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Literary Research on Alport syndrome

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Alport syndrome is rare genetic disorder of the glomeruli in the kidneys that can lead to kidney failure, hearing loss, and eye abnormalities. The production of collagen in the glomeruli, which are the microfiltration units inside the kidneys, is affected by mutations in the alpha-3, alpha-4, and alpha-5 chains of type IV collagen. These mutations cause a decrease in the production of alpha-3, alpha-4, and alpha-5 chains, leading to the development of Alport syndrome.

The pathophysiology of Alport syndrome is complex in nature. According to the information obtained from Alport Syndrome Foundation websites, the syndrome is caused by genetic mutations that affect the glomeruli, which are the microfiltration units inside the kidney. The glomeruli contain GBM (glomerular basement membrane) that allows filtration of blood through the membrane. The normal structure of the GBM is replaced by scarring tissue, leading to failure in the filtration system causing kidney failure in AS. Type IV collagen is a major part of important tissue structures called basement membranes that are present in the kidneys. These proteins spread and result in GBM thickening and impairment of selectivity with subsequent glomerular sclerosis, interstitial injuries, and renal failure. Type IV collagen comes from a family of six proteins known as alpha-1 through alpha-6. Mutation in alpha-1, alpha-4, and alpha-5 chains cause Alport syndrome. K-linked AS is the most common form that accounts for 80% to 85% of the cases and results from mutations in the alpha-5 chain type IV collagen found inside gene COL4A5 (Kaewpoowat, 2012). Autosomal Recurrent AS accounts for 15% to 25% of the cases and is caused by mutations in the alpha-3 or alpha-4 chains found in COL4A3 or COL4A4 genes. Finally, the rare cases of autosomal dominant AS is due to heterozygous transformation in COL4A3 or COL4A4 also occur in 5% of cases (Banares, Morrow, & Mercuro, 2010).

Signs and Symptoms

According to National kidney Foundation (SNF) patients with AS symptoms may include: Blood in the urine (hematuria), protein in the urine (proteins尿), and high blood pressure (hypertension). It causes damage to the kidney through scarring in the glomerular basement membrane (GBM) of a normal kidney structure. Studies published by NIHR also shows that 90% to 95% of people with AS-linked Alport syndrome develops hearing loss at some point in their lives, both gender with a mean age of 17 years. Research has shown that people with AS also have slow decline of vision, which may lead to cataracts formation and corneal edema (Khalil). Some people with this disease have abnormal pigment of the retina called dot-and-bluish retinal spots. Alport syndrome may also have symptoms in the nose, ankle, foot, and around the eyes (Iijima, Kami, Nakayama, Ito, Maniniha, & Maintz, 2010).

The top image obtained from Medscape website shows the electron micrograph of a kidney biopsy from a patient with Alport syndrome. Notice the scar tissue formation in the glomerular basement membrane (arrow signs).

Understanding Pathophysiology and its Significance

The pathophysiology of AS is complex in nature. According to the information obtained from Alport Syndrome Foundation websites, the syndrome is caused by genetic mutations that affect the glomeruli, which are the microfiltration units inside the kidney. The glomeruli contain GBM that allows filtration of blood through the membrane. The normal structure of the GBM is replaced by scar tissue, leading to a failure in the filtration system causing kidney failure in AS. Type IV collagen is a major part of important tissue structures called basement membranes that are present in the kidneys. These proteins spread and result in GBM thickening and impairment of selectivity with subsequent glomerular sclerosis, interstitial injuries, and renal failure. Type IV collagen comes from a family of six proteins known as alpha-1 through alpha-6. Mutation in alpha-1, alpha-4, and alpha-5 chains cause Alport syndrome. K-linked AS is the most common form that accounts for 80% to 85% of the cases and results from mutations in the alpha-5 chain type IV collagen found inside gene COL4A5 (Kaewpoowat, 2012). Autosomal Recurrent AS accounts for 15% to 25% of the cases and is caused by mutations in the alpha-3 or alpha-4 chains found in COL4A3 or COL4A4 genes. Finally, the rare cases of autosomal dominant AS is due to heterozygous transformation in COL4A3 or COL4A4 also occur in 5% of cases (Banares, Morrow, & Mercuro, 2010).

Implication for Nursing Care

AS plays an important role in secondary and tertiary prevention against AS. Institute for Work and Health states that the goal of secondary prevention is to slow the process of disease in its earliest stages (2014). Nurses need to continue encourage patients with the family history of AS to have regular exams and screening tests to prevent complications from such diseases. They educate patient about the importance of blood pressure management to prevent further complications. They educate patient about diet, limiting fluids, and other treatments options. They perform counseling and education to increase coping skills among patient with such disease. At dialysis center, nurses assist phlebotomists in performing hemodialysis and peritoneal dialysis on patient with renal failure secondary to AS. Nurses also can help patients in learning new skills such as lip reading or sign language and getting/hearing aids. They teach younger men with Alport syndrome the importance of using hearing protection in noisy environments.

Finally, the goal of tertiary prevention is to prevent further physical deterioration and maintain quality of life (Institute for Work and Health, 2014). It focuses on helping people manage complicated health problem such as hearing loss, blindness, and kidney failure secondary to AS. Nurses assist patients with AS by providing information about support groups such as Alport Syndrome Foundation and National Organization for Rare Disorders. They also provide advice about lifestyle modifications including diet, medication administration and other appropriate treatments. They educate patient undergoing kidney transplant secondary to end stage kidney failure. Some trained nurses also provide genetic counseling to the patient with AS because the disease is inherited. They also have patient with rehabilitation program and set up patient support groups.

References