# Multiple Abdominal Vascular Anomalies in a Patient with Alagille Syndrome

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**Alagille syndrome affects multiple organ systems. The most common vascular manifestation of Alagille syndrome is peripheral pulmonary artery stenosis. A few cases of abdominal vasculature involvement have been reported, particularly in the pediatric age group. Herein, the authors describe an adult patient with Alagille syndrome who presented with multiple visceral vascular abnormalities, including a high-grade stenosis of the celiac artery, superior mesenteric artery (SMA), aneurysms of the distal common hepatic artery, and distal SMA detected with computed tomographic angiography.**

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**Abbreviations**: MIP = maximum intensity projection, SMA = superior mesenteric artery

ALAGILLE syndrome is an autosomal-dominant disorder characterized by abnormalities of multiple organs, including liver, heart, eyes, vertebrae, face, and kidneys (1,2). Cardiac manifestations mainly include murmur, tetralogy of Fallot, truncus arteriosus, ventricular septal defect complex, and anomalous venous return. Skeletal manifestations are butterfly vertebrae, shortened interpedicular distance, shortened phalanges, short stature, pathologic fractures, and spina bifida occulta. Posterior embryotoxon, shallow anterior chamber, cataracts, strabismus, ectopic pupil, and glaucoma are the main ocular manifestations. Patients have particular facies, thinned cortical bones, large ears, macrocephaly, and sinus abnormalities. Renal manifestations consist of neonatal renal insufficiency, solitary kidney, ectopic kidney, horseshoe kidney, cystic and multicystic kidney, and infantile renal tubular acidosis. Vascular anomalies are one of the characteristic findings of this syndrome. The most common vascular manifestation is pulmonary artery involvement characterized by peripheral pulmonary artery stenosis (3). Herein, we describe computed tomographic (CT) angiography findings of an adult with Alagille syndrome who presented with multiple abdominal vascular involvement. Our findings are described in of the literature and with an overview of the syndrome.

## CASE REPORT

Our institution did not require institutional review board approval for this retrospective case report.

A 43-year-old white man with Alagille syndrome had been followed up by one of the gastroenterologists (D.M.J.) in our outpatient clinic. He had been diagnosed with Alagille syndrome almost 30 years ago when he presented with failure to thrive and chronic cholestasis. Liver biopsy showed paucity of bile ducts. He also had short stature, cerebral basilar artery aneurysms, coarctation of the aorta with repair, cardiac arrhythmia, fibrous dysplasia, and mandibular deformity due to osteomas and bone cysts. He had a period of 10 years of alcohol abuse and discontinued drinking 10 years before presentation. He had undergone abdominal ultrasonography (US) during his follow-up and a liver lesion was detected. The patient underwent abdominal CT to rule out hepatocellular carcinoma, and findings were consistent with cirrhosis accompanied by portal hypertension. The patient was further evaluated with CT angiography due to the presence of various abdominal vascular abnormalities at CT. At CT angiography, the aorta had a small caliber, the mid-abdominal aorta measured 1 cm, and there was high aortic bifurcation just proximal to L3. There were focal severe stenoses of the origins of the celiac and superior mesenteric artery (SMA) (**Fig 1**) and bilateral stenoses at renal arteries at the aortic origin with poststenotic dilatation (**Fig 2**). There was an aneurysm of the distal common hepatic artery just proximal to the proper hepatic and gastroduodenal arteries as well as an aneurysm of an SMA branch vessel in the medial left hemiabdomen (**Figs 3**, **4**). The proper hepatic artery was poorly filled but patent. There were multiple aneurysms in the inferior pancreaticoduodenal arcade (**Fig 3**). There was hypertrophy of the inferior mesenteric artery with moderate retrograde flow into

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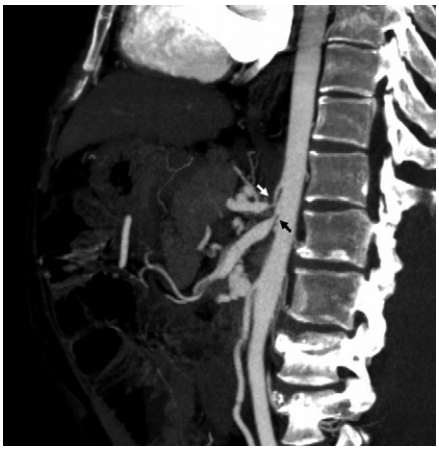
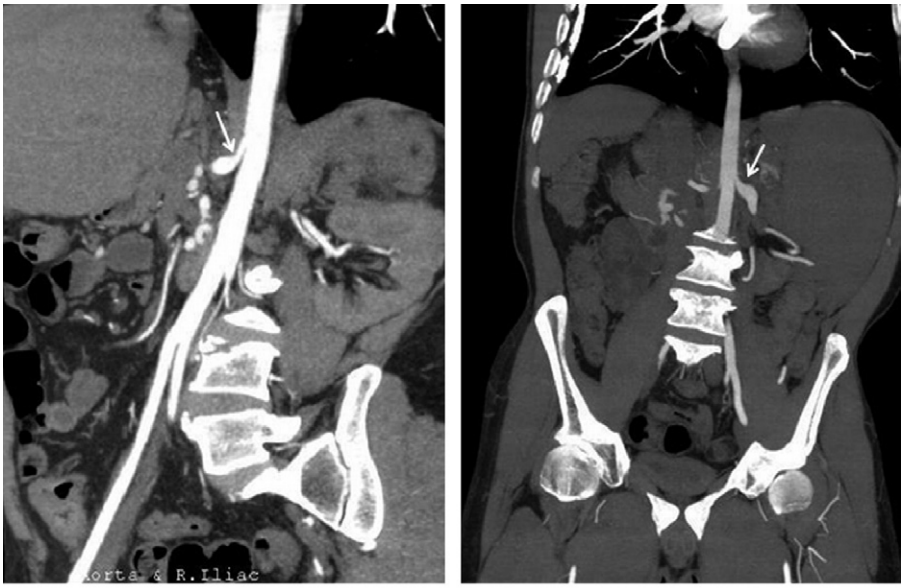


**Figure 1.** Sagittal three-dimensional CT arterial slab maximum intensity projection (MIP) image demonstrates a high-grade stenosis in the celiac trunk (white arrow) and SMA (black arrow). Poststenotic dilatation is present.




**a.** **b.**

**Figure 2.** **(a)** Coronal oblique three-dimensional CT arterial slab MIP image demonstrates a stenotic right renal artery with poststenotic dilatation (arrow). **(b)** Coronal three-dimensional CT arterial slab MIP image shows the stenotic left renal artery with poststenotic dilatation (arrow).

the meandering artery, which provided collateral blood supply to the epigastric region (**Fig 4**).

The patient has been treated medically so far. He now has cirrhosis–-probably due to a combination of both the Alagille syndrome and his history of alcohol abuse. He is currently medically stable with no jaundice. He may need a transplant in the future.

## DISCUSSION

Since the first description of Alagille syndrome by Alagille and Watson (1,4), more than 600 cases have been diagnosed, and a large number of abnormalities have been associated with Alagille syndrome (**Table**) (3). Alagille syndrome (also known as arteriohepatic dysplasia) is mainly characterized by cholestasis from bile duct paucity, butterfly vertebrae, peripheral pulmonary artery stenosis, ocular anomalies, and peculiar facies. Clinical manifestations related to the hepatobiliary system include intrahepatic bile duct paucity, mild to severe cholestasis, neonatal hepatitis, fibrosis, cirrhosis, portal hypertension, liver failure, hepatocellular carcinoma, and nodular hamartoma (5). Jaundice is present as a conjugated hyperbilirubinemia in the neonatal period. The other manifestations are hepatosplenomegaly and increased levels of serum cholesterol, bile salts, aminotransferases, and other enzymes (6).

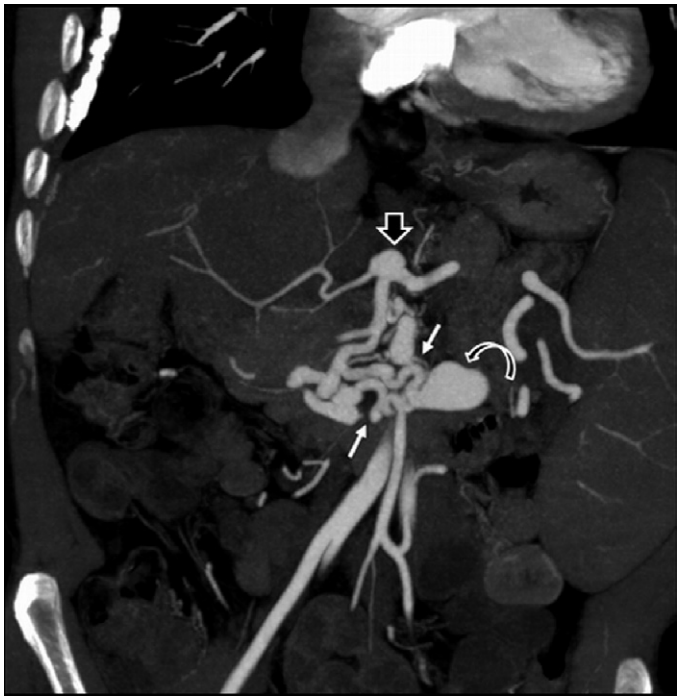


**Figure 3.** Coronal three-dimensional CT arterial thick slab MIP image demonstrates multiple aneurysms in the inferior pancreaticoduodenal arcade (straight solid arrows). The gastroduodenal artery and pancreaticoduodenal arcade are enlarged due to stenosis of the celiac trunk and SMA. Aneurysms of the distal common hepatic artery (straight open arrow) and SMA branch vessel (curved arrow) are seen. Splenomegaly is present.

Vascular anomalies have also been defined in Alagille syndrome (2,4). The most common manifestation is pulmonary artery involvement manifesting as peripheral pulmonary artery stenosis. Alagille syndrome has