

Brief Clinical Report

Choledochal Cyst Associated With Rare Hand Malformation

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We report on an 8-year-old boy with choledochal cyst associated with most unusual hand malformation. Review of the literature and possible etiopathogenesis are discussed. © 1995 Wiley-Liss, Inc.

KEY WORDS: choledochal cyst, hand malformation, Catel-Manzke dysostosis

INTRODUCTION

Choledochal cysts are rare malformations. They have been reported in association with other gastrointestinal malformations. We report on a young boy with an exceptional hand malformation associated with choledochal cyst.

CLINICAL REPORT

An 8-year-old boy was admitted for vomiting, fever and abdominal pain for 3 days. On admission he had mild hepatomegaly, jaundice, right quadrant abdominal pain, and a symmetrical hand malformation. The index was medially deviated and located in a parallel position to the thumb. Both feet had a markedly shorter first toe. No other malformation was detected.

Initial investigation showed increased direct serum bilirubin, liver enzymes, alkaline phosphatase, and serum amylase.

The child was born at term to non-consanguineous parents (father's age 34 and mother's age 27 years). They had six other normal sibs. No limb malformation was reported in the immediate family and none of the siblings had similar complaint. The child came to medical attention at the age of 2 years for an episode of

fever, jaundice, and abdominal pain which was diagnosed as hepatitis. No neonatal jaundice was reported. Two similar episodes were reported at the age of 5 and 6 years.

The history and physical findings were highly suggestive of obstructive jaundice due to choledochal cyst. Abdominal ultrasound confirmed the presence of a cystic mass measuring 5 by 5 cm at the level of the common bile duct. The cyst was classified as Todani type Ib by the operative cholangiogram (Fig. 1).

Hand roentgenograms showed a symmetrical anomaly with shortness of the first metacarpals; the second metacarpals were composed of two separate bones with external deviation of the distal one (Fig. 2). Both first metatarsal bones were short giving to feet a particular aspect (Fig. 3). No other skeletal abnormality was detected.

The postoperative course was smooth and no operation was proposed to correct the hand abnormalities as the boy's functional hand capacities were excellent.

DISCUSSION

Choledochal cyst is a rare malformation generally diagnosed in children and young adults. Its incidence was estimated as 1 in 13,000 hospital admissions in the USA [Crittenden and McKinley, 1985] and 1 in 2,000,000 live births [Howell, 1983]. About 2w3 of reported cases are from Japan [Yamagushi, 1980].

The most commonly accepted contemporary classification of congenital choledochal and other bile-duct cyst is that of Todani [Todani et al., 1977] which is an adaptation of the original categorization of Alenso-Lej [1959].

The cause of choledochal cyst is not yet settled. The extrahepatic biliary system forms between the fifth and the seventh week of intrauterine life [Linder, 1964]. Choledochal cyst may be the result of an embryonic malformation causing "weakness" of the wall of the bile duct. This weakness is coupled with a distal obstruction to produce the typical cystic anomaly [Alenso-Lej et al., 1959; Kasai et al., 1970]. Yotuyanagi postulated an inequality of proliferation of epithelial cells of the bile duct between the distal and proximal parts which,

Received for publication March 25, 1994; revision received October 17, 1994.

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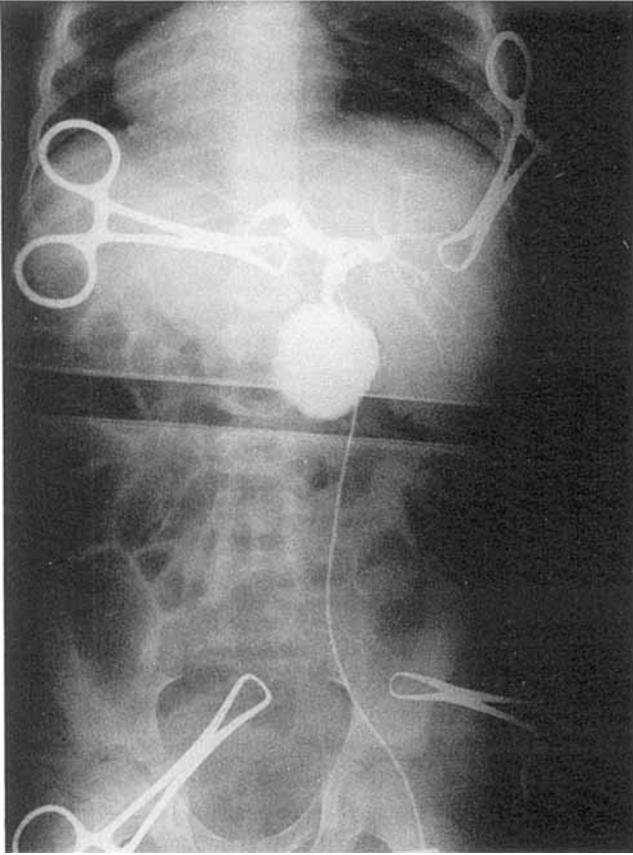


Fig. 1. Operative cholangiography. Choledochal cyst type Ib.

when recanalization occurs, will lead to a dilated proximal part and to a relatively narrower (or in severe cases atretic) distal part [Yotuyanagi, 1912]. An anomalous arrangement of the pancreatic duct with the common bile duct was proposed as an alternative explanation for the development of the choledochal cyst [Babbit et al., 1969].

Cases of annular pancreas [Nunez-Hoyo et al., 1982], duodenal atresia [Barlow et al., 1976], malrotation [Yamagushi, 1980; Babbit, 1969; Nunez-Hoyo et al., 1982; Grosdidier et al., 1974] have been described in association with choledochal cysts. Extra-intestinal malformations such as renal agenesis or polycystic kidneys [Bottger, 1951], or thoracic hemivertebrae [Kirwan, 1974] are rare associations in choledochal cyst cases.

To our knowledge, Witzel in 1880 [quoted by Bottger, 1951] described the only case with associated choledochal cyst and hand malformation. He reported on a newborn infant with choledochal cyst and polydactyly of hands and feet; the left hand also had a seventh rudimentary finger.

The hand anomaly described in this child can be classified as Catel-Manzke dysostosis. The initial detailed description of this anomaly in association with Pierre Robin sequence is due to Manzke [1966]. Further cases

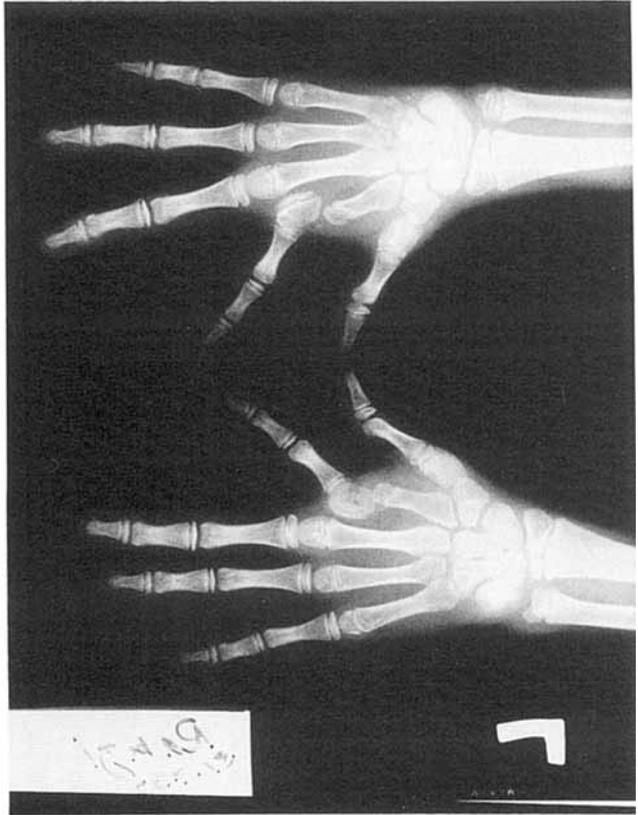


Fig. 2. Catel-Manzke anomaly of both hands.

have been described in association with cleft palate [Gewitz et al., 1978], Pierre Robin sequence [Holt-husen, 1972; Sundaram et al., 1982; Brude, 1984; Dignan et al., 1986], heart malformation [Stevenson et al., 1980], and dislocatable knees [Thompson et al., 1986]. An isolated similar hand malformation in mother and daughter was reported [Holmes and Remensnyder, 1972]. Two half brothers by the same mother with Robin sequence and digital anomalies were described by Chitayat et al.: patient 1 had hypoplastic distal phalanges and supranumerary ossification center at the base of the second metacarpal bones [Chitayat et al., 1991]. None of these reports mention the presence of choledochal cyst in association of the hand malformation in the proband or his relatives.

The mode of inheritance of Catel-Manzke dysostosis is not settled yet. X-linked recessive inheritance was suggested in cases reported in association with Robin sequence [Brude, 1984]. The association of choledochal cyst and Catel-Manzke malformation could be fortuitous. This possibility is very unlikely as both malformations are extremely rare. We strongly suggest that choledochal cysts, as other malformations described in association with Catel-Manzke syndrome, constitute possibilities of expression of the same gene responsible of the Catel-Manzke anomaly.

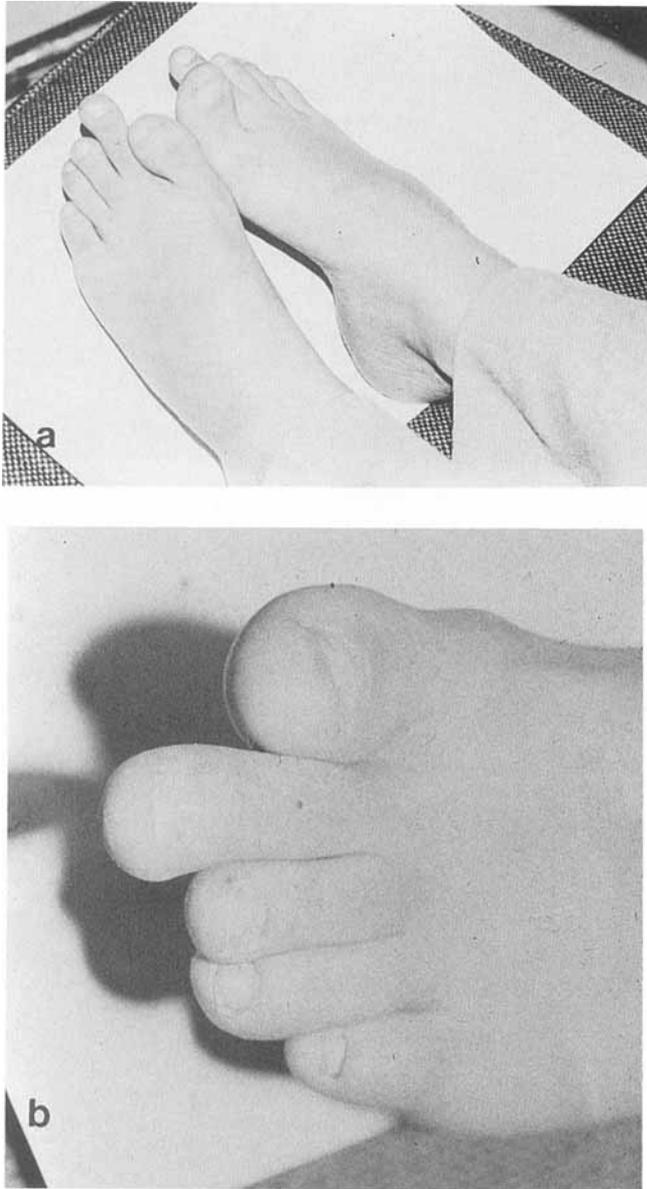


Fig. 3. a,b: Symmetrical anomaly of feet.

REFERENCES

Alonso Lej F, Rever WB Jr, Pessagno DJ (1959): Congenital choledochal cyst with a report of 2 and analysis of 94 cases. *Int Abstr Surg* 108:1-30.

Babbit DP (1969): Congenital choledochal cysts: New etiological concept based on anomalous relationships of the common bile duct and pancreatic bulb. *Ann Radiol* 12:231-240.

Barlow B, Tabor E, Blanc WA, Santulli TV, Harris RC (1976): Choledochal cyst: A review of 19 cases. *J Pediatr* 89:934-940.

Bottger H (1951): Zur atologie der choledochuszysten. *Zbl Allg Path.* 87:407-412.

Brude E (1984): Pierre Robin sequence and hyperphalangy: A genetic entity (Catel Manzke syndrome). *Eur J Pediatr* 142:222-223.

Chitayat D, Meunier CM, Hodgkinson KA, Azouz ME (1991): Robin sequence with facial and digital anomalies in two half-brothers by the same mother. *Am J Med Genet* 40:167-172.

Crittenden SL, Mckinley MJ (1985): Choledochal cyst-clinical features and classification. *Am J Gastroenterol* 80:643-647.

Dignan PStJ, Martin LW, Zenni EJ Jr (1986): Pierre Robin anomaly with an accessory metacarpal of the index finger: The Catel-Manzke syndrome. *Clin Genet* 29:168-173.

Gewitz M, Dinwiddie R, Yuille T, Hill E, Carter CO (1978): Cleft palate and accessory metacarpal of index finger syndrome: Possible familial occurrence. *J Med Genet* 15:162-164.

Grosdidier J, Boissel P, Richaume B, Baudesson D (1974): Resection du kyste et reimplantation hepatico-duodenale termino-laterale dans la dilatation kystique congenitale de la voie biliaire principale. *J Chir(Paris)* 107:117-124.

Holmes LB, Remensnyder JP (1972): Hypoplasia of the second metacarpal in mother and daughter. *J Paediatr* 81:1165-1167.

Holthusen W (1972): The Pierre Robin syndrome:unusual associated developmental defects. *Ann Radiol (Paris)* 15:252-262.

Howell CG, Templeton JM, Weiner S, Glassman M, Betts JM, Witzleben CL (1983): Antenatal diagnosis and early surgery for choledochal cyst. *J Pediatr Surg* 18:387-393.

Kasai M, Asakura Y, Taira Y (1970): Surgical treatment of choledochal cysts. *Ann Surg* 172:844-851.

Kirwan WO (1974) : Choledochal cyst. *Br J Surg* 61:147-153.

Linder HM, Green RB (1964) : Embryologic and surgical anatomy of the extra-hepatic biliary tree. *Surg Clin North Am* 44:1273-1285.

Manzke VH (1966): Symmetrische hyperphalangie des zweiten fingers durch ein akzessorischesmetacarpale. *Fortschr Roentgenstr* 105: 425-427.

Nunez Hoyo M, Lees CD, Herman RE (1982): Bile duct cysts, experience with 15 patients. *Am J Surg* 144:295-299.

Stevenson RE, Taylor HA, Burton OM, Hearn HB III (1980): A digitopalatal syndrome with associated anomalies of the heart, face and skeleton. *J Med Genet* 17:238-242.

Sundaram V, Taysi K, Hartman AFJr, Shackelford GD, Keating Jp JP (1982): Hyperphalangy and clinodactyly of the index finger with Pierre Robin anomaly; Catel-Manzke syndrome: A case report and review of the literature. *Clin Genet* 21:407-410.

Thompson EM, Winter RM, Williams MJH (1986): A male infant with the Catel-Manzke syndrome and dislocatable knees. *J Med Genet* 23:271-273.

Todani T, Watanabe Y, Narusue Tabuchi K, Okajima K (1977) : Congenital bile duct cysts. *Am J Surg* 134:263-269.

Yamagushi M (1980): Congenital choledochal cyst. *Am J Surg* 140:653-657.

Yotuyanagi S (1936): Contribution of the aetiology and pathogeny of the idiopathic cystic dilatation of the common bile duct with report of three cases: A new aetiological theory based on supposed unequal epithelial proliferation at the state of the physiological epithelial occlusion of the primitive choledochus. *Gann* 30: 601-605.