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Literary Research on Alport Syndrome

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Introduction

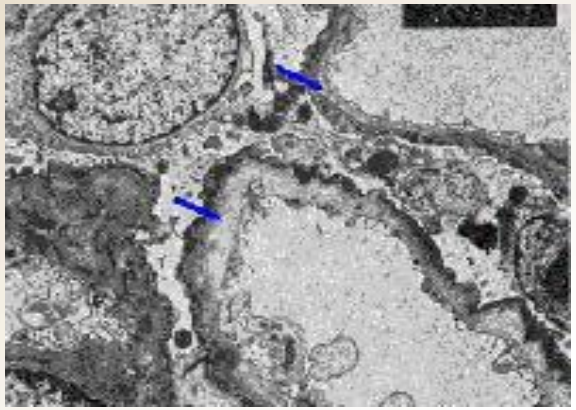
Alport syndrome is rare genetic disorder of the glomerulus in the kidneys that can be X-linked, Autosomal recessive, or Autosomal dominant in nature. Clinical manifestation includes hearing loss, hematuria, proteinuria, and hypertension (Cheungpasitporn, Kaewpoowat, Suksaranjit, Kittanamongkolchai, Srivali, Ungprasert, & Rangan, 2012). It is most common disease in males. A 24-hour urine specimen is usually obtained to check for proteinuria and elevated creatinine, which indicate acute renal failure (Cosgrove, 2012). A renal biopsy and ultrasounds is used to confirm the diagnosis of Alport Syndrome. Patients that are diagnosed with such disease require dialysis and referral to an ophthalmology and audiology. The following poster will help in understanding the signs and symptoms, pathophysiology, and nursing implication of Alport syndrome based on information found in literature research.

Signs and Symptoms

According to National kidney Foundation (NHF) 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 a normal kidney structures. Studies published by NHF also shows that 80% of boys with X-linked Alport syndrome develops hearing loss at some point in their lives. Both gender with autosomal recessive AS typically have childhood hearing loss and their parents develop hearing loss at a later stage. Finally studies published by NHF shows that, people with AS also have slow decline of vision, which may lead to cataract formation and corneal erosion if left untreated. Some people with this disease have abnormal pigment of the retina called dot-and-fleck retinopathy. Alport syndrome may also results in the swelling of legs, ankle, feet, and around the eyes (Iijima, Nozu, Kamei, Nakayama, Ito, Matsuoka & Matsuo, 2010).

Understanding Pathophysiology and its Significance

The pathophysiology of AS is complex in 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 microfiltration units inside the kidney. The glomeruli contain GBM that allows filtration of blood through the membrane. The normal structures of the GMB are replaced by scar tissue, leading to failure in the filtration system causing kidney failure in the 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 fibrosis, and renal failure. Type IV collagen comes from a family of six proteins know as alpha-1 through alpha-6. Mutation in alpha-3, alpha4, and alpha-5 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 of the alpha-5 chain type IV collagen found inside gene COL4A5 (Rheault, 2012). Autosomal Recessive AS accounts for 10% to 15% of the cases and is caused by mutation 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 (Bassareo. Marras, & Mercuro, 2010).



The above image obtained from Medscape website shows the electron micrograph of a kidney biopsy from a patient with Alport syndrome. Note the scar tissue formation in the glomerular basement membrane (see arrows).

Implication for Nursing Care

Nurses play 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 progress 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 complication from such disease. They educate patient about the importance of blood pressure management to prevent further complication. 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 physician 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 maximize 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 Rear Disorders. They also provide education about lifestyle modifications including diet, medication administration and other preventive measure for patients undergoing kidney transplant secondary to end stage kidney failure. Some trained nurses also provide genetic counseling to the patient with AS because the disorder is inherited. They also AS patient with rehabilitation program and setting up patient support groups. Assist

Conclusion

The content of this poster helped in identifying the common signs and symptoms of AS, pathophysiology, and its implication to nursing care. The disease is more common among man and can be diagnosed as early at 2 years old. This genetic disorder not only affects the kidney, but also their vision, and hearing leading to end stage renal failure, hearing loss, and blindness. It is important to provide patients with AS secondary and tertiary prevention methods to manage complications from such genetic disorder. Through early screening using genetic test can help in detecting this disease and can protect these patient from developing complication and injury. The evaluation can also be made using blood test, urine test, ultrasound, and kidney biopsy (Kashtan, Ding, Gregory, Gross, Heidet, Knebelmann, & Licht, 2013).

There is no specific treatment available against AS. People with such disease are treated symptomatically. Kidney transplantation has showed some success in-patient with end-sate 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 healthcare providers can help patient with AS to manage their disease.

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