

## Familial occurrence of congenital bile duct dilatation

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### Abstract

Congenital bile duct dilatation (CBD) that developed in a parent and son is presented. Familial occurrence of CBD is rare, with only a few male cases having been reported. Since the initial report of CBD occurring in siblings in 1981, a total of 20 cases (10 pairs) have been published as of 2007. Clinical and genetic features of CBD are discussed.

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**Key words:** Congenital bile duct dilatation; Familial occurrence; Choledochal cyst; Choledochoceles; Anomalous pancreaticobiliary duct junction

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### INTRODUCTION

Choledochal cyst is a rare congenital dilatation of the extrahepatic and/or intrahepatic biliary tract. Some

possibilities have been postulated regarding inheritance of congenital bile duct dilatation (CBD), since several familial CBD cases have been reported. However, the etiology of CBD is essentially unknown. In this report, a case of familial occurrence of CBD is presented along with a review of the literature.

### CASE REPORT

Case 1 was a 65-year-old woman, the mother of case 2, who was admitted to Matsusaka City Hospital, Japan, complaining of back pain and fever. Serum amylase level was normal. Liver function impairment was noted: aspartate aminotransferase (AST) 216 U/L, alanine aminotransferase (ALT) 228 U/L, and total bilirubin 1.3 mg/dL. A CT scan revealed a cystic dilatation of the common bile duct. With the diagnosis of choledochal cyst, the patient underwent cholecystectomy, resection of the dilated bile duct, and hepaticojejunostomy. Intraoperative cholangiopancreatography showed extrahepatic bile duct dilatation and an anomalous pancreaticobiliary duct junction (APBDJ) (Figure 1A and B). Amylase level in the bile was 4735 U/L. Microscopic examination of the resected specimen revealed no evidence of malignancy. The postoperative course was uneventful.

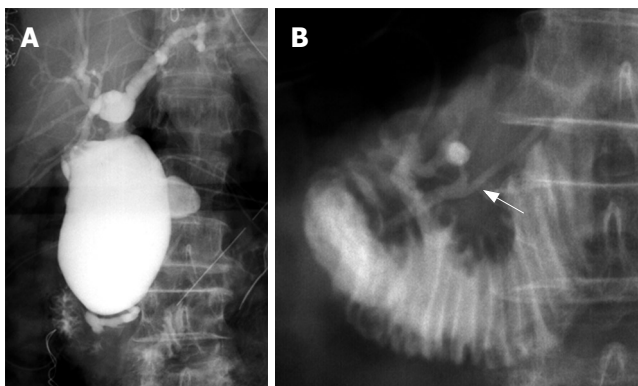
Case 2 was a 25-year-old man, the son of case 1, who was admitted to Mie University Hospital, Japan, with complaints of epigastric pain and fever. Amylase level in the serum and urine was 2639 and 56 720 U/L, respectively. Endoscopic retrograde cholangiopancreatography (ERCP) showed cystic dilatation of the extra- and intrahepatic biliary ducts associated with APBDJ (Figure 2). With the diagnosis of choledochal cyst, cholecystectomy, excision of the dilated bile duct, and hepaticojejunostomy were performed. Amylase level in the bile was 138 700 U/L. No malignancy was found histologically in the resected specimen. The patient was discharged without any complication.

### DISCUSSION

Congenital choledochal cyst is a relatively rare disease and its incidence is between 1 in 130 000 and 1 in 2 million<sup>[10]</sup>. There is a female predominance (ratio 1:3)<sup>[11,12]</sup> and the lesions are more common in Asians, with the majority of reported cases in Japan<sup>[11]</sup>. Choledochal cysts usually present in infancy or childhood, which supports a congenital origin. Although anomalous pancreaticobiliary duct junction (APBDJ) is thought to be an essential causative factor of CBD, the etiology of CBD remains unclear. Since Chiba *et al* first described in 1981 cases of CBD occurring in

Table 1 Reported cases of familial occurrence of congenital bile duct dilatation

Year	Authors	Combination	Age	Extrahepatic bile duct	Intrahepatic bile duct dilatation	Anomalous pancreaticobiliary duct junction (APBDJ)
1981	Chiba <sup>[1]</sup>	Elder sister	4 yr 10 mo	Fujiform	+	+
		Younger sister	3 yr 9 mo		+	+
1985	Iwama <sup>[2]</sup>	Mother	48 yr	Fujiform	-	+
		Daughter	11 yr	Cystic	-	unknown
1986	Akiyama <sup>[3]</sup>	Mother	34 yr		Unknown	unknown
		Daughter	3 mo	Cystic	+	unknown
1987	Date <sup>[4]</sup>	Elder brother	7 yr	Cystic	-	+
		Younger sister	10 yr	Fujiform	-	unknown
1990	Iwafuchi <sup>[5]</sup>	Mother	25 yr	Cystic	+	+
		Daughter	2 yr 7 mo	Fujiform	-	+
1996	Narita <sup>[6]</sup>	Mother	48 yr		Unknown	+
		Daughter	34 yr	Cystic	+	+
1998	Iwata <sup>[7]</sup>	Mother	33 yr	Choledochocoele	-	-
		Daughter	7 d	Cystic	Unknown	+
1999	Lane <sup>[8]</sup>	Dizygotic twins	3 yr	Cystic	-	+
		(Females)	3 yr	Fujiform	+	+
2005	Tokuhara <sup>[9]</sup>	Dizygotic twins	2 yr 4 mo	Fujiform	+	+
		(Females)	2 yr 6 mo	Cystic	-	+
2002	Our case	Mother	65 yr		-	+
		Son	25 yr	Cystic	+	+



**Figure 1** A: Intraoperative cholangiopancreatography, showing cystic dilatation of the common bile duct; B: Intraoperative cholangiopancreatography, showing an APBDJ (arrow).

siblings<sup>[1]</sup>, a total of 20 cases (10 pairs), including ours, of familial occurrence have been reported (Table 1). Eighteen patients (90%) were women and only two (10%) were men. Of these 10 pairs, four were siblings and six were parent and child. APBDJ was noted in 15 cases (75%). In addition, two pairs (20%) were dizygotic twins who both developed CBD. As for genetic background of CBD, Iwama *et al* have postulated two possible patterns of inheritance: an X-linked dominant trait or an autosomal dominant trait with relatively low penetrance in males<sup>[2]</sup>. The present case of CBD with APBDJ suggests a maternally inherited condition, i.e. an X-linked dominant trait. Besides our literature review, a total of six pairs of monozygotic twins have been described in which CBD appeared in one of each pair of twins<sup>[13-16]</sup>. The phenotypic discordance in monozygotic twins cannot be explained only by inheritance. Other factors, such as acquired or environmental conditions, might have a role in the development of CBD. To clarify the etiological basis of CBD, it will be necessary to accumulate more familial cases of CBD.



**Figure 2** ERCP showing dilatation of the extra- and intrahepatic bile ducts, as well as an APBDJ (arrow).

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