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Abstract Topic: - Molecular effects of genetic variation

Abstract Title: - Association of Copy Number Variation in Complement Factor H and Complement Factor H-related genes with Chronic Kidney Disease of Unknown Etiology (CKDu).

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Aims: - Our aim was to investigate the significance and relevance of systemic assessment of complement genes in individuals diagnosed with chronic kidney disease of unknown etiology (CKDu).

Methods: - It was a single-centric prospective case-control study that examined the occurrence of chronic kidney disease of unknown etiology (CKDu) in patients between the period of 2021-2023. A total of 51 patients and 39 controls were enrolled. Complement Factor H-related (CFHR) gene testing via Multiplex Ligation-dependent Probe Amplification (MLPA) was performed in all enrolled samples to identify larger deletions and duplications in the CFHR gene. Chi-square analysis was performed for the association between the case and control group by estimating the odds ratio (OR) and 95% confidence intervals (95% CI).

Results: - Among the 51 CKDu patients, 36 (70.6%) were males and 15 (29.4%) were females. Out of 39 controls, there were 23 (59%) males and 16 (41%) females. Complement Factor H-related CFHR3/CFHR1 gene, duplication was found in 28 (54.9%) patients and 11 (28.21%) controls. Duplication of CFHR3/CFHR1 has been associated with risk of developing CKDu [OR = 3.099; CI = (1.27-7.54); p = 0.011]. The CFHR3/CFHR1 homozygous deletion was 4 (7.84%) CKDu patients and 6 (15.38%) controls. CFHR4 gene duplication was observed in 1 (1.96%) of patients, with no occurrences in the control group, while heterozygous deletion was found in 1 (1.96%) of patients and 1 (2.56%) of controls. CFHR5 gene heterozygous deletion was observed in 1 (1.96%) patient only. Heterozygous deletion in the CFH gene was present in 5 (9.8%) of patients and 6 (15.38%) of controls.

Conclusions: - Duplication of the CFHR3/CFHR1 gene has been associated with the risk of CKDu. However, a larger sample size study is required to understand the association of CFHR3/CFHR1 duplication with CKDu.

Keywords: - Among the 51 CKDu patients, 36 (70.6%) were males and 15 (29.4%) were females. Out of 39 controls, there were 23 (59%) males and 16 (41%) females. Complement Factor H-related CFHR3/CFHR1 gene, duplication was found in 28 (54.9%) patients and 11 (28.21%) controls. Duplication of CFHR3/CFHR1 has been associated with risk of developing CKDu [OR = 3.099; CI = (1.27-7.54); p = 0.011]. The CFHR3/CFHR1 homozygous deletion was 4 (7.84%) CKDu patients and 6 (15.38%) controls. CFHR4 gene duplication was observed in 1 (1.96%) of patients, with no occurrences in the control group, while heterozygous deletion was found in 1 (1.96%) of patients and 1 (2.56%) of controls. CFHR5 gene heterozygous deletion was observed in 1 (1.96%) patient only. Heterozygous deletion in the CFH gene was present in 5 (9.8%) of patients and 6 (15.38%) of controls.