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Mitochondrial hepatopathy in infants younger than two years

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LETTER TO THE EDITOR

In a recent article, McKiernan et al. reported about 39 patients <2 years with acute liver failure (ALF) of whom 5 had mitochondrial depletion syndrome (MDS) due to mutations in *DGUOK* (n=2), *POLG* (n=2), or *MPV17* (n=1) [1]. We have the following comments and concerns.

Mitochondrial ALF may not only occur in patients with MDS but also in patients with mtDNA deletions [2] or in patients with mutations in non-MDS nDNA genes, such as TRMU [3] or HADHB [4]. Were the 12 patients with undetermined cause, particularly patient 24 with mitochondrial liver disease without a causative mutation and those with undetermined mtDNA depletion, investigated for mtDNA deletions or non-MDS nDNA gene mutations?

We should be informed about the drugs the 39 patients were regularly taking. Acute liver failure may be triggered by liver-toxic medication [5]. Particularly antiepileptic drugs, such as valproic-acid, carbamazepine, phenytoin, and barbiturates may be mitochondrion-toxic and may trigger ALF [6]. How many of the 39 had epilepsy requiring antiepileptic medication? In how many of those with MDS who died were drugs, including immunosuppressives for LT, responsible for the fatal outcome?

Patients 3 and 4 died from ALF [1]. Did these two patients undergo autopsy? Why was no histological, biochemical and genetic work-up of liver tissue carried out in these two patients? Were cerebral lesions on MRI in the 5 MDS patients compatible with stroke-like episodes? Stroke-like episodes not only occur in MELAS but rarely also in other types of mitochondrial disease. Stroke-like episodes freuqnetly go along with epilepsy requiring antiepileptic treatment.

Overall, this interesting retrospective study could profit from investigations for mutations in alternative genes, from providing more extensive data about the history, provision of the currnet medication, and from comprehensive work-up of liver tissue at autopsy or biopsy.

REFERENCES

[1] McKiernan P, Ball S, Santra S, et al. Incidence of Primary Mitochondrial Disease in Children Presenting With Acute Liver Failure Under 2 Years of Age. J Pediatr Gastroenterol Nutr 2016 Jul 30.

[2] Lee WS, Sokol RJ. Mitochondrial hepatopathies: advances in genetics, therapeutic approaches, and outcomes. J Pediatr 2013; 163:942-8.

[3] Gaignard P, Gonzales E, Ackermann O, Labrune P, Correia I, Therond P, Jacquemin E, Slama A. Mitochondrial Infantile Liver Disease due to TRMU Gene Mutations: Three New Cases. JIMD Rep 2013; 11:117-23.

[4] Kobayashi T, Minami S, Mitani A, Tanizaki Y, Booka M, Okutani T, Yamaguchi S, Ino K. Acute fatty liver of pregnancy associated with fetal mitochondrial trifunctional protein deficiency. J Obstet Gynaecol Res 2015; 4:799-802.

[5] Hynynen J, Komulainen T, Tukiainen E, Nordin A, Arola J, Kälviäinen R, Jutila L, Röyttä M, Hinttala R, Majamaa K, Mäkisalo H, Uusimaa J. Acute liver failure after valproate exposure in patients with POLG1 mutations and the prognosis after liver transplantation. Liver Transpl 2014;20: 1402-12.

[6] Björnsson E. Hepatotoxicity associated with antiepileptic drugs. Acta Neurol Scand 2008; 118:281-90.

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