INTRODUCTION

3-Methylcrotonylglycinuria (MCG) is an inborn error of the leucine catabolism resulting from isolated biotin-insensitive deficiency of 3-methylcrotonyl-CoA carboxylase (3 MCC), the enzyme converting 3-methylcrotonyl-CoA to 3-methylglutaconyl-CoA (1).

The metabolic phenotype characterizing MCC deficiency is the elevated excretion of the diagnostic compounds 3-methylcrotonylglycine and 3-hydroxyisovaleric acid, and the presence of abnormally elevated blood levels of 3-hydroxyisovaleryl carnitine (C5-OH), as determined by tandem mass spectrometry (MS/MS). Expanded newborn screening for inborn errors of metabolism using MS/MS has demonstrated that 3-MCC deficiency is one of the most commonly detected inherited organic acidurias. Patients with MCG, which is inherited as an autosomal recessive trait, show a highly variable clinical phenotype, ranging from asymptomatic to severe presentation. In the latter phenotype, episodes of acute metabolic decompensation can lead to coma, lethargy, and death (2).

PATIENT AND METHODS

The authors present six cases in a universe of thirty patients with an increase of C5-OH in the acylcarnitine profile.

Blood spot samples from newborns are collected between day 3 and 6 in Watman 903 filter paper. Acylcarnitines in samples are analysed by MS/MS (3). Molecular characterization of genes MCCA and MCCB that encodes the enzyme 3-MCC were studied by reported methods.

RESULTS

Table 1 presents the molecular results and C5-OH value obtained from the Guthrie card of the six cases.

REFERENCES