

Supplemental Data

Brain magnetic resonance imaging (MRI) and (1H-MRS) imaging was performed with 1.5 Tesla MR (Philips Ingenia 1.5T, Philips Medical System) at the level of the basal ganglia monovoxically spectroscopically. In the evaluations, proton spectra were obtained using a head coil. PRESS (Point-resolved surface coil spectroscopy) sequence (TR = 1500 TE = 2000) with a high signal-to-noise ratio (SNR) was preferred for the acquisition of spectra from voxels. The examination period was determined as 10-15 minutes. "Gradient shimming", suppression of the water signal and data processing were done automatically with a package program. The images obtained were postprocessed (Intellispace Service Philips, Netherlands). N-Acetyl aspartate and other N-acetyl containing compounds (NAA): 2.02 ppm, creatinine: 3.03 ppm, choline and other trimethyl-amine containing compounds: 3.20 ppm, lipids: 0.90 and 1.3 ppm, glutamate and glutamine: 2.35 ppm, alanine: 1.48, glycine and / or myoinositol: 3.55 ppm.

Clinical severity score (Bruun et al., 2018); A clinical severity score was applied including three clinical features.

- 1) GDD/ID scored 0 = normal, 1 = borderline or mild, 2 = moderate, 3 = severe;
- 2) Seizures, 0 = none, 1 = occasional seizures with no anti-epileptic drug (AED) treatment, 2 = AED responsive seizures, 3 = AED resistant seizures;
- 3) Behavioural disorder, 0 = none; 1 = autism spectrum disorder (ASD) OR hyperactivity OR attention deficit and hyperactivity disorder (ADHD), 2= >1 behavioural disorder with no aggressive or self-injurious behaviour, 3= >2 behaviour disorder or aggressive and self-injurious behaviour.

Scores of all three clinical features were summed and phenotype defined as mild = 0–3, moderate = 4–6 and severe = 7–9.

Bruun, T. U. J., Sidky, S., Bandeira, A. O., Debray, F. G., Ficicioglu, C., Goldstein, J., . . . Mercimek-Andrews, S. (2018). Treatment outcome of creatine transporter deficiency: international retrospective cohort study. *Metab Brain Dis*, 33(3), 875-884. doi:10.1007/s11011-018-0197-3