Der Carnitin-Transporter-Defekt: Eine Screening-Erkrankung?

Carnitine transporter defect: a suitable condition for screening?

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Carnitine transporter defect (CTD) is a potentially life-threatening disorder causing acute metabolic decompensation, cardiac arrhythmia, and cardiac and skeletal myopathies. CTD is included in many newborn screening (NBS) programs. The screening parameter free carnitine, however, is influenced by maternal conditions due to placental transfer. CTD is part of a NBS pilot study in Bavaria, Germany. This study reviewed the NBS results and the long-term follow-up of the identified patients treated in our center between January 1999 and June 2018. Among 1,816,000 Bavarian NBS samples, six newborns were diagnosed with CTD (incidence of 1:302,667; positive predictive value (PPV) of 1.63% from 2008 to 2018). In the 24 newborns presented to our center for confirmatory testing, we detected four newborns and six mothers with CTD, one newborn and three mothers in whom CTD was presumed but not genetically confirmed, and one mother with glutaric aciduria type I. In 11 newborns, no indication for an inborn error of metabolism was found. The newborns and mothers with CTD had no serious cardiac adverse events or relevant muscular symptoms at diagnosis and during treatment for up to 14 years. Three mothers were lost to follow-up. Revealing a lower incidence than expected, our data confirm that NBS for CTD most likely misses newborns with CTD. It rather produces high numbers of false-positives and a low PPV picking up asymptomatic mothers with a diagnosis of uncertain clinical significance. Our data add to the growing evidence that argues against an implementation of CTD in NBS programs.