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CASE REPORT

Choledochal cyst in a pediatric patient with Down syndrome, a diagnostic challenge

Quiste de Colédoco en paciente pediátrico con síndrome de Down, un desafío diagnóstico

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ABSTRACT

Introduction. Down syndrome (DS) is the most common chromosomal alteration. Mortality during the first year of life in this group is high. Newborns with this condition have a higher risk of developing cholestasis. Infants with DS can develop cysts. of common bile duct secondary to cholestasis, in pediatric patients common bile duct cysts are a rare pathology.
Clinical Case. 9-month-old infant with a history of DS. On general physical examination, the skin and mucous membranes were generally moist and jaundiced, isochoric and photoreactive pupils, icteric sclerae, with hepatomegaly 3 cm below the costal margin and splenomegaly 2 cm from the rib cage, abdominal perimeter 49 cm. Laboratory and imaging studies, including cholangioresonance, are performed to confirm the diagnosis of Todani IVa choledochal cyst.
Conclusions. In patients with Down syndrome who present a clinical picture of vomiting, abdominal pain and jaundice, the diagnosis of choledochal cyst should be considered. Cholangioresonance is the study that confirms the presence of a choledochal cyst. Our clinical case was evaluated by the surgery service and a surgical procedure was scheduled for resection of the cyst.

Keywords: Down syndrome, choledochal cyst, cholestasis, cholangioresonance.

RESUMEN

Introducción. El síndrome de Down (SD) es la alteración cromosómica más frecuente, la mortalidad durante el primer año de vida en este grupo es elevada, los recién nacidos con esta condición tienen un mayor riesgo de desarrollar colestasis, los lactantes con SD, pueden desarrollar quiste de colédoco secundario a la colestasis, en el paciente pediátrico los quistes de colédoco son una patología poco frecuente.
Caso Clínico. Lactante de 9 meses de edad con antecedentes de SD. Al examen físico general, piel y mucosas húmedas e ictéricas de forma generalizada, pupilas isocóricas y fotoreactivas, escleras ictéricas, con hepatomegalia a 3 cm por debajo del reborde costal y esplenomegalia a 2 cm de la parrilla costal, perímetro abdominal 49 cm. Se realizan estudios de laboratorio y de imagen la colangioresonancia confirmar el diagnóstico de quiste de colédoco Todani IVa.
Conclusiones. En pacientes con síndrome de Down que presenten un cuadro clínico de vómitos, dolor abdominal e ictericia se debe considerar el diagnóstico de quiste colédoco. La colangioresonancia es el estudio que confirma la presencia de quiste de colédoco, nuestro caso clínico fue valorado por el servicio de cirugía y se programó el proceder quirúrgico para resección del quiste.

Palabras Claves: Síndrome de Down, quiste colédoco, colestasis, colangioresonancia.

INTRODUCTION

Down syndrome (DS) is the most common chromosomal disorder, with a high mortality rate in the first year of life of 17.8%, of which 37.5% die in the neonatal period. Newborns with this condition are at greater risk of developing cholestasis, according to a study carried out between January 2005 and September 2011 in Stockholm County, Sweden, in which 206 infants with Down syndrome participated, 3.9% developed choledochal cyst secondary to cholestasis1. This represents a 100-fold increase in the incidence in the general population. In Bolivia, no studies or reports of clinical cases address this issue despite its importance in the pediatric age of this group.

Cholestasis in Down syndrome is due to the likelihood of a smaller size of the circulating bile acid deposit, a lower rate of synthesis, reduced recirculation of bile acids, and an immature function of the canalicular bile acid transport system.2

In pediatric patients, choledochal cysts are a rare pathology with a slight increase in our environment. These malformations of the biliary system include all those cystic dilations in the intra- and extrahepatic biliary tract. There are numerous pathogenic hypotheses.2 The main controversy is between congenital or acquired origin. Its incidence is low; in the western population, it ranges between 1/100,000-150,000 cases with a predominance in females3. Todani's classification distinguishes five types according to the location, morphology, and extent of the lesions; the form of clinical presentation is highly variable according to age and the type of cyst.

A small percentage develop the classic triad of jaundice, abdominal pain, and a mass in the right hypochondrium. In infants, the presence of cholestatic jaundice is familiar due to a decrease in bile flow or excretion. From a biochemical point of view, it is characterized by a level of direct bilirubin in the blood > 1 mg/dl3. In patients over 2 years of age, fusiform and segmental lesions are typical, and the main symptom in these patients is abdominal pain4. In our context, there are few studies related to this topic. Therefore, we present the case of an infant with DS, with the presence of a choledochal cyst, in which the timely diagnosis was made, and serious late complications were avoided.

CLINICAL CASE

A 9-month-old infant with a history of Down syndrome confirmed by a karyotype study, referred from the Villa Tunari Hospital, the mother reports a clinical picture of about 13 days' evolution characterized by liquid stools and vomiting on several occasions of a milky type. She goes to her referral hospital, they decide to admit her, and they start antibiotic treatment with Imipenem and Vancomycin for 4 days. The picture is exacerbated by generalized icteric tinge, choluria, acholia, and elevated bilirubin levels.

The patient has a history of having been admitted at 2 weeks of age for neonatal hyperbilirubinemia, treated with light therapy without complications. On general physical examination, skin and mucous membranes were moist and icteric in a generalized manner; pupils were isochoric and photoreactive, sclera was icteric; the nasal bridge was broad and flat; ears were low-set; the nose was saddle-shaped, the mouth was open and micrognathia, the neck was short, there were no cardiopulmonary adenopathies, the patient was clinically stable, the abdomen was distended with RHA (+) hyperactive, with hepatomegaly 3 cm below the costal margin and splenomegaly 2 cm from the costal arch, abdominal perimeter 49 cm, small extremities, right hand with simian crease, peripheral pulses perceptible, capillary refill time 2 seconds. Rest of the physical examination without alterations. Complementary examinations are carried out (table 1)

Table 1. Supplementary exams

	19/08/2024	21/08/2024	23/08/2024	24/08/2024	25/08/2024	27/08/2024	02/09/2024
Leukocytes (mm3)	27, 400	17, 700	17, 970	-	9,29	8,55	6.770
Segmented (%)	69	69	66	-	65	47	47
Lymphocytes (%)	25	19	28	-	32	49	49
Monocytes (%)	6	5	4	-		1	1
Eosinophils (%)	-	3	1	-	3	3	3
Hemoglobin	7,7	6,9	12. 6	-	11,6	11,7	11.2
Hematocrit	22	20	36	-	35	36	34
Platelets	203, 000	226, 000	210, 000	-	233, 000	340, 000	347,000
RBCs	-	4	1	-	-	-	-
CRP (mg/dl)	-	-	96	-	135	73	Negativo
Blood sugar	72	68		83	72	68	88
Urea	8	10	9	8	12	13	39
Creatinine	0,5	0,5	0,4	0,5	0,6	0,4	0.4
Total bilirubin	4,6	3,6	8,5	9	6,6	2,8	1.6
Direct bilirubin	2,4	2,8	6,7	7	3,4	1,6	0.9
Indirect bilirubin	2,2	0,8	1,8	2	3,2	1,3	0.7
GGT (U/L)	539	408	791	837	22	-	151
GPT (U/L)	36	19,3	20	21	758	25	54
GOT (U/L)	56	23	41	48	60	47	55
LDH (U/L)	448	157	-	-	242	-	
Albumin (g/l)	-	2,3	3,4	-	2,9	-	3.7

Alkaline phosphatase	-	-	-	-	572	423	-
Lipase / Amylase	-	-	-	-		423 / 55	-
Sodium (mmol/dl)	138	135	138	-	141	-	144
Potassium (mmol/dl)	4,8	4,5	4,2	-	4,5	-	4.8
Chlorine (mmol/dl)	105	107,3	105	-	113	-	106

Laboratory tests indicate that the patient is suffering from an infectious process with neutrophilia, a red blood cell count with a decrease in hemoglobin compatible with severe anemia, a renal profile within normal parameters, total and fractionated bilirubin increased with a predominance of direct bilirubin, negative hepatitis A, B, C serology, peripheral blood smear with microcytosis and mild hypothermia, moderate anisocytosis with a predominance of macrocytosis.

After analyzing the laboratory findings, a decision was made to transfuse 120 ml of packed red blood cells, and antibiotic therapy with imipenem and vancomycin was continued. The internal environment is within normal parameters, and stool and urine cultures are negative. The blood culture shows no bacterial growth, and the general urine test is not suggestive of infection.

The patient is observed with a more extraordinary accentuation of jaundice in a generalized way; it was decided to carry out complementary control tests with an increase in total bilirubin, direct bilirubin, and liver profile. In addition, an abdominal ultrasound was indicated (Figure 1A–1B–1C), which described hepatomegaly as being associated with signs of dilatation of the intra- and extrahepatic ducts. Dilated choledochus of 16 mm; mild amount of free fluid.

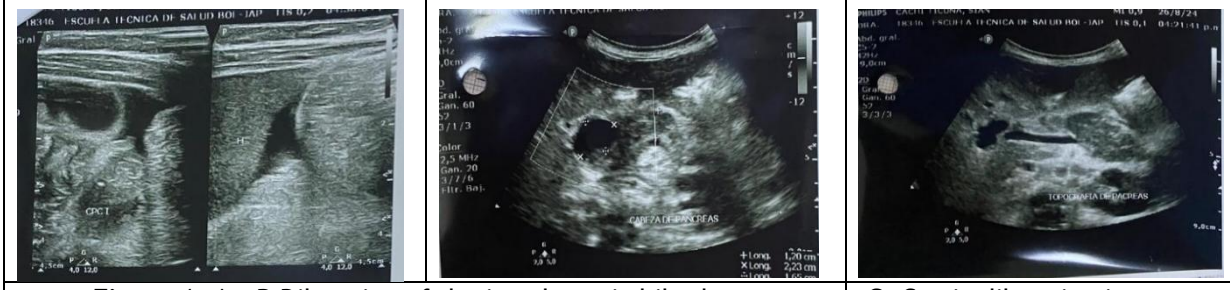

Figure 1. A - B Dilatation of the intrahepatic bile duct. **C.** Cystic dilatation in segment V. Cystic image in the head of the pancreas with hyperechogenic content that may correspond to a complex cyst of the head of the pancreas.

Anteroposterior (AP) abdominal X-ray is also indicated, where distended intestinal loops are evident with the presence of fecal mass in the intestinal tract. (figure 2)

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Figure 2. Chest AP radiograph on admission: Horizontal intercostal arches with distended intestinal loops and presence of fecal mass in the intestinal tract, with no evidence of obstruction.

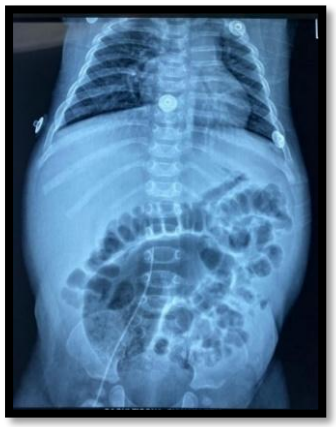
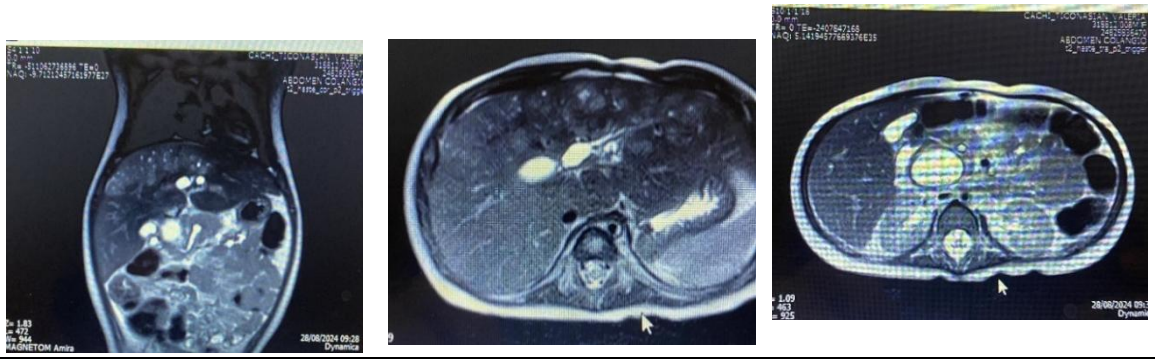


Figure 2. Chest AP radiograph on admission: Horizontal intercostal arches with distended intestinal loops and presence of fecal mass in the intestinal tract, with no evidence of obstruction.

Continue with antibiotic therapy based on Vancomycin and Imipenem on day 6. The results of TORCH. Toxoplasmosis IgG reactive (3.2), Toxoplasmosis IgM non-reactive (0.6), cytomegalovirus IgG reactive (4.2), cytomegalovirus IgM non-reactive (0.3), herpes ½ IgG – IgM non-reactive, hepatitis A (IgM) non-reactive 0.1, Hepatitis B Core (Anti - HBC) reactive 287, Hepatitis B (HBC-AG surface antigen) non-reactive 0.1, Hepatitis C (HCV antigen) non-reactive. A contrast-enhanced magnetic resonance cholangiography (Figure 3A–3B–3C) was also performed, reporting findings consistent with Todani IVa choledochal cyst, complicated with choledocholithiasis, biliary sludge, and evidence of cholangitis. The small lesions referred to are suspected microabscesses. She is scheduled for surgery.



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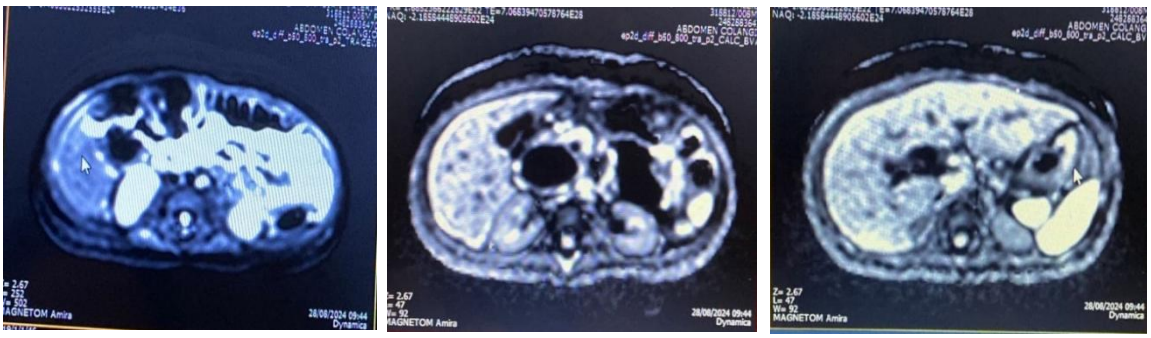

Figure 4. Contrast-enhanced cholangiopancreatography: Todani IVa choledochal cyst, complicated by choledocholithiasis and signs of cholangitis.

CONCLUSIONS

In patients with Down syndrome who present with vomiting, abdominal pain, and jaundice, a diagnosis of choledochal cyst should be considered. Advances in fetal ultrasound have improved the prenatal diagnosis of this condition, although in our clinical case, it could not be performed. Magnetic resonance cholangiography is the study that confirms the presence of a choledochal cyst. Laparoscopic surgery is advantageous over open surgery. For type IVa cysts, resection of the extrahepatic cysts is recommended, and in advanced cases, a liver transplant is necessary. In our clinical case, after diagnosis, it was evaluated by the surgery department, and surgery was scheduled for cyst resection.

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