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**ALPORT SYNDROME: A MULTISYSTEM DISEASE AND THE POTENTIAL OF MODERN TREATMENT APPROACHES**

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# ALPORT SYNDROME: A MULTISYSTEM DISEASE AND THE POTENTIAL OF MODERN TREATMENT APPROACHES

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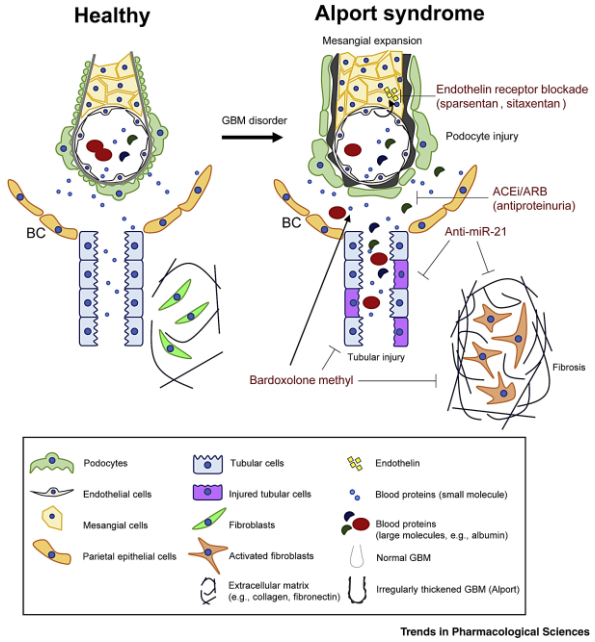
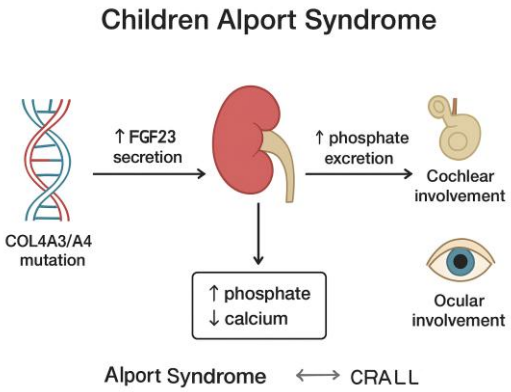
**Research Relevance**: Alport syndrome is a rare but significant genetic disorder that affects multiple systems of the body, including the kidneys, hearing, and vision. It is one of the leading causes of inherited kidney failure, which progresses with age. Despite advancements in diagnostics and treatment, issues of early detection and appropriate therapy remain relevant in nephrology, ophthalmology, and otolaryngology. Effective treatment and early diagnosis are crucial for slowing disease progression and improving the quality of life of patients.




**Research Goal:** The goal of the study is to analyze the genetic, clinical, and therapeutic aspects of Alport syndrome, as well as to assess the effectiveness of modern diagnostic and treatment methods, with the aim of improving prognosis and therapeutic approaches.

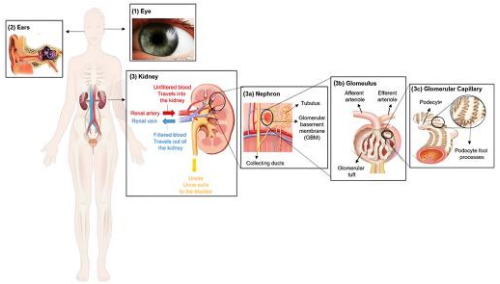
**Materials and Methods:** Genetic testing (analysis of mutations in the COL4A3, COL4A4, COL4A5 genes), kidney biopsy with electron microscopy, audiometry, ophthalmological examination, application of ACE inhibitors and angiotensin receptor blockers (ARBs), analysis of the effectiveness of kidney transplantation and the use of hearing




**Results:** The study yielded important data on the diagnosis, treatment, and prognosis of Alport syndrome.

Genetic testing, including the analysis of mutations in the COL4A3, COL4A4, and COL4A5 genes, allows for a high degree of accuracy in confirming the diagnosis of Alport syndrome. This also provides an opportunity for genetic counseling for the patient's family members, which is crucial for identifying the risk of the disease in close relatives. Kidney biopsy with electron microscopy, revealing thinning and breaks in the basement membrane of glomeruli, plays an important role in the morphological confirmation of the diagnosis. This method helps to distinguish Alport syndrome from other nephropathies with similar clinical manifestations. Audiometry and ophthalmological examination are also integral parts of the diagnosis. Audiometry identifies bilateral sensorineural hearing loss, which often develops during adolescence, while ophthalmological examination reveals changes in the eyes, such as anterior lenticonus and maculopathy, confirming the multisystem nature of the disease. Treatment of Alport syndrome is aimed at slowing the progression of kidney failure and correcting extrarenal symptoms. The use of ACE inhibitors and ARBs has proven effective in reducing proteinuria levels and stabilizing blood pressure, which helps slow the progression of chronic kidney disease.

These drugs are used in the early and middle stages of the disease, helping to prevent or delay the onset of kidney failure. Additionally, hearing aids and other devices help patients with hearing loss maintain their quality of life. Regular ophthalmological monitoring is important for preventing vision deterioration and intervening promptly when retinal changes progress. In cases where kidney failure reaches the terminal stage, patients require dialysis or kidney transplantation, which is a necessary measure for stabilizing their condition.



**Conclusion**: Alport syndrome is a multisystem genetic disorder requiring comprehensive diagnosis and management. Genetic testing and kidney biopsy confirm the disease, while modern treatments—ACE inhibitors, ARBs, and kidney transplantation—slow kidney failure and improve prognosis. Hearing aids and regular eye monitoring reduce extrarenal complications. Early detection and timely therapy are key to better long-term outcomes.

Alport Syndrome: Achieving Early Diagnosis and Treatment – PubMed https://pubmed.ncbi.nlm.nih.gov/32712016/
Alport syndrome: Hereditary nephropathy associated with mutations in genes coding for type IV collagen chains – PubMed https://pubmed.ncbi.nlm.nih.gov/27816395/
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