

Social outcome in adults with maple syrup urine disease (MSUD)

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Summary Background: In MSUD, dietary treatment aims at the protection of the brain from functional disturbances and structural damage by keeping the branched-chain amino acids in plasma permanently in the near-normal range. Unfortunately, delay in effective treatment of the neonatal manifestation and poor long-term metabolic control are common in MSUD patients, leading to impaired cognitive outcome. Some studies have analysed cognitive capacity but only few data are available on social status (educational qualification, interpersonal relationships, lifestyle) in adult patients, which is one measure for the success of treatment. **Aim:** In this study we analysed sociodemographic data of 22 adult patients suffering from classic or very severe variant MSUD in comparison with data on an age-matched control collective of the population of Germany. **Results:** The

analysis revealed low educational and professional levels with a low rate of participation in the labour force by adult MSUD patients. The educational level in patients of migrant origin was lower than the educational qualifications in the native patient group. A large number of patients did not live a normal adult life. Patients could not live autonomously, did not have a steady partnership and had no children. **Conclusion:** Despite substantial improvement in the treatment of MSUD in recent years, unimpaired outcome of patients with classic or very severe variant MSUD remains a rare finding. Care must be further optimized in order to improve intellectual and, thereby, social outcome. Particular care must be exercised in the treatment of migrant patients who offer special problems due to cultural peculiarities and language difficulties.

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Electronic Supplementary Material

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