

P03.03B / B - **COL4A5 G624D: abundance of the Alport syndrome mutation in Russia along with Greek, Hungarian and Slovenian populations suggests it is a frequent mutation in Eastern Europe with mild phenotype**

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 Poster Area

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Disclosures

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Abstract

Introduction: Alport syndrome (AS) is a familial hematuria caused by mutations in *COL4A3*, *COL4A4* and/or *COL4A5* genes which lead to defects in glomerular filtration barrier. So far, some founder mutations have been identified with many of them being region- or ethnicity-specific. *COL4A5* c.1871G>A, p.(Gly624Asp) pathogenic variant is known to be prevalent in AS patients from Slovenia (6 out of 17), Hungary (3 out of 10) and Greece and lead to late age at onset of end stage renal disease. In contrast to these populations, the mutation is considered to be rare in the US, Northern and Western Europe or Japan. Here we show that the mutation was detected in 7 AS patients out of 49 with genetically confirmed diagnosis from Russia. Materials and methods: The population sample contained 76 apparently unrelated pediatric patients (1 to 17 years old) from diverse range of regions in the European part of Russian Federation with confirmed or suspected diagnosis of AS according to current guidelines. NGS sequencing was performed using Ion PGM (AmpliSeq panel). Results: We confirmed the diagnosis in 49 patients including 43 with X-linked AS (harboring *COL4A5* gene mutations) and 1 with digenic *COL4A5* and *COL4A3* inheritance. Seven of them were bearing the *COL4A5* c.1871G>A, p.(Gly624Asp) mutation which corresponds to 14% frequency in the sample. We demonstrate that the mutation is characterized by late age at onset of hematuria (>48 months) and absence of proteinuria in childhood. The research was supported by RFBR grant 18-34-00708 to L.I.S. and Minzdrav government grant №115022070016.