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MOLECULAR GENETIC CHARACTERIZATION AND PREVALENCE OF FATTY ACID OXIDATION DISORDERS IDENTIFIED THROUGH NEWBORN SCREENING IN UKRAINE

Background. Fatty acid oxidation disorders (FAODs) are a group of inherited metabolic diseases caused by defects in mitochondrial β -oxidation of fatty acids or carnitine transport. Since October 2022, expanded newborn screening for 21 inherited disorders, including six FAODs (CUD, MCADD, VLCADD, LCHADD, TFP, and MADD), has been implemented in Ukraine. This program enabled evaluation of the prevalence, molecular spectrum, and genetic characteristics of FAODs in the Ukrainian population. **Methods.** Molecular genetic analysis was performed using NGS on the Ion GeneStudio S5 (Thermo Fisher Scientific) and confirmed by Sanger sequencing on the 3500DX Genetic Analyzer (Applied Biosystems). The potential clinical significance of previously unreported variants was assessed using the automated variant interpretation platforms VarSome and Franklin. **Results.** Between October 2022 and December 2025, a total of 519,535 newborns were screened through the expanded newborn screening program in Ukraine. FAODs were confirmed in 25 newborns. The identified disorders included 14 cases of MCADD (incidence 1:37110; 56% of all FAODs cases), 5 cases of CUD (1:103907; 20%), 3 cases of LCHADD (1:173178), 1 case of VLCADD (1:519535), and 1 case of MADD (1:519535). In addition, one newborn with CPT II deficiency was diagnosed during differential diagnostic evaluation (1:519535). The major variant in the ACADM gene among MCADD patients was p.Lys329Glu (85,7%). One MCADD patient was homozygous for the ACADM variant c.31-2A>G. In patients with CUD, the most common SLC22A5 variants were p.Thr232Met (20%), c.-149G>A (20%), and p.Val151 = (20%). The study identified three previously unreported pathogenic missense variants: the ACADM (p.Ile-134His), SLC22A5 (p.Glu111Gly), and ETFDH (p.Leu327Pro). **Conclusion.** MCADD (56% of FAOD cases) and CUD (20%) were the most common FAODs identified through expanded newborn screening in Ukraine. Molecular genetic analysis revealed a predominance of the ACADM p.Lys329Glu variant and identified three novel likely pathogenic variants in the ACADM, SLC22A5, and ETFDH genes. These findings improve understanding of the molecular epidemiology of FAODs in the Ukrainian newborn population.

Keywords: FAODs, NGS, screening.

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