



Congress Abstract

Alport Syndrome: Current Concepts

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ABSTRACT

Background: Alport syndrome is an inherited disorder of the glomerular basement membrane caused by pathogenic variants in COL4A3, COL4A4, or COL4A5. The disease is characterized by persistent microscopic hematuria, progressive proteinuria and chronic kidney disease, with possible hearing and ocular involvement. Early recognition is important for timely nephroprotective management and family screening.

Methods: This narrative review synthesizes the available information on the molecular basis, inheritance patterns, clinical manifestations, diagnostic approaches, monitoring, and treatment of Alport syndrome. Particular attention was given to genetic testing, renal manifestations, extrarenal features, and nephroprotective strategies.

Results: Alport syndrome demonstrates X-linked, autosomal recessive, and autosomal dominant inheritance patterns. X-linked disease is associated predominantly with COL4A5 variants, whereas COL4A3 and COL4A4 variants are responsible for autosomal forms. Persistent hematuria is an important early renal manifestation and may progress to proteinuria, glomerulosclerosis, chronic kidney disease, and end-stage renal disease. Hearing impairment and ocular abnormalities may provide additional diagnostic clues. Molecular genetic testing is an important approach to diagnosis and classification. Screening of relatives is valuable for identifying affected or at-risk family members. Nephroprotective therapy, particularly renin-angiotensin-aldosterone system blockade with angiotensin-converting enzyme inhibitors or angiotensin receptor blockers, is emphasized in patients with persistent proteinuria. Long-term management also requires renal function monitoring and audiological and ophthalmological assessment.

Conclusion: Alport syndrome requires early recognition of persistent hematuria and appropriate genetic evaluation. Timely nephroprotective treatment, family screening, and multidisciplinary follow-up may help slow progression of kidney disease and improve comprehensive patient care.

Keywords: Alport Syndrome; COL4A3; COL4A4; COL4A5; Hematuria; Nephroprotection