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Autor(en) des Beitrags:
Freisinger, P; Fütterer, N; Lankes, E; Gempel, K; Berger, TM; Spalinger, J; Hoerbe, A; Schwantes, C; Lindner, M; Santer, R; Burdelski, M; Schaefer, H; Setzer, B; Walker, UA; Horváth, R

Titel des Beitrags: Hepatocerebral mitochondrial DNA depletion syndrome caused by deoxyguanosine kinase (DGUOK) mutations.

Abstract:
BACKGROUND: Autosomal recessive mutations in deoxyguanosine kinase (DGUOK) have been identified in the hepatocerebral form of mitochondrial DNA (mtDNA) depletion syndrome. OBJECTIVES: To describe the clinical spectrum of DGUOK-related mtDNA depletion syndrome in 6 children and to summarize the literature. RESULTS: We identified pathogenic mutations in DGUOK in 6 children with the hepatocerebral form of mtDNA depletion syndrome. We describe the clinical, neuroradiologic, histologic, and genetic features in these children. All children showed severe hepatopathy, while involvement of other organs (skeletal muscle and brain) was variable. We identified 5 novel mutations (1 of them in 2 children) and 2 previously described mutations. Three different mutations affected the initial methionine, suggesting a mutational hot spot. One of our patients underwent liver transplantation; pathologic findings revealed (in addition to diffuse hepatopathy) a hepatocellular carcinoma, implying a possible link between mtDNA depletion syndrome and tumorigenesis. CONCLUSION: We studied 12 children with infantile hepatoencephalopathies and mtDNA depletion syndrome and found pathogenic DGUOK mutations in 6, suggesting that this gene defect is a frequent but not an exclusive cause of...
the hepatic form of mtDNA depletion syndrome.

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