

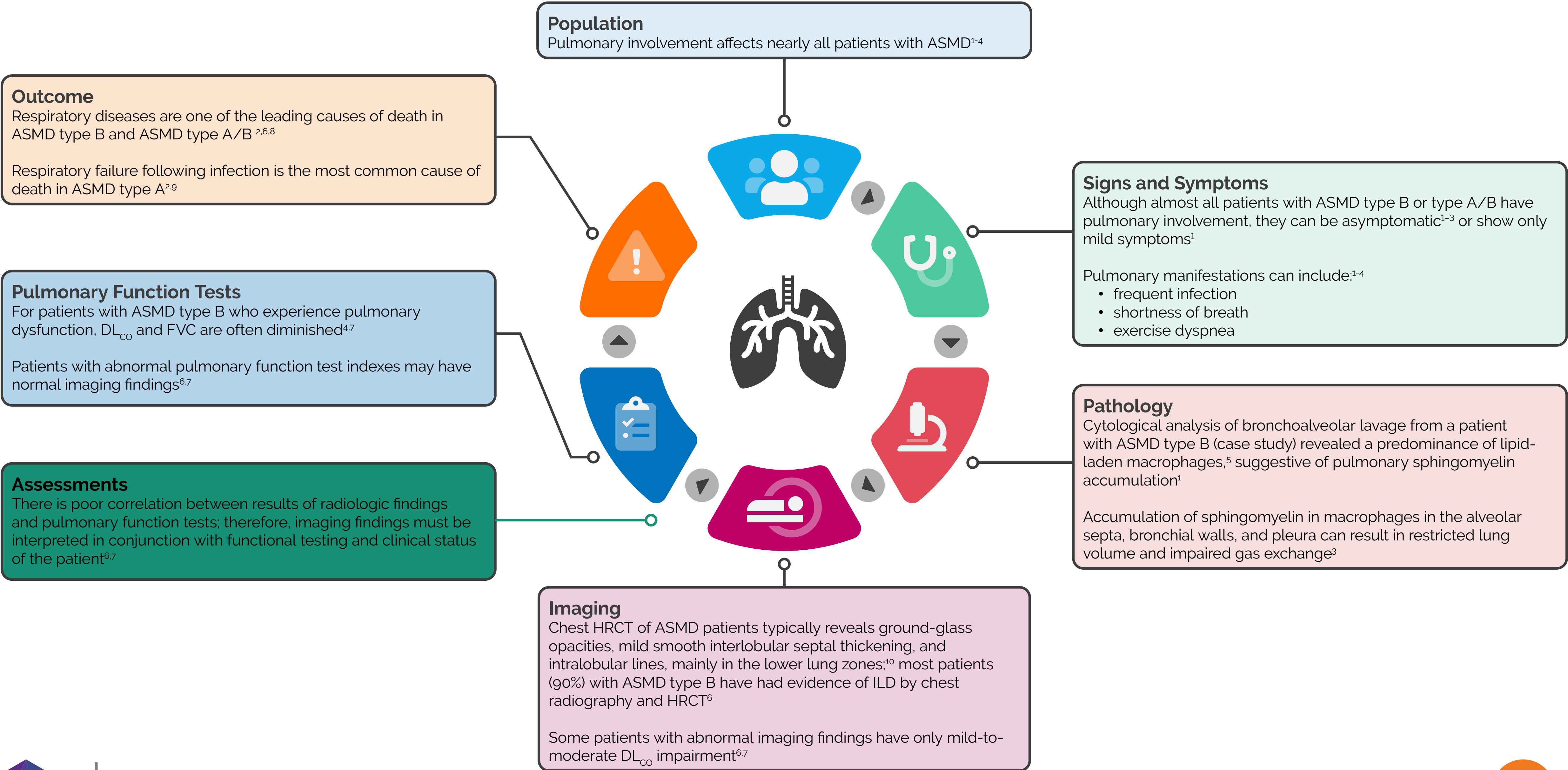
# **FOCUS ON LUNG AND LIVER:** Summary of the Lung and Liver Manifestations of ASMD

Please select an organ

**Lung**

**Liver**

# Lung Manifestations of ASMD



# Liver Manifestations of ASMD

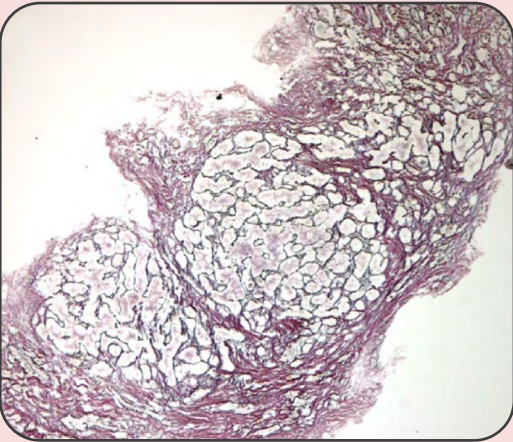
**Population**  
Hepatomegaly occurs in nearly all patients with ASMD and is usually one of the first presenting clinical features of ASMD together with splenomegaly<sup>1</sup>

**Outcome**  
Liver disease is the second most common cause of death in ASMD type B, and third leading cause of death in ASMD type A/B<sup>7</sup>

**Signs and Symptoms**  
Symptoms of hepatomegaly may include enlarged abdomen<sup>2</sup> and abdominal pain<sup>3</sup>  
  
Fibrosis can range from mild to severe with progressive fibrosis leading to hepatic dysfunction and/or liver failure/cirrhosis<sup>1,4,5</sup>  
  
Adults with ASMD have a high risk of developing liver cirrhosis, portal hypertension, and variceal bleeding;<sup>5,6</sup> cirrhosis is a risk factor for hepatic carcinoma<sup>7</sup>

**Imaging**  
Hepatomegaly can be detected and monitored by clinical examination and imaging modalities such as ultrasound, X-ray, CT, or MRI scans<sup>1,6</sup>  
  
Accurate monitoring of fibrosis may be achieved using ultrasound-based transient elastography<sup>6</sup>

**Pathology**  
In ASMD, sphingomyelin accumulation is observed in the hepatocytes,<sup>2,8</sup> Kupffer cells, and bile duct epithelium of the liver;<sup>8</sup> the infiltration of sphingomyelin-laden hepatic macrophages into sinusoidal spaces can lead to liver dysfunction<sup>5,7</sup>



Liver biopsy with fibrosis grade of 4 (cirrhosis)<sup>5</sup>

**Liver Function Tests**  
Periodic liver panels (every 6–12 months), e.g. ALT, AST, GGT, coagulation, albumin, are useful in assessing liver function; however, the extent of liver disease and fibrosis is commonly underestimated<sup>6</sup>  
  
Levels of ALT and AST are elevated in 65–75% of patients with ASMD;<sup>1,2,6,9</sup> liver enzyme levels can be normal/only slightly elevated at early stages, so should be monitored alongside liver imaging<sup>6</sup>



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