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Prevalence of Predicted Pathogenic COL4A3–COL4A5 Variants in the Korean General Population

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Objectives : Alport syndrome (AS) has traditionally been considered a rare inherited kidney disease with an estimated prevalence of 1 in 50,000. However, recent analyses of population genomic datasets such as gnomAD and the Exome Variant Server suggest that pathogenic COL4A3–COL4A5 variants may be more common than previously recognized. The prevalence of AS and the frequency of pathogenic variants in COL4A3, COL4A4, and COL4A5 in the Korean population remain unclear. This study aimed to investigate the frequency of predicted pathogenic variants in COL4A3–COL4A5 using population genomic datasets from Korea.
Methods : Genome data from 9,687 individuals in the National Biobank of Korea Pilot Project (Phase 2), the Korean Genome and Epidemiology Study (KoGES; 4,998 unrelated adults), and the Korean Variant Archive (KOVA) were analyzed. These datasets represent the Korean general population and enable assessment of predicted pathogenic COL4A variants in individuals not selected for kidney disease. Predicted pathogenic variants were classified according to ACMG/AMP criteria incorporating population frequency and in silico pathogenicity prediction.
Results : Predicted pathogenic COL4A3–COL4A5 variants were identified in 138 individuals from rare disease families and 15 individuals from the general population. In the general population, two unique variants were identified in COL4A3, 11 in COL4A4, and two in COL4A5. Predicted pathogenic COL4A3 or COL4A4 variants were present in 1 in 384 individuals, whereas COL4A5 variants were present in 1 in 2,499. Among the 15 unique variants identified in the general population, 11 involved glycine substitutions within collagenous domains and four were located in non-collagenous domains. No compound heterozygous or digenic variants were identified.
Conclusions : Predicted pathogenic COL4A3–COL4A5 variants are not rare in the Korean general population and show a prevalence comparable to that reported in European populations. These findings may support earlier recognition of individuals at risk for AS and facilitate early diagnosis and prevention.

[AS prevalence] KSN_table.png

Gene	Prevalence of predicted pathogenic variant		
	Rare disease	**KoGES**	**KOVA**
COL4A3	1 per 109 1	1 per 2,499	27 variants
COL4A4	per 285 1 per	1 per 454 1	19 variants
COL4A3* or *COL4A4	79 1 per 646	per 384 1	46 variants
COL4A5		per 2,499	3 variants