



Types of Congenital Liver Diseases

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Abstract

Liver disease in early infancy surrounds a wide spectrum of conditions, including infectious, metabolic, and hematologic disorders, congenital vascular and heart diseases, drug-related toxicity, hypoxia, and gestational alloimmune liver disease related with neonatal hemochromatosis.

Keywords:

Chronic, Liver.

Introduction

Healthcare providers don't know the exact source of congenital liver defects. Most likely they are caused by something that transpired as the unborn baby was advancing or around the time of birth. This might occur because of one or more of the following: A viral or bacterial infection after birth, An immune system problem, such as when the immune system assaults the liver or bile ducts for unknown reasons, A genetic mutation. This is a long-lasting transpose in a gene's structure. A problem during liver and bile duct growth in the unborn baby, Contact with toxic substances. Inherited liver diseases are a classification of metabolic and genetic defects that typically cause early chronic liver involvement. Most are due to a deficient of an enzyme/transport protein that alters a metabolic pathway and employs a pathogenic role mainly in the liver.

An early chronic liver involvement may be noticed in a number of genetic and metabolic diseases although with different penetrance, age at onset, and result. Clinical symptoms and laboratory data are frequently converging, thus rendering a differential diagnosis difficult. A great advancement both in imaging [1] and in molecular genetics.

Alpha-1 antitrypsin (AAT) deficiency

Alpha-1 antitrypsin (AAT) deficiency (OMIM 613490) is an autosomal dominant (codominant) disease due to mutations in the SERPINA1 gene that conceals the serine protease inhibitor AAT. The protein, mainly synthesized by liver cells, hinders proinflammatory proteases such as neutrophil elastase, thus, safeguarding the lung from proteolytic damage. AAT deficiency has an incidence of 1:2,000–5,000 but the number of diagnosed patients is underrated.

Cystic fibrosis

Cystic fibrosis (CF, OMIM 219700) is the most persistent lethal autosomal recessive disease among Caucasians. CF is a systemic disease that emerges mainly with pancreatic insufficiency in more than 90% of cases and pulmonary disease due to inflammation and opportunistic colonization that moderately causes respiratory insufficiency [14]. About 20% of patients experience meconium ileus.

Wilson disease

Wilson disease (WD, 277900) is an autosomal recessive disorder with an occurrence of about 1:30,000. It typically materializes with liver disease in the second decade and neurological disorders in the third decade, even if cases with preliminary or later onset have been described [30]. Wilson disease depends on mutations in the gene encrypting the ATP7B Cu translocase, a protein mainly expressed by the hepatocyte that synchronizes the levels of copper in the liver.

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