenal atresia (Fig. 8)[1-3,5,6,9,16].

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ดูโอดีนั่มตันในครอบครัวเดียวกัน : รายงานผู้ป่วยสองพี่น้อง†

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ดูโอดีนั่มตันที่เกิดในครอบครัวเดียวกันพบได้น้อยมาก ผู้รายงานได้รายงานผู้ป่วยดูโอดีนั่มตันในพี่น้องสองคนซึ่งได้รับการผ่าตัดเป็นผลสำเร็จที่แผนกศัลยกรรมเด็ก กลุ่มงานศัลยกรรม โรงพยาบาลราชบุรี เท่าที่ผู้รายงานทราบรายงานนี้เป็นรายงานแรกของผู้ป่วยดูโอดีนั่มตันที่เกิดในครอบครัวเดียวกันในประเทศไทย

คำสำคัญ : ครอบครัว, ดูโอดีนั่มตัน

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