PRINT: ISSN 0972-3757 ONLINE: ISSN 2456-6330
International Journal of
HUMAN GENETICS

Full text open access online (Since 2001)

PRINT: ISSN 0972-3757 ONLINE: ISSN 2456-6330
Int J Hum Genet, 26(2): 117-125 (2026)
DOI: 10.31901/24566330.2026/26.02.934

Comprehensive Analysis of Persistent Isolated Hypermethioninemia: A Single Centre Study in Korea Over Two Decades

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KEYWORDS Aminoacid Metabolism. Genetic Testing. Hypermethioninemia. Neonatal Screening. Phenotype

ABSTRACT Persistent isolated hypermethioninemia (PIH) is a metabolic condition primarily caused by methionine adenosyltransferase I/III (MATI/III) deficiency due to a variant in the MAT1A gene. This retrospective study aimed to evaluate the prevalence, genetic spectrum, and clinical outcomes of PIH identified through newborn screening (NBS) at a single centre in Korea. A total of 46 patients referred for hypermethioninemia between March 1999 and July 2023 were reviewed. Among 46 patients, 30 were diagnosed with PIH, 8 with homocystinuria, and 8 with transient hypermethioninemia. PIH patients showed a significant decrease in methionine levels over time. Genetic analysis revealed *MAT1A* variants, including the novel variant R84K, in all 30 PIH patients. The study underscores the importance of NBS in early detection and suggests that PIH typically presents a favourable prognosis, emphasising the need for expanded screening and tailored management strategies.