

ABSTRACT

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Title: Genetic diseases of the liver

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Liver function may be affected by various factors including genetic diseases. The aim of this bachelor thesis is to collect and summarize current information about genetic diseases of the liver.

Diseases with the highest incidence in the population are Dubin-Johnson syndrome, Rotor syndrome, Crigler-Najjar syndrome and Gilbert's syndrome. They are known as inherited hyperbilirubinemia characterized by an impairment in bilirubin metabolism. These genetic diseases are very rare with exception of Gilbert's syndrome. However, despite low prevalence and incidence it is necessary not to prolong their diagnosis. Most of them do not have any complications and do not require any treatment. The exception is Crigler-Najjar's syndrome, as untreated can have fatal consequences.

Wilson's disease and hereditary hemochromatosis are inherited disorders of metal metabolism. Wilson's disease is a rare disease caused by an impairment in copper metabolism. Inherited hemochromatosis causes excessive iron deposition in the liver and other organs. Alpha-1 antitrypsin deficiency is characterized by impaired alpha-1 antitrypsin protein production.

On the basis of the obtained information about genetic diseases of the liver was made comparison of their clinical picture, diagnosis, therapy, prevalence and incidence.

Key words: liver, genetic diseases, impairment of metabolism, hyperbilirubinemia