Microvesicular steatosis, hemosiderosis and rapid development of liver cirrhosis in a patient with Pearson's syndrome

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BACKGROUND/AIMS:

Pearson's marrow-pancreas syndrome consists of refractory sideroblastic anemia with vacuolization of marrow precursors and exocrine pancreas dysfunction. Patients with this disease usually have large deletions of the mitochondrial genome. We report a patient with Pearson's syndrome who had predominantly hepatic manifestations such as microvesicular steatosis, hemosiderosis and rapidly developing cirrhosis.

METHODS:

Analysis of the mitochondrial and nuclear genomes, determination of enzyme activities and of the hepatic iron content were performed using standard techniques of molecular biology and biochemistry.

RESULTS:

The patient had typical ringed sideroblasts in a bone marrow smear and a 7436-bp deletion of the mitochondrial genome in all tissues investigated, compatible with Pearson's syndrome. He died within 3 months after birth due to liver failure. Histopathological analysis of the liver revealed complete cirrhosis with signs of chronic cholestasis, microvesicular steatosis and massive hemosiderosis. In addition, the patient was heterozygous for the C282Y and H63D mutations of the hemochromatosis gene.

CONCLUSIONS:

Pearson's syndrome should be added to the list of neonatal diseases which can cause microvesicular steatosis, hepatic accumulation of iron and liver cirrhosis.