Alport Syndrome

Alport syndrome is a genetic cause of nephritic syndrome commonly inherited in an X-linked dominant pattern. It is caused by a mutation in type IV collagen, which is an important structural component of basement membranes present in the kidney, eyes, and ears. Therefore, patients with this syndrome develop hematuria with progression to chronic renal failure along with deafness and various eye disorders. The abnormalities of type IV collagen in the kidney basement membranes lead to gradual scarring of the kidneys with progression to chronic renal failure. Progression of the disease leads to thinning of the basement membrane with a split lamina densa.

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Pathophysiology

Nephritic
Nerd-cricket

Nephritic syndrome is characterized by inflammation of the glomeruli and includes the following clinical features: hematuria, hypertension, oliguria, and less than 3.5 grams per day of proteinuria. Patients with nephritic syndrome also commonly present with red cell casts in the urine and azotemia. Nephritic syndrome can be caused by several diseases including Alport syndrome, Berger's disease, poststreptococcal glomerulonephritis and rapidly progressive glomerulonephritis.

X-linked
X-suit

This disease is inherited as an X-linked trait in approximately 85% of cases. It is commonly called X-linked dominance because males express the full syndrome while female carriers can have manifestations of disease like hematuria. Alport syndrome can also be inherited in an autosomal recessive fashion, as well as autosomal dominant manner in about 5% of cases.

Mutation in Type IV Collagen
Mutants and (4) Fork in Cola-gem

Alport syndrome is caused by a mutation in type IV collagen, which is an important structural component of basement membranes present in the kidney, eyes, and ears.

Diagnosis

Split Basement Membrane
Split Basement

Progression of the disease leads to thinning of the basement membrane with a split lamina densa.

Basket Weave Appearance
Basket

On microscopy, the split basement membranes in Alport syndrome show irregular thinning and thickening, described as a lamellated "basket weave" appearance.

Signs and Symptoms

Ocular Disorders
Crazy Eyes

Various ocular disorders are a major component of Alport syndrome because type IV collagen is abundant in the ocular system. Manifestations include posterior cataracts and corneal dystrophy. A pathognomonic ocular feature of Alport syndrome is anterior lenticonus.

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Deafness

Headphones

Difficulty hearing results because type IV collagen is an essential piece of the auditory system. Type IV collagen is a crucial component of the cochlea in the ear. Therefore, a mutation in type IV collagen can cause deafness, which is a major component of Alport syndrome. However, auditory defects may be subtle as well.

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