Genetics • Genomics • Genetic engineering • Biotechnology • Bioinformatics

eISSN 2566-431X

Genetics & Applications

An Aspiring Interdisciplinary Journal of Genetic Research

special edition

ABMBBIH CONFERENCE 2023

International Conference of Biochemists and Molecular Biologists in Bosnia and Herzegovina

The Official Publication of the
Institute for Genetic Engineering and Biotechnology
University of Sarajevo

ISSN 2566-431X

ISSN 2566-2937

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An Aspiring Interdisciplinary Journal of Genetic Research

Volume 7, Number 2

Special edition

Book of abstracts

International Conference of Biochemists and Molecular Biologists in Bosnia and Herzegovina - ABMBBIH

May, 2023

Indexed/Abstracted

This journal is indexed or abstracted by:

EBSCO, *DOAJ*, *CAB Abstracts*, *Google Scholar*, *Global Health database, Crossref*, *Index Copernicus, EuroPub, Scilit and MIAR.*

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Publisher of *Genetics & Applications*

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Session 3 – Poster presentation *S3-15*

HIGH-RISK POPULATION SCREENING FOR FABRY DISEASE IN PATIENTS WITH CHRONIC RENAL FAILURE OF UNKNOWN ETIOLOGY

Parezanovic Marina, Andjelkovic Marina, Stevanovic Nina, Spasovski Vesna, Ugrin Milena, Komazec Jovana, Klaassen Kristel, Stankovic Sara, Pavlovic Sonja, Stojiljkovic Maja, Skakic Anita

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Fabry disease (FD) is a rare X-linked disorder caused by variants in the GLA gene leading to the deficiency of lysosomal α-galactosidase-A and progressive accumulation of globotriaosylceramide affecting the heart, nervous system, and kidneys. FD has overlapping phenotypes and often remains undiagnosed. Therefore, a precise molecular-genetic diagnosis and the earliest possible treatment are essential to avoid significant disease progression. The study aimed to determine the strategy for establishing routine molecular genetic diagnostics of FD in Serbia to provide an early application of appropriate therapy and genetic advice to families with a high risk for the birth of a child with FD. We analyzed 95 (34 female and 61 male) hemodialysis patients with clinical suspicion of FD using Sanger sequencing of all coding exons (7) and flanking intron regions of the GLA gene and measured the relative expression of the GLA gene in available samples. The genetic analysis revealed 3 patients with a missense variant (p.Asp313Tyr), and 10 patients with combinations of non-coding variants, described as complex intronic haplotypes (CIHs). CIH1 (c.-10C>T, c.370-81_370-77delCAGCC, c.640-16A>G, c.1000-22C>T), the most frequent haplotype, was detected in 7 (7.4%) patients. Lyso-Gb3 biomarker levels were within the normal range in each tested patient. However, RT-qPCR analysis revealed decreased relative expression of the GLA gene in PBMC of 2 female patients with CIH1 and one female patient carrying only c.-10C>T variant by 9,1%, 7,4%, 46,3%, respectively, pointing out that further analyses are needed to confirm/exclude FD in these patients. Because the effects of CIHs are not yet fully understood, our work highlights the importance of analyzing intronic regions of the GLA gene as genetic modifiers and the need to include expression analysis in the diagnostic algorithm.

Keywords: Fabry disease, precise molecular-genetic diagnosis, high-risk population screening

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