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O-226 Inherited Antithrombin III Deficiency: A Case Report of Familial Pedigree and Gene Mutation Screening

Case Reports

Rongrong Zhu [*], **Zhanjiang Cao, Weiwei Wu**

Vascular Surgery, Beijng Tsinghua Changgung Hospital, Beijing, China

Introduction - Inherited antithrombin III deficiency is an autosomal dominant genetic disease, which can lead to various forms of thrombus and is associated with pathological pregnancy.
Methods - In this study, we reported a familial pedigree with three daughters diagnosed as AT III deficiency with different clinical manifestations. The first one was diagnosed with deep vein thrombus (DVT) during pregnancy and developed into acute pulmonary embolism (PE) under anticoagulation of low molecular heparin. The second one developed unprovoked DVT in left lower extremity and received the treatment of inferior vena filter, catheter-directed thrombolysis, pharmacomechanical thrombectomy and anticoagulation with rivaroxaban. The last one experienced an abortion because of placental abruption in the second trimester of pregnancy and the pathology indicated thrombosis in the microvessels of the placenta. We screened the whole family members for AT activity and antigen level, including the patients' parents and sister without venous thromboembolism (VTE) history. Whole exome sequencing was performed in the first case to identify variations in the AT gene and Sanger sequencing was further adopted for other family members to verify the gene mutation responsible for the pedigree. We also conducted a long-term follow up of this family to help them with the anticoagulation during pregnancy and daily life.
Results - Four members of the family, including the three sick daughters and their father, showed a deceased AT III activity and antigen level. However, their father did not have any VTE history before. The whole exome sequencing identified three nonsynonymous mutations, including c.1273C>T(p.R425C) mutation in SERPINC1, c.1200A>C(p.E400D) mutation in SERPINA1 and c.494C>T(p.T165M) mutation in F2. Then the Sanger sequencing for other family members verified the first mutation, c.1273C>T(p.R425C) mutation in SERPINC1, was the pathogenic gene for this family because of the consistence of mutation and decreased AT III activity. During the 2-year follow up, the father was attacked by DVT and two of the sick daughters gave birth to a healthy baby respectively with anticoagulation during pregnancy.
Conclusion - Inherited antithrombin III deficiency is a hypercoagulable state associated with an increased risk for venous thrombosis, which cannot be relieved by low molecular heparin except NOACs like rivaroxaban. It is a disease with low incidence, few researches and no guidelines, so when and how to give anticoagulation remain controversial, especially during pregnancy. Further studies are necessary to give clues for clinical practicing of patients with this rare disease.

O-227 Subclavian Pseudoaneurysm in a Young Patient: Rare Cause to Think

Case Reports

Satchithanantham Vinojan [1,*], **Gayan Bandara** [2], **Thushan Gunaratne** [2], **Ranjuka Ubayasiri** [2]

[1] *Department of Surgery, Faculty of medicine, Jaffna, Sri Lanka*
[2] *Teaching Hospital, Karapitiya, Sri Lanka*

Introduction - Tuberculosis is an extremely rare cause for pseudo aneurysm. Infected aneurysm is a diagnostic challenge that is associated with significant morbidity and mortality. Treatment consists of antibiotic therapy ,aggressive surgical debridement and selective vascular reconstruction.
Methods - Case History

A 21 year old boy presented with the history of progressively increasing size of left supraclavicular lump of 3 months duration. It was associate with mild grade of evening pyrexia, loss of appetite and loss of weight. He didn't have a history of blunt trauma or cannulation in the neck. Upon examination there was a pulsatile lump in his left supraclavicular region. His inflammatory markers were high. (CRP and ESR was 70mg/dl, 105mm/1sthr respectively) Mantoux reading was positive. Sputum and blood cultures were negative. CT angiogram revealed a pseudoaneurysm arising from the 2nd part of the left subclavian artery.

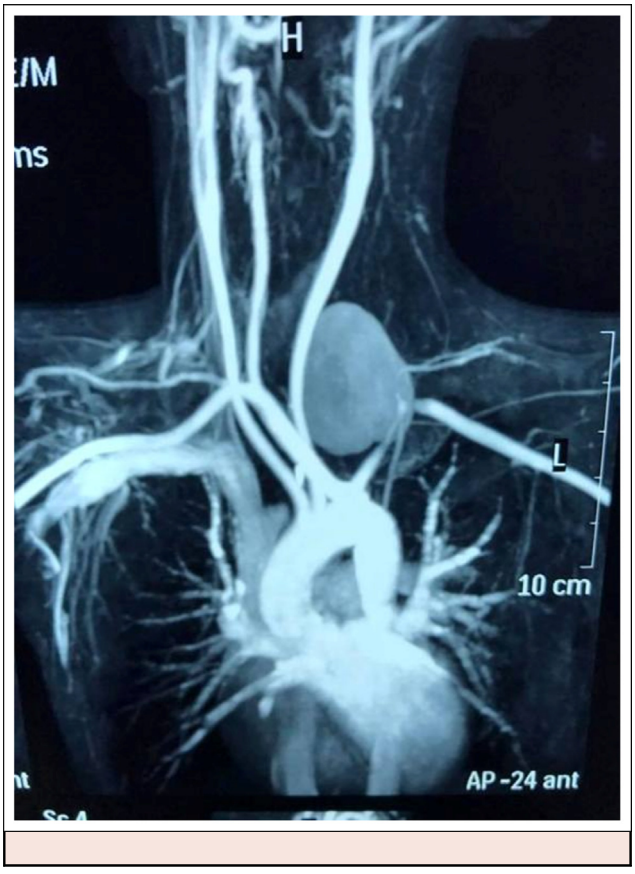
The patient underwent surgery, which revealed a pseudoaneurysm arising from the 2nd part of the subclavian artery. It was excised and thoroughly debrided. Aneurysml part was excluded by ligation and distal perfusion to the left upper limb was achieved with extra anatomical carotid to axillary artery bypass with reverse saphenous vein graft. Gram stains from the resected specimen showed only few neutrophils and there were no bacteria. Probable diagnosis of tuberculous pseudoaneurysm was made and he was started on anti TB medication.
Results - Discussion

Subclavian artery pseudo aneurysms are rare. Pseudo aneurysms in this location usually occurs after trauma. (1) But rarely it happens after infections. Staphylococcus aureus, salmonella and streptococcus species are the commonest organisms causing mycotic aneurysms. (2)

Mycobacterium tuberculosis is the common organism among the mycobacterium species. Even though we didn't have any positive cultures for these organisms our case is most probably due to infection because he had constitutional symptoms associated with raised inflammatory markers and his symptoms diminished following a resection and anti TB treatment. We have started him on anti TB treatment based on his symptoms, high inflammatory markers and positive mantoux test. Inaddition tuberculosis is quite common in south Asian countries. There are several other case reports of tuberculosis arterial infections have been reported without positive microbiology results. (3)

Seeding of mycobacterium in the vessel wall is explained by several theories. It can be due to haematogenous spread, inoculation from vasovasorum or direct local spread.(4)

Treatment of mycotic aneurysms can be endovascular or open surgical repair. Endovascular treatment is less invasive one and can be done in patients who are at high risk for open surgical intervention. As this aneurysm harbors infection some authors don't prefer stent placement in addition stent has its own complications like thrombosis, migration , rupture and endoleaks. (1) As our patient was young and fit for surgery and the aneurysm also was in the second part of the subclavian artery we offered him open surgical repair. Direct reconstruction in this case after exclusion is difficult because of short segment of the 1st part of the subclavian artery so we did extra anatomical bypass.



Conclusion - Tuberculosis mycotic aneurysms are very rare. Even though endovascular treatment options are emerging open surgical repair remains as a standard option in patients who are fit for surgery.

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O-228 Hybrid Approach to Symptomatic Innominate Artery Stenosis with Direct Embolic Protection

Case Reports

Nelson Camacho [1,*], Frederico B. Gonçalves [2], Gonçalo Alves [1], Rita S. Ferreira [1], Joana Catarino [1], Ricardo Correia [1], Rita Bento [1], Maria E. Ferreira [1]

[1] *Angiology and Vascular Surgery, Hospital de Santa Marta, Lisbon, Portugal*
[2] *Centro Hospitalar Lisboa Central, Hospital de Santa Marta, Lisbon, Portugal*

Introduction - Innominate artery (IA) lesions are an uncommon but important source of symptomatic extracranial cerebrovascular disease and may be associated with significant morbidity. For symptomatic lesions, open surgery or endovascular techniques are valuable options. While endovascular repair offers a minimally invasive alternative to extra-anatomic bypass or arch debranching techniques, cerebral embolic complications remain an important concern, especially in highly calcified or more extensive lesions.

Methods - The authors present a retrospective analysis of 5 patients with symptomatic stenosis of the IA treated with a hybrid operation. We evaluate short and medium term results of the procedure on these patients. The primary endpoints were symptoms after the surgery and restenosis on follow-up.

Results - All five patients were symptomatic and clinical presentation included dizziness (n=2), vertigo (n=1), gait ataxia (n=1), transient ischemic attack with hemiparesis (n=2) and right upper limb claudication (n=1). Mean age was 66±8 years (54-75 years) with male gender predominance (n=4). Radiological features of the lesions at the IA included ulcerated plaque (n=1), pre-occlusive stenosis with or without mural thrombus (n=4) and occlusion of the right subclavian artery (SCA) (n=1).

All patients were submitted to a hybrid operation under general anesthesia with surgical exposure of the right