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Titel des Beitrags:
Biallelic Mutations in NBAS Cause Recurrent Acute Liver Failure with Onset in Infancy.

Abstract:
Acute liver failure (ALF) in infancy and childhood is a life-threatening emergency. Few conditions are known to cause recurrent acute liver failure (RALF), and in about 50% of cases, the underlying molecular cause remains unresolved. Exome sequencing in five unrelated individuals with fever-dependent RALF revealed biallelic mutations in NBAS. Subsequent Sanger sequencing of NBAS in 15 additional unrelated individuals with RALF or ALF identified compound heterozygous mutations in an additional six individuals from five families. Immunoblot analysis of mutant fibroblasts showed reduced protein levels of NBAS and its proposed interaction partner p31, both involved in retrograde transport between endoplasmic reticulum and Golgi. We recommend NBAS analysis in individuals with acute infantile liver failure, especially if triggered by fever.

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