Clinical and Molecular Evaluation of 15 Korean MCAD Patients Detected by Newborn Screening

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Introduction

Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency is a rare metabolic disorder that prevents the body from converting medium chain fats to energy during fasting. The MCAD gene 985A>G (p.K329E) is not common within the Asian population. We report on Korean MCAD molecular characteristics.

Biochemical genetic testing including plasma acylcarnitine and urine organic acid analysis, IVP (in vitro probe assay) and sequencing of ACADM was performed on a Korean family with a newborn who had an elevated octanoyl (C8) carnitine concentration by newborn screening (NBS). Preventive and management protocol have been used.

Methods

Biochemical genetic testing including plasma acylcarnitine and urine organic acid analysis, IVP (in vitro probe assay) and sequencing of ACADM was performed on a Korean family with a newborn who had an elevated octanoyl (C8) carnitine concentration by newborn screening (NBS). Preventive and management protocol have been used.

Results

With the exception of one symptomatic patient, all MCAD patients in Korea were detected by NBS. All patients had mildly elevated blood C8 and C10:1 carnitine and hexanoylglycine, suberylglycine or 3-phenylpropionylglycine in urine. The common mutations are 1189T>A (Y397N) and 449_452delCTGA in Koreans. All Korean MCAD patients used preventative care and emergency management protocol, avoiding fasting. All patients have normal growth and development.

Conclusion

449_452delCTGA, 1189T>A(Y397N) mutation is common in Korea. We didn’t find the common Caucasian mutation, 985A>G (p.K329E), in our study. MCAD emergency management protocol can prevent metabolic crisis and brain damage in newborns.

Reference