Inherited metabolic liver diseases in infants and children: an overview

Prirojene metabolne bolezni jeter pri dojenčkih in otrocih: pregled

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Abstract

Inborn errors of metabolism, which affect the liver are a large, continuously increasing group of diseases. Their clinical onset can occur at any age, from intrauterine period presenting as liver failure already at birth to late adulthood. Inherited metabolic disorders must be considered in differential diagnosis of every unexplained liver disease. Specific diagnostic work-up for either their confirmation or exclusion should start immediately since any postponing can result in delayed diagnosis and death or irreversible disability. This can be particularly painful while many inherited metabolic liver diseases are relatively easily treatable if diagnosed on time, for instance galactosemia or hereditary fructose intolerance by simple dietary means. Any unexplained liver disease, even one looking initially benign, should be considered as a potential liver failure and therefore should deserve proper attention. Diagnosis in neonates is additionally complicated because of the factors which can mask liver disease, such as physiological neonatal jaundice, normally relatively enlarged liver and increased transaminases at that age. In everyday practice, in order to reveal the etiology, it is useful to classify and distinguish some clinical patterns which, together with a few routine, widely available laboratory tests (aminotransferases, prothrombine time, albumin, gammaGT, total and conjugated bilirubin, ammonia, alkaline phosphatase and glucose) make the search for the cause much easier. These patterns are isolated hyperbilirubinemia, syndrome of cholestasis in early infancy, hepatocellular jaundice, Reye syndrome, portal cirrhosis and isolated hepatomegaly. Despite the fact that some diseases can present with more than one pattern (for instance, alpha-1-antitrypsin deficiency as infantile cholestasis, but also as hepatocellular jaundice), and that in some diseases one pattern can evolve into another (for instance, Wilson disease from hepatocellular damage into portal cirrhosis), this classisfication is still very useful. Diagnostic work-up in an unexplained liver disease can be very complex and tricky, particularly in patients with (sub)acute liver failure, when the time for interventions is limited, many pathological laboratory results can be a secondary abnormality, and many specific tests should be done and interpreted properly in parallel with various treatment measures. In such clinical setting, specific knowledge, skills, experience, special drugs and properly equiped diagnostic laboratories are, as a rule, necessary for good clinical outcome. Therefore, it is highly recommended to transfer the patient as soon as possible to a specialized center where all these requirements can be fulfilled by an experienced specialized team comprising pediatric gastroenterologist, subspecialist in metabolic diseases, intensivist, surgeon, biochemist and others. Such an approach is the only way to decrease the still high proportion of patients with inherited metabolic liver diseases who remain causally undiagnosed and whose outcome is far from optimal.