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Case Report

Association of Poland and Caroli Syndromes: A Case Report

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ABSTRACT:
Poland syndrome and Caroli's syndrome are two Conditions not usually found in association. This unusual case of Poland syndrome highlights once again the wide spectrum of clinical presentation of this rare condition. A 9-year-old boy presented with hepatomegaly. Clinical examination revealed a right anterior chest wall depression, hypoplasia of the right nipple, hypoplasia of the right pectoralis major muscle, pectus excavatum, hepatomegaly and splenomegaly. The diagnosis of Poland syndrome was made on the basis of clinical and radiological features. Liver biopsy showed congenital hepatic fibrosis and magnetic resonance cholangiography revealed cystic dilatation of the common bile duct, dilatation of the right and left hepatic ducts with visibility of intrahepatic bile ducts and portal hypertension which was confirmed by upper gastrointestinal endoscopy. This case emphasizes the need of hepatic involvement screening in patients with Poland syndrome to allow early treatment and prevent complications.

Keywords: Poland Syndrome, Caroli Syndrome, Pectolaris Major Muscle Agenesis, Congenital Hepatic Fibrosis

CASE REPORT:

INTRODUCTION:

Poland Syndrome (PS) is a disorder in which affected individuals are born with missing or underdeveloped muscles on one side of the body, resulting in abnormalities that can affect the chest, breast, shoulder, arm, and hand. The extent and severity of the abnormalities vary among affected individuals [1] In 1841 during an autopsy as a Medical Student, Sir Alfred Poland, reported a case of pectoralis major and minor muscle agenesis associated with other muscle deficiencies and ipsilateral hand brachysyndactyly. Since then, many reports of PS were published [2]. The pathogenic mechanisms underlying PS are still unknown. It could result from a vascular insult during early embryological stages [2]. Caroli syndrome (CS) consists of Caroli's disease and congenital hepatic fibrosis (CHF) [3]. We report an unusual case of Poland syndrome associated with Caroli's syndrome.

Patient and Observation:

A 9-year-old boy was referred to our hospital for hepatomegaly. He was the second child born to consanguineous parents with no family history of a similar condition. His weight was 21 Kg (-2,73 z-score), height 123 cm (-2,18 z-score), he had peculiar facial features with protruding ears, chest asymmetry with right anterior chest wall depression, hypoplasia of the right nipple, hypoplasia of the right pectoralis major muscle and pectus excavatum. Examination of his abdomen revealed hepatomegaly, which was 5 cm palpable below the costal margin and his spleen was palpable 04 cm below the left costal margin. Spine examination revealed kyphosis. The physical examination of the others systems was normal. Biochemical and hematological assessment showed a blood count formula without abnormalities, an absence of cholestasis and moderate cytolysis. (Table 1). The chest CT scan reported chest deformity with right hemithorax retraction and ipsilateral supernumerary rib articulating with the 3rd dorsal vertebra (Figure 1).

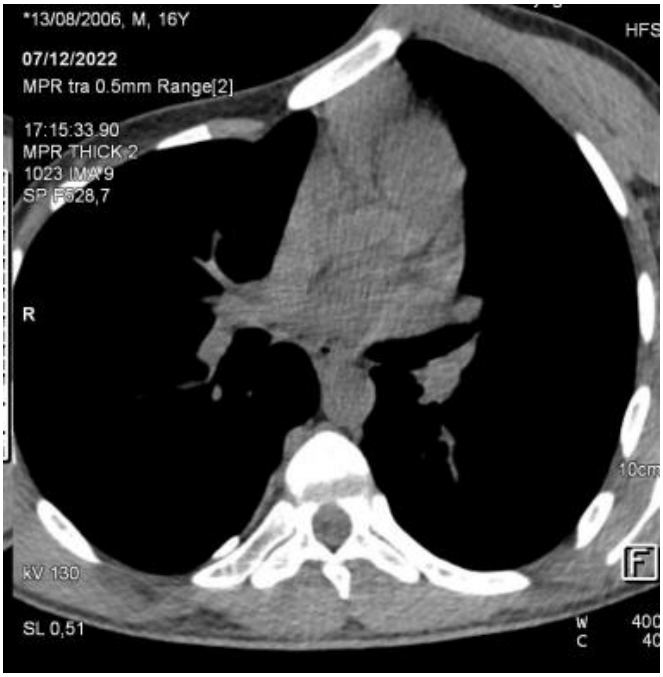

Figure 1 : Chest CT Scan

Table 1 Biochemical and hematological exams		
Parameters	Values in the patient	Normal values
Leukocytes (10e3/µl)	5,9	5.2- 12.4
Platelets (10e3/µl)	170	130- 400
Hemoglobin (g/dl)	12.5	12- 18
Glycemia (g/dl)	0.92	0.70- 1.10
Albumin (g/l)	29.57	32- 50
Prothrombin activity (%)	100	70- 100
AST (u/ml)	50.9	10- 50
ALT (u/ml)	61.4	10- 50
GGT (u/ml)	46	8- 61
Alkaline phosphatase (u/ml)	218	< 375
Total bilirubin (mg/l)	05	< 10
Direct bilirubin (mg/l)	0	< 2
Creatinine (mg/l)	0.65	2- 5
Urea (g/l)	0.34	0.15- 0.25
Calcemia (mg/l)	88	81- 104
Phosphoremia (g/l)	45	40- 70
Alpha fetoprotein (u/ml)	1,06	< 121.6
Alpha 1 globilin (g/l)	2.88	1.00- 4.00
Beta globulin (g/l)	7.64	6.00- 13.00
Gamma globulin (g/l)	6.91	7.00- 15.00
AST: aspartate transaminase, ALT: Alanine transferase, GGT: gamma glutamyl transpeptidase		

Abdominal ultrasonography with Doppler showed dysmorphic liver with heterogeneous echostructure and irregular contours with signs of portal hypertension, splenomegaly, normal kidney appearance, abnormal visibility of some intrahepatic bile ducts, and enlargement of the common bile duct. Magnetic resonance cholangiography revealed hepatic cirrhosis with portal hypertension, cystic dilatation of the common bile duct, dilatation of the right and left hepatic ducts with visibility of intrahepatic bile ducts (figure 2). Our patient had a normal Transthoracic echocardiography, there was no dextrocardia or pulmonary arterial hypertension. An upper gastrointestinal endoscopy showed esophageal varices form II according to Japanese classification which were ligated. Liver biopsy was performed and the histopathologic exam demonstrated features compatible with congenital hepatic fibrosis. Our patient was diagnosed as having a Poland Syndrome (hypoplasia of the right major pectoralis muscle) associated with Caroli's syndrome (congenital hepatic fibrosis and biliary duct dilatation) He was treated with ursodeoxycholic acid 600mg/m2/day, with endoscopic monitoring of esophageal varices already ligated. For his muscular defect, he did not undergo surgery we initially proposed a therapeutic abstention with monitoring of thoracic deformations and will be referred to thoracic surgery after growth completion. Physical therapy was required for kyphosis.

DISCUSSION:

Poland Syndrome is a rare condition, with an estimated incidence of approximately 1 per 30,000 births [2] it is three times more common in males than females [4]. To date, the etiology and pathogenesis of PS are still unknown. Most PS cases are sporadic [1] however De novo deletion of chromosome 11q12.3 in monozygotic twins affected by Poland Syndrome was found [5]. Diagnosis of PS is clinical, the mandatory feature of PS is the agenesis or hypoplasia of the pectoralis major muscle, the sterno-costal head is always affected. In most cases, PS is unilateral. Presumed bilateral PS needs a more extensive differential diagnosis. Additional diagnostic criteria are hypo/aplasia of the homolateral mammary gland and nipples, and malformations of the homolateral upper limb [1]. Ribs agenesis and supernumerary ribs have been described [1,6]. Some authors diagnosed PS only on defective pectoralis muscle, without the ipsilateral limb abnormalities [7] such as in our case. Several abnormalities associated with PS have been described such as: hematological abnormalities (thrombocytopenia, leukemia), renal agenesis, ocular abnormalities (strabismus, epicanthus); genital abnormalities (cryptorchidism, hypospadias); bone abnormalities (scoliosis, clubfoot, mandibular malformation); dextrocardia. These lesions may be associated with other syndromes (Sprengel, Klippel-Feil, Moebius, Adams-Olivier [8]. Caroli's syndrome

(CS) is a rare congenital disorder characterized multifocal segmental non-obstructive saccular or fusiform dilatation of the intrahepatic bile ducts associated with congenital hepatic fibrosis on biopsy.MR imaging is a non-invasive technique which can confirm the diagnosis especially in the large or small cystic patterns. Whereas congenital hepatic fibrosis is a histopathological diagnosis. Histopathological intrahepatic bile duct ectasia and proliferation are associated with severe periportal fibrosis and confirm the congenital hepatic fibrosis component of Caroli's syndrome [9]. The treatment of Caroli's disease depends on the clinical features and the location of the biliary abnormalities [10]. Our patient was treated with ursodesoxycholic acid in order to prevent future episode of hepatolithiasis and cholangitis [12]. Variceal bleeding was prevented with endoscopic band ligation. Liver abnormalities in patients with PS is rarely described in the literature, liver exstrophy has been sporadically reported [11,12], while the association between PS and biliary duct malformation remains extremely rare [6].

Tore et al identified 122 patients with Poland syndrome, in only one case intrahepatic biliary duct dilatation was detected associated with dextrocardia and hypertellorisme [6]. The present patient presented PS with cystic hepatic duct biliary dilatation and congenital hepatic fibrosis diagnosed as having PS associated with Caroli's syndrome, to our knowledge there are no reported cases of hepatic congenital fibrosis related to PS. The patient with PS needs to be assisted by a multidisciplinary team, tailored on the basis of the real needs of the patient and family. In general, the team should involve the following specialists: Pediatric/thoracic surgeon, Plastic surgeon, Orthopedic surgeon, Hand surgeon, Radiologist, Geneticist, Psychologist, Cardiologist, Ophthalmologist, and other professionals as needed [6]. Autologous fat graft should be the first surgical procedure but it is strictly dependent on the grade of deformities, BMI index and chest wall involvement [6]. No surgery is required if there are no functional limitations [6]. Our patient was referred to physical medicine for his kyphosis problem, he will be transferred to thoracic surgery at the end of its growth period.

CONCLUSION:

Biliary duct dilatation has been previously described in very few cases of PS, however, to our knowledge, it is the first published case of the association of PS and CS

Disclosure:

The authors declare no competing interest.

Authors' Contributions:

All authors contributed equally to the write up of this article. They also read and approved the final version of the manuscript.

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