CASE REPORT

Bardet Biedl Syndrome with Choledochal Cyst: A Very Rare Case Report

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Introduction:

The Bardet-Biedl syndrome (BBS) is a rare genetically heterogeneous, autosomal recessive inherited ciliopathy with wide variability in expression. It presents with varied clinical manifestations like retinitis pigmentosa, polydactyly, central obesity, mental retardation and renal dysfunction. Prevalence of this disease is 1:160 000 in North Europe, 1:13 500 in Arabs, 1:140 000 in North America population[1]. Its diagnosis is done on the basis of genetic analysis[2].

Choledochal cyst is also a relatively rare congenital anomaly of bile duct. Its incidence is approximately 1:100000–1:150000 in Western countries[3] and high geographical variations with a reported incidence of 1:1000 in Asian populations[4]. Diagnosis can be made during episodes with abdominal symptoms or as an incidental finding during abdominal ultrasound. Few reportsare available on antenatal detection[5].Laboratory inflammatory and cholestasis parameters, sonography, CT scan, hepatobiliaryscintigraphy with Technetium 99 (HIDA), MRCP, or ERCP are helpful for the diagnosis of choledochal cyst[6]. Combination of these two conditions are rare. Surprisingly, no such reports are available on reviewing literature of last 10 years.The only two papers to our knowledge are before 1980[7,8]. As it is a very rare condition and the first case of our country, we want to share our experience of management of this particular patient with special reference to perioperative course and prognosis of the patient in the light of literature review.

Case report

A 19-year-old boy was admitted to the Department of Hepatobiliary Pancreatic and Liver Transplant Surgery, Bangabandhu Sheikh Mujib Medical University with the complaints several episodes of abdominal pain, fever and jaundice for last 3 years. Each episode persisted 5 to 7 days and subsided either spontaneously or sometimes after taking a course of antibiotic and pain killer. For evaluating the present complaints an ultrasonogram of whole abdomen and an MRCP (Fig.1) were done. Both reports showed that gallbladder was distended and the common bile duct was fusiformly dilated which contained multiple stones. Right and left intrahepatic ducts were also grossly dilated and there was a stricture at common hepatic duct level. These findings confirmed the diagnosis of choledochal cyst type IV with choledocholithiasis.

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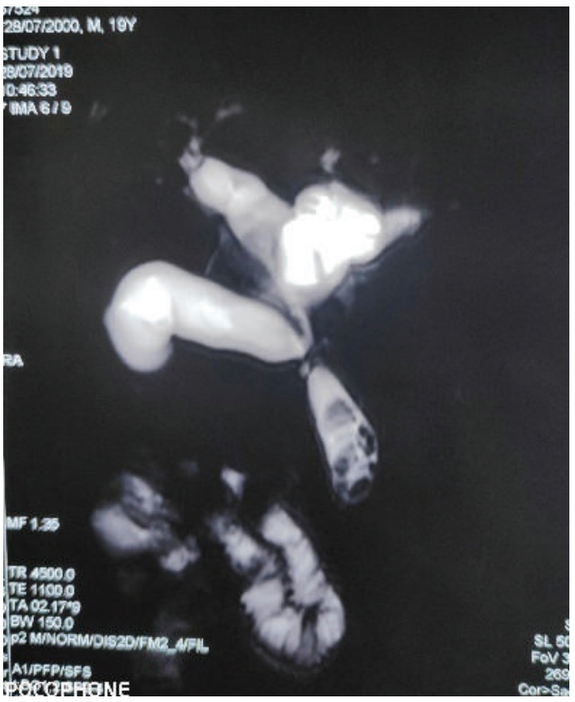
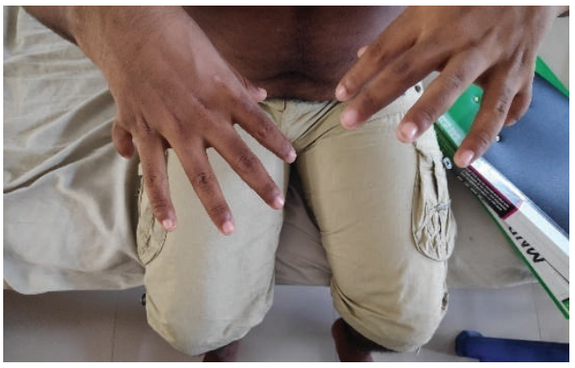


Figure 1: MRCP showing choledochal cyst with choledocholithiasis

On further evaluation, the boy's IQ level was very low and he had slurred speech and for this reason he did not receive any schooling. A thorough clinical examination showed that he had polydactyly in both hands and both foots (Fig. 2) and gynecomastia. He had squint in his both eyes and he complained of shortness of vision. An ophthalmological consultation was taken for eye problems. His visual acuity was counting fingers from one foot.



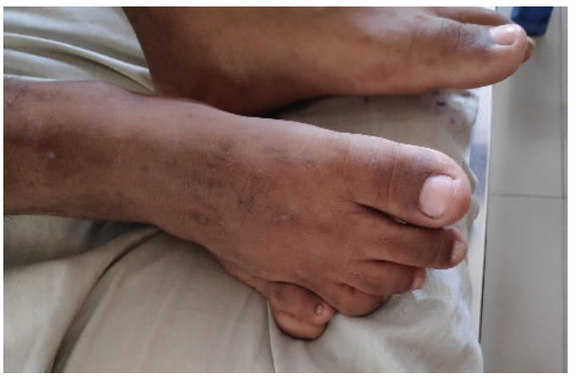


Figure 2: Polydactyly of both foots and both hands

Fundoscopic examination showed a mottled appearance of the retinal pigment epithelium, waxy appearance of the optic nerve and attenuation of blood vessels in the retina, which are the features of retinitis pigmentosa. On the basis of clinical features, the boy was diagnosed as Bardet-Biedl syndrome.[9] He was advised to take vitamin Aroutinely to reduce ophthalmic deterioration. After getting fitness for Anesthesia and Ophthalmology, complete excision of extrahepaticcholedochal cyst was performed. Choledochoscopic examination of both intrahepatic duct systems was performed for excluding the presence of any pathology. There were no stones, stricture or tumor in intrahepatic ducts. The operation was completed by doing Roux-en-Y hepatico-jejunostomy. Postoperatively, patient developed psychosis, otherwise he was conscious and his vital parameters were within normal limit. He did not follow the commands, became restless and removed his intra-abdominal drain tube and nasogastric tube himself. He was restrained and managed by haloperidol. His postoperative course was otherwise normal except minor wound infection which was healed by regular dressing. Patient discharged from hospital on 10th postoperative day.

Discussion:

BBS is an autosomal recessive disorder characterized by retinal dystrophy,

obesity, post-axial polydactyly, renal dysfunction, learning difficulties and hypogonadism. The diagnosis is based on clinical findings and can be confirmed by sequencing of known disease-causing genes in 80% of patients[2]. In the last 2 decades, 21 BBS genes (BBS1–BBS21)have been identified[10-12]. BBS proteins localize to the primary cilium basal body complex, a ubiquitously expressed highly evolutionarily conserved organelle functioning primarily for cell-to-cell signaling. The genes that cause BBS can also cause other ciliopathies, with the classic example being CEP290, which can cause Joubert syndrome, Leber congenital amaurosis, Meckel syndrome, and Senior-Loken syndrome in addition to BBS[13].
Modified diagnostic criteria by Beales, et al[9]. suggest that either four primary features or three primary and two secondary features are required to make a clinical diagnosis. Primary features are-rod-cone dystrophy, polydactyly, genital abnormality, renal anomaly and learning difficulty.Secondary features include speech delay, developmental delay, diabetes mallitus, dental anomaly, congenital heart disease, syndactyly or brachydactyly, ataxia or poor co-ordination, anosmia. The patient had retinitis pigmentosa, polydactyly of all limbs, learning problems as well as speech delay and developmental delay. Genetic testing can also help confirming diagnosis with an increasing number of modifying loci reported[14]. But Gene tests were not done on this patient because of lack gene primer in our lab.

Choledochal cyst is a rare combination with BBS. So far in the published literature BBS is associated with congenital problems in finger, eye, gonads, kidney, heart, tooth, and brain are noted. This present case probably is the first case report of BBS with associated biliary duct abnormality. The surgery for choledochal cyst associated with BBS is our first experience. In addition to usual anesthesiology consultation, ophthalmologicalconsultation was taken. As there was no acute problem related to eye and others vital organs was operation for choledochal cyst was performed safely. But as the patient had poor IQ, he developed postoperative psychosis which was effectively controlled by medication.

Conclusion:

Surgery for choledochal cyst with BBS is performed safely and as it is experienced for first time the case is reported herewith.

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