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Congenital abnormalities associated with extrahepatic portal hypertension

M. ODIÈVRE, G. PIGÉ, AND D. ALAGILLE

From Unité de Recherche d'Hépatologie Infantile INSERM U56, and Clinique de Pédiatrie, Université Paris-Sud, Hôpital d'Enfants, Bicêtre, France

SUMMARY Congenital abnormalities were present in 12 out of 30 (40%) children with extrahepatic portal hypertension of unknown cause, but in only 2 out of 17 (12%) children with extrahepatic portal hypertension secondary to umbilical vein catheterization or omphalitis. The most frequent abnormalities in this series and in published reports were atrial septal defect, malformation of the biliary tract, and anomalous inferior vena cava. These findings are consistent with the view that some cases with extrahepatic portal hypertension are congenital in origin.

Extrahepatic portal hypertension may be due to portal vein obstruction arising secondary to omphalitis, umbilical vein catheterization, or intra-abdominal sepsis. In many instances no cause is evident and a developmental defect is inferred, though an unrecognized antepartum or post-partum thrombosis cannot be excluded (Clatworthy, 1974). The observation of several congenital abnormalities associated with extrahepatic portal hypertension prompted us to review our experience with this condition in children.

Material and methods

The records of all patients with extrahepatic portal hypertension admitted from 1965 to 1976 have been reviewed. During this period 30 children were admitted with an extrahepatic portal hypertension of unknown origin (group I). 17 other children with a neonatal history of umbilical vein catheterization or omphalitis were used as a control group (group II). Age at admission in group I ranged from 6 months to 16 years, and in group II from 8 months to 6 years. The extrahepatic portal obstruction was shown by percutaneous splenic porta-venography and/or mesenteric angiography. Only abnormalities obvious on clinical examination or causing symptoms are recorded. Where appropriate, special investigations were undertaken to define these.

Results (Table 1)

Twelve out of the 30 children in group I, and 2 out of the 17 in group II presented with one or several congenital abnormalities. Cardiac and/or vascular abnormalities were the most frequent (5 cases); abnormality of the urinary tract was noted in 3 patients. In 2 other patients the biliary tract was abnormal and the absence of periportal fibrosis was a striking feature in both: one patient (Case 11) had multiple dilated and dysmorphic interlobular bile ducts, and the other (Case 12) had a partial choledochal stenosis. Finally, Turner's syndrome was present in one patient. 6 patients in group I had several abnormalities. There were no affected parents and sibs, and no parental consanguinity in either group.

Discussion

In our series of 47 patients, postnatal portal vein thrombosis was suggested by a history of umbilical catheterization in 16 and of omphalitis with neonatal septicaemia in one. Of these 17 patients, only 2 (12%) had associated congenital abnormalities, while such abnormalities were present in 12 out of 30 (40%) similar patients with extrahepatic portal hypertension but with no evident cause. These results suggest that portal vein obstruction could arise as a result of a developmental defect. It should be stressed that in neither group were systematic investigations or necropsy performed, so that the true incidence of malformation is unlikely to have been recorded.

In large published series of extrahepatic portal hypertension including 228 cases, the paucity of congenital abnormalities has been stressed (Hsia and Gellis, 1955; Clatworthy and Boles, 1959; Fonkalsrud *et al.*, 1974; Voorhees and Price, 1974).

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Table 1 *Congenital abnormalities associated with extrahepatic portal hypertension among children without (group I) and with (group II) a history of umbilical vein thrombosis or omphalitis*

Case no.	*Cardiac and/or vascular abnormality*	*Urinary tract abnormality*	*Biliary tract abnormality*	*Other congenital abnormalities*
Group I (n=30)				
1		Bilateral uretero hydronephrosis		
2	Intestinal telangiectases			Turner's syndrome; bilateral inguinal hernias
3	Atrial septal defect			Deformity of external ear
4		Hydronephrosis with ureterovesical junction stenosis		Deformity of hands and feet
5	Supraorbital angioma			
6	Persistent ductus arteriosus; ventricular septal defect; valvular aortic stenosis; angioma of the skin			
				Cleft palate and lip; absence of 1st costal arch; congenital rectal prolapse
8	Atrial septal defect; bilateral pulmonary stenosis; inferior and superior vena cava obstruction			Bilateral coloboma
9				Ovarian cyst
10		Left kidney agenesis		
11			Intrahepatic biliary 'dysplasia'	
12			Incomplete choledochal stenosis	
Group II (n=17)				
13				Oesophageal atresia
14				Dislocated hip

However, abnormalities that were present included (Table 2) atrial septal defect in 4 patients, extrahepatic biliary malformation in 4, and abnormality of inferior vena cava in 3.

The morphology of the portal vein system as observed by porta-venography or by the surgeon does not differ in patients with a suspected developmental defect from those where an antecedent event may have led to thrombosis. Congenital stenosis or atresia of the portal vein is rare (Raffensperger *et al.*, 1972; Marks, 1973; Clatworthy, 1974). In 2 cases coming to necropsy obstructing valves were shown within the lumen of either the splenic vein or the portal vein or both (Hsia and Gellis, 1955); another patient had a double portal vein with obliteration by an old organized thrombosis (Hsia and Gellis, 1955). This last case might be an example of congenital abnormality with secondary throm-

Table 2 *Congenital abnormalities associated with extrahepatic portal hypertension in the literature*

Authors	*No. of patients with extrahepatic portal hypertension*	*No. of patients with congenital abnormality*	*Abnormalities*
Lamy *et al.* (1961)	2	1	Atrial septal defect, absence of gallbladder
Esposito (1966)	1	1	Agenesis of left kidney
Seringe *et al.* (1966)	1	1	Absence of inferior vena cava
Moretti *et al.* (1968)	3	3	Pigmented naevi, cystic lymphangioma, arterial cerebral aneurysm; Dislocated hip, Stein-Leventhal syndrome; Crouzon's disease, arachnodactyly
Rösch and Dotter (1971)	38	1	Congenital anomaly of the bile ducts
Raffensperger *et al.* (1972)	14	3	Imperforate anus, atrial septal defect, mitral valve insufficiency Anomalous inferior vena cava Choledochal duct cyst, malrotation, anomalous drainage of inferior vena cava
Pinkerton *et al.* (1972)	23	1	Choledochal cyst
Keighley *et al.* (1973)	10	2	Atrial septal defect Atrial septal defect, anomalous pulmonary venous drainage
Myers and Robinson (1973)	54	7	Omphalocele, pyloric stenosis, hypoplastic mandible, laryngeal cleft, bilateral pulmonary stenosis

bosis. Subsequent recanalization and development of perivascular vessels can produce the most common cavernomatous disposition (Clatworthy, 1974). The late onset of clinical manifestations of portal hypertension several years after birth does not preclude that the abnormality is congenital.

An awareness of the possibility of associating congenital abnormalities in extrahepatic portal hypertension has practical implications. Multiple abnormalities in another system might contraindicate any major surgical procedure. In practical terms the splenic, superior mesenteric, and left renal veins, as well as the inferior vena cava should be investigated so that appropriate surgery may be planned.

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Correspondence to Dr. M. Odièvre, Unité de Recherche d'Hépatologie Infantile, Hôpital de Bicêtre, 78, Rue du Général-Leclerc, 94270 Bicêtre, France.