Literary Research on Alport Syndrome

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Alport syndrome is a rare genetic disorder of the glomerulus in the kidneys that can lead to kidney failure, hearing loss, and vision problems. It affects the glomerular basement membrane (GBM) of a normal kidney structures. Studies published by Ungprasert and others show that 80% of boys with X-linked Alport syndrome (AS) have mutations in the alpha 5 chain of COL4A5, which is the most common form that accounts for 80% to 85% of the cases and results from defects in the alpha 5 chain. Type IV collagen is a major component of basement membrane, which is the microfilaments inside the kidney. The glomeruli contain GBM that allows filtration of blood through the membrane. The normal structures of the GBM are replaced by scar tissue, leading to failure in the filtration system causing kidney failure in the AS. Type IV collagen is a major component of basement membrane, which is responsible for the failure in the filtration system causing kidney failure in the AS. Type IV collagen comes from a family of six proteins known as alpha-1 through alpha-6. Mutations in alpha-5, alpha-4, and alpha-3 chains cause Alport syndrome. X-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 GBM. Recent studies have shown that mutations in COL4A3 and COL4A4 also occur in 5% of cases (Bassano, Marras, & Mercuro, 2010). The rare cases of autosomal dominant AS is due to heterozygous mutation in COL4A3 or COL4A4 also occur in 5% of cases (Bassano, Marras, & Mercuro, 2010).

Understanding Pathophysiology and its Significance

The pathophysiology of AS is complex and nature. According to the information obtain from Alport Syndrome Foundation website, the syndrome is caused by genetic mutations that affect the glomeruli which are the microfilaments inside the kidney. The glomeruli contain GBM that allows filtration of blood through the membrane. The normal structures of the GBM are replaced by scar tissue, leading to failure in the filtration system causing kidney failure in the AS. Type IV collagen is a major component of basement membrane, which is present in the kidneys. These proteins spread and result in GBM thickening and impairment of selectivity with subsequent glomerular sclerosis, interstitial fibrosis, and renal failure. Type IV collagen comes from a family of six proteins known as alpha-1 through alpha-6. Mutations in alpha-5, alpha-4, and alpha-3 chains cause Alport syndrome. X-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 GBM. Recent studies have shown that mutations in COL4A3 and COL4A4 also occur in 5% of cases (Bassano, Marras, & Mercuro, 2010). The rare cases of autosomal dominant AS is due to heterozygous mutation in COL4A3 or COL4A4 also occur in 5% of cases (Bassano, Marras, & Mercuro, 2010).

Implication for Nursing Care

Nurses play an important role in secondary and tertiary prevention against AS. For Work and Health states that the goal of secondary prevention is to slow the progression 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 disease. 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 patient in performing hemodialysis and peritoneal dialysis on patient with renal failure secondary to AS. Nurses also help patients in learning new skills such as lip reading or sign language and getting/giving ophthalmology. These proteins spread and result in GBM thickening and impairment of selectivity with subsequent glomerular sclerosis, interstitial fibrosis, and renal failure. Type IV collagen comes from a family of six proteins known as alpha-1 through alpha-6. Mutations in alpha-5, alpha-4, and alpha-3 chains cause Alport syndrome. X-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 GBM. Recent studies have shown that mutations in COL4A3 and COL4A4 also occur in 5% of cases (Bassano, Marras, & Mercuro, 2010). The rare cases of autosomal dominant AS is due to heterozygous mutation in COL4A3 or COL4A4 also occur in 5% of cases (Bassano, Marras, & Mercuro, 2010).

Signs and Symptoms

According to National kidney Foundation (NKF) patients with AS symptoms may include: Blood in the urine (hematuria), protein in the urine (proteinuria), and high blood pressure (hypertension). It causes damage to the kidney through scar formation in the glomerular basement membrane (GBM) of normal kidney structures. Studies published by Ungprasert and others show that 80% of boys with X-linked Alport syndrome (AS) develops hearing loss at some point in their lives. Both gender with autosomal dominant AS have childhood hearing loss and their parents develop hearing loss at a later stage. Finally studies published by Ungprasert shows that, people with AS also have slow decline of vision, which may lead to cataract formation and visual acuity loss. Some people with this disease have abnormal pigment of the retina called dot and strip retinal dystrophy. Alport syndrome may also cause neck, ankle, foot, and around the eyes (Iijima, Kunii, Kamei, Nakayama, Ino, Minamida & Mihama, 2010).

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There is no specific treatment available against AS. People with such disease are treated symptomatically. Kidney transplantation has shown some success in patient with and without kidney failure. Medical researchers are constantly working hard on understanding why people develop such disease and its treatments. Several Laboratory tests are being conducted on animals with AS to find the best treatment. (Temme, Kramer, Jager, Lange, Peters, Müller, & Gross, 2012). Overall the joint effort among nurses and other health care providers can help patient with AS to manage their disease.

References


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References Contd...