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Short Communication

Citrullinemia type I: molecular screening of the *ASS1* gene by exonic sequencing and targeted mutation analysis

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ABSTRACT. We developed a mutation-screening protocol for the *ASS1* gene in order to guide clinical management of neonates with elevated citrulline detected during routine newborn screening. An exon-based amplification and sequencing method was designed and successfully applied to patients to identify disease-associated mutations. The sequencing-based method was applied to three patients with mild or asymptomatic clinical courses. Identification of a homozygous mutation in these patients, c.787G>A (p.Val263Met), led to the development of a tetra-primer ARMS-PCR method that successfully detected the mutation in DNA extracted from blood or from Guthrie card spots.

Key words: *ASS1* gene; Tetra-primer ARMS-PCR; Sequencing; Citrullinemia type I

