

Carnitine-acylcarnitine translocase deficiency. *Clinical course of 3 Saudi children with a severe phenotype*

To the Editor

I have 2 comments on the interesting case report by Al-Sannaa and Cheriyan.¹

First, the authors did well in confirming the diagnosis of carnitine-acylcarnitine translocase (CACT) deficiency in the studied patient by measuring the level of CACT activity in the cultured fibroblasts, as they did not totally rely upon plasma acylcarnitine profile. It is worthy to mention that acylcarnitine profiling in plasma is the assay of choice for the diagnosis at a metabolite level. However, since the acylcarnitine profile observed in CACT-deficient patients is identical to that in carnitine palmitoyltransferase II-deficient patients, definitive identification of CACT-deficiency in a certain patient requires determination of the activity of CACT.²

Second, the inclusion of tandem mass spectrometry (MSMS) into an existing newborn screening program is promising. Implementation of that program has significantly reduced the morbidity and mortality of long-chain fatty acid oxidation defects, including CACT deficiency. It also identifies a great number of mildly affected patients who might never develop clinical symptoms throughout life. Disease prevalence has increased with newborn screening.³ The MSMS has shown a sensitivity of 95.9% and specificity of 99.8%, with a positive predictive value of 18%.⁴ I wonder whether screening for CACT deficiency is incorporated into the neonatal screening program in Saudi Arabia.

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Reply from the Author

I fully support the comment of Dr. Al-Mendalawi regarding acylcarnitines profile of CACT deficiency. In regards to newborn screening for inborn error of metabolism, unfortunately, this has not been established yet at our center. This has been already discussed in a published study by our group.⁵

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Ethical Consent

All manuscripts reporting the results of experimental investigations involving human subjects should include a statement confirming that informed consent was obtained from each subject or subject's guardian, after receiving approval of the experimental protocol by a local human ethics committee, or institutional review board. When reporting experiments on animals, authors should indicate whether the institutional and national guide for the care and use of laboratory animals was followed.