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

Full text open access online (Since 2001)
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PRINT: ISSN 0972-3757 ONLINE: ISSN 2456-6330
Int J Hum Genet, 25(3): 114-120 (2025)
DOI: 10.31901/24566330.2025/25.3.919

Disorders of Lipid Metabolismin Children with Clinical Phenotypic Features Based on Second Generation Sequencing

Weiyuan Wang[1] and Tingting Yu[2,*]

[1]Department of Pediatrics, The Fourth Affiliated Hospital of Nanjing Medical University, Nanjing City, Jiangsu Province, China
[2]Department of Pediatrics, Nanjing Lishui People's Hospital, Nanjing City, Jiangsu Province, China

KEYWORDS Gene Mutation. Genetic Association Studies. High-Throughput Nucleotide Sequencing. Lipid Metabolism Disorders. Phenotype

ABSTRACT This study aimed to explore pathogenic genes associated with pediatric dyslipidemia using second-generation sequencing and analyse their correlation with clinical phenotypes. The researchers retrospectively analysed clinical and genetic data from 20 pediatric patients. Skin xanthoma (60%) and hypercholesterolemia (55%) were common presentations. Second-generation sequencing targeting lipid metabolism genes was performed. Causative mutations were identified in 14 of 18 tested patients, involving genes such as LDLR, LPL, ABCG5, ABCA1, and HFE2. Distinct genotype-phenotype correlations were observed, for example, ABCG5, HFE2, LDLR, and ABCA1 mutations were associated with xanthomas, while LPL and LDLR mutations were linked to hypertriglyceridemia and hypercholesterolemia patterns, respectively. These findings suggest that pediatric dyslipidemia presents with diverse clinical phenotypes that are strongly associated with mutations in specific genes. Molecular genetic diagnosis based on sequencing significantly improves clinical assessment and deepens the understanding of these disorders, offering valuable guidance for early detection and precision medicine.