THROMBOPHILIA AND RECCURENT THROMBOSIS OF AV FISTULAS IN PATIENT WITH ALPORT SYNDROME

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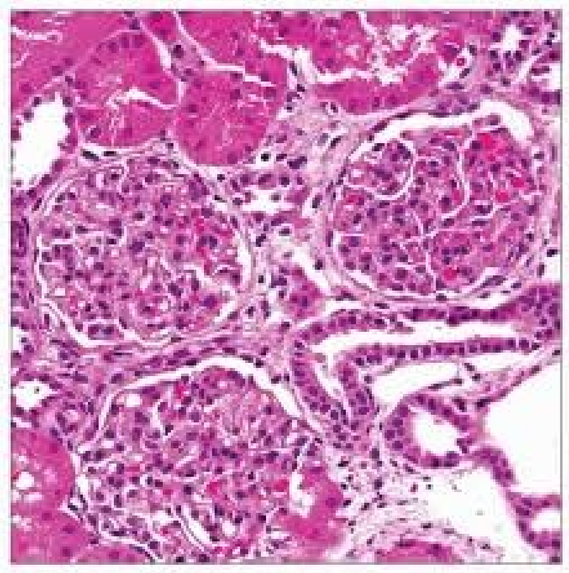
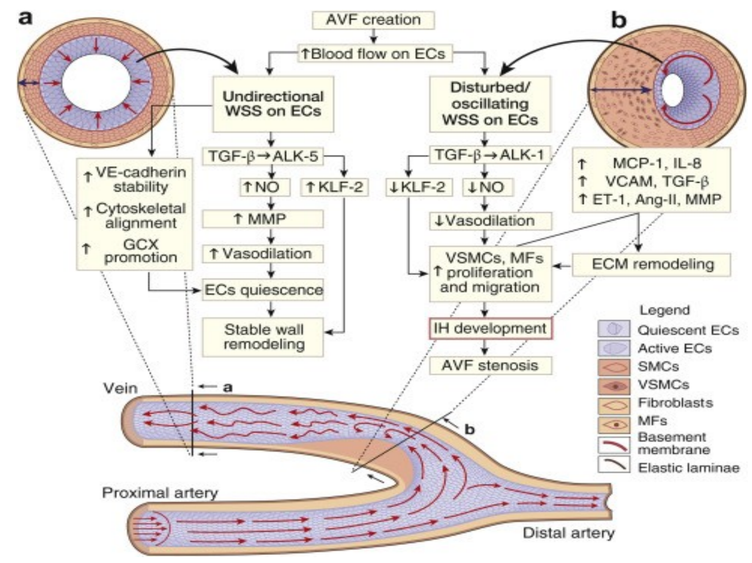
BACKGROUND

The maintenance of adequate vascular access is crucial to patient survival on hemodialysis. Complications related to vascular access account for 20 to 25% of all hospitalizations in dialysis patients. Thrombosis is the leading cause of arteriovenous fistula and graft failure. Thrombophilias are inherited or acquired predispositions to thrombosis and have been suggested as a possible cause of dialysis access thrombosis. Studies to date have been conflicting, with some suggesting a significant association whereas others have not. There are few cases of Alport syndrome as underlying CKD (chronic kidney disease) with diagnosed genetic thrombophilia described in literature.



Picture 1. Kidney biopsy findings in patient with Alpor syndrom



Picture 2. The mechanisms of hemodialysis vascular access failure

CASE REPORT

Male patient, 25 years old, presented with hematuria and non nephrotic proteinuria in childhood, age of two years. At the age of eleven years old he presented with elevated levels of serum creatinine and urea for the first time. Kidney biopsy was performed and Alport syndrome diagnosed. Patient developed severe bilateral hypoacusis also in early childhood. Six years ago the progression of CKD was noticed. He developed the end stage chronic renal failure at the age of 24 years old, when treated with RRT with hemodialysis. Patient had multiple reccurent thrombosis of all AV fistulas created in the next period of three years, and with recurrent thrombosis of central venous dialysis catheters use as vascular accesses while creating and growing AV fistulas. He was treated with anticoagulation therapy all the time, beside the dialysis anticoagulation. By the time he developed thrombosis of all vascular accesses and the treatment with CAPD was started in the age of 25.The hematological evaluation was performed and the antiphospholipid syndrome was proven. Genetical analyses on inherited thrombophilias showed that the presence of homozygosy in C667T polymorphism and heterozygosy in A1298C polymorphism in MTHFR gene. Mutation in PAI-1 4G/5G gene in homozygous status was also proven. Polymorphisms for factor V Leiden, in factor II prothrombin mutation and mutation in factor II genes were not detected. He had thrombosis of last AV fistula, although treated with acenocumarol as anticoagulant therapy with LMWH low molecular weight heparin during hemodialysis. He was switched to peritoneal dialysis because of accesses failure.

CONCLUSION

The presence of thrombophilia is associated with hemodialysis access thrombosis. In patients with Alport syndrome inherited thrombophilias disorders should be diagnosed in every case of first vascular access thrombosis and earlier than in other groups of CKD patients in order to prevent the thrombosis of next hemodialysis vascular accesses with proper anticoagulant therapy.