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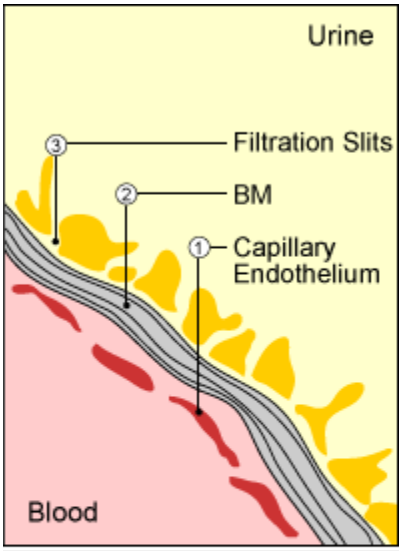
Alport syndrome



Filtration barrier in the kidney. To enter urine, waste molecules pass through: 1, gaps between the lining of the blood vessel; 2, the BM; and 3, slits between epithelial cells. In AS, the BM deteriorates, allowing proteins and red blood cells to enter the urine.

Alport syndrome (AS) is a genetic disease in which a collagen mutation affects the kidneys, the ears, and the eyes. The syndrome was named for Dr. Alport who in 1927 described a British family in which many members developed renal disease as well as deafness. He noted that affected men in the family died as a result of their kidney problems, whereas females were less affected and lived until old age.

It is now known that most cases of AS are caused by a mutation in the collagen gene *COL4A5*. This gene encodes for the alpha-5 chain of collagen type IV and is located on the X chromosome. Because women have two X chromosomes (XX), affected women usually have one normal copy and one abnormal copy of the gene. Men only have one copy of the X chromosome (XY). If they inherit the *COL4A5* mutation, this abnormal copy of the gene is the only copy they have and the effects are more severe.

Type IV collagen is found in basement membranes (BM), which are selective barriers between cells. In the kidney, the glomerular BM filters waste products into the urine while keeping useful molecules within the blood stream. In AS, the abnormal collagen disrupts this filter, leading to the loss of proteins and red blood cells into the urine. Blood in the urine (hematuria) is a sign common to all types of AS. In the ear, abnormal collagen in the cochlea results in a progressive deafness in which the ability to hear high tones is lost first. Abnormal collagen can also affect the lens of the eye.

Currently, renal failure due to AS is treated by dialysis or, for some, renal transplantation. However, gene therapy may one day be able to provide a cure for AS by replacing the faulty *COL4A5* gene.

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