

Renal involvement in a patient with cobalamin A type (cblA) methylmalonic aciduria: a 42-year follow-up

A Haarmann¹, M Mayr, S Kölker, E R Baumgartner, J Schnierda, H Hopfer, O Devuyst, M R Baumgartner

Affiliations + expand

PMID: 24095221 DOI: [10.1016/j.ymgme.2013.08.021](https://doi.org/10.1016/j.ymgme.2013.08.021)

Free article

Abstract

Chronic renal failure is a well-known long-term complication of methylmalonic aciduria (MMA-uria), occurring even under apparently optimal metabolic management. The onset of renal dysfunction seems to be dependent on the type of defect and vitamin B12-responsiveness. We report on a patient with a vitamin B12-responsive cobalamin A type (cblA) MMA-uria caused by a homozygous stop mutation (p.R145X) in the cobalamin A gene (MMAA). She was diagnosed with chronic kidney disease (CKD) stage III at the age of 12 years. Following re-evaluation, the patient received vitamin B12 (hydroxocobalamin) treatment, resulting in a significant decrease in the concentration of methylmalonic acid (MMA) in urine and plasma. Until age 29 years glomerular filtration rate remained stable probably due to hydroxocobalamin treatment slowing down progression to end-stage renal failure. Kidney biopsies showed non-specific manifestations of chronic interstitial inflammation. The patient received a renal transplant at age 35 years. Under continuous treatment with hydroxocobalamin there is no evidence of kidney damage due to MMA-uria until the last follow-up 6 years after transplantation. This case report illustrates (i) a long-term follow-up of a patient with MMA-uria due to cblA deficiency, (ii) the involvement of the kidney as a target organ and (iii) the importance of early and adequate vitamin B12 substitution in responsive patients. Further investigation will be necessary to prove the protective effect of hydroxocobalamin in the kidney in vitamin B12-responsive patients.