Abstracts

Introduction

Catastrophic Antiphospholipid Syndrome (CAPS) represents the most severe manifestation of Antiphospholipid Syndrome (APS), affecting approximately 1% of cases, and is associated with a high mortality rate. It is characterized by multorgan failure and widespread microvascular thrombosis but mainly without vasculitis. This case report aims to contribute to the limited literature available on CAPS cases co-occurring with vasculitis.

Case Report

A 36-year-old male diagnosed with primary APS in 2003 due to two deep vein thrombosis episodes and testing positive for anti-beta 2 glycoprotein IgG antibodies and lupus anticoagulant. Systemic lupus erythematosus was ruled out, and the patient was treated with warfarin. Amid the COVID-19 pandemic, he lost regular follow-up and discontinued oral anticoagulation for a year.

In 2021, he developed an upper airway infection, tested negative for COVID-19, and exhibited focal neurological symptoms with global aphasia. Within four days, necrosis affected his 4th toe on the right foot and the 2nd and 3rd toes on the left foot. Three days later, he presented with proteinuria (1g/24hrs), a threefold increase in creatinine levels from baseline, and severe thrombocytopenia. A skin biopsy of the right foot dorsum (figure 1) prompted the initiation of treatment: methylprednisolone, intravenous immunoglobulin, and anticoagulation. During follow-up the patient developed pneumonia, sepsis and ultimately, death.

Abstract P1 Table 1

<table>
<thead>
<tr>
<th>Characteristics</th>
<th>All patients (n=258)</th>
<th>Intra-renal involvement (n=17)</th>
<th>No intra-renal involvement (n=241)</th>
<th>p value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Male sex</td>
<td>56/258 (21.7)</td>
<td>6/17 (35.3)</td>
<td>50/241 (20.7)</td>
<td>0.218</td>
</tr>
<tr>
<td>Caucasians</td>
<td>244/258</td>
<td>15/17 (88.2)</td>
<td>229/241 (95.0)</td>
<td>0.233</td>
</tr>
<tr>
<td>Age at disease onset, years</td>
<td>32.0 (25.0–44.0)</td>
<td>29.0 (24.0–38.0)</td>
<td>33.0 (25.0–44.0)</td>
<td>0.377</td>
</tr>
<tr>
<td>Thrombotic APS</td>
<td>173/258</td>
<td>6/17 (35.3)</td>
<td>167/241 (69.3)</td>
<td>0.004</td>
</tr>
<tr>
<td>Obstetric APS</td>
<td>109/252</td>
<td>5/11 (45.5)</td>
<td>104/191 (54.5)</td>
<td>0.561</td>
</tr>
<tr>
<td>Catastrophic APS</td>
<td>4/258 (1.6)</td>
<td>1/17 (7.6)</td>
<td>1/241 (0.4)</td>
<td>0.001</td>
</tr>
<tr>
<td>‘Extra-criteria’ APS*</td>
<td>19/258 (7.4)</td>
<td>8/17 (47.1)</td>
<td>11/241 (4.6)</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>Thrombocytopenia</td>
<td>36/202 (17.8)</td>
<td>2/11 (18.2)</td>
<td>34/191 (17.8)</td>
<td>1.000</td>
</tr>
<tr>
<td>Heart valve lesions</td>
<td>21/258 (8.1)</td>
<td>2/17 (11.8)</td>
<td>19/241 (7.9)</td>
<td>0.637</td>
</tr>
<tr>
<td>Livedo reticularis</td>
<td>22/258 (8.5)</td>
<td>1/17 (5.9)</td>
<td>21/241 (8.7)</td>
<td>1.000</td>
</tr>
<tr>
<td>Epilepsy</td>
<td>17/258 (6.6)</td>
<td>4/17 (23.5)</td>
<td>13/241 (5.4)</td>
<td>0.018</td>
</tr>
<tr>
<td>Single aPL profile</td>
<td>87/250 (34.8)</td>
<td>2/15 (13.3)</td>
<td>85/241 (35.6)</td>
<td>0.072</td>
</tr>
<tr>
<td>Double aPL profile</td>
<td>59/250 (23.6)</td>
<td>4/15 (26.7)</td>
<td>56/235 (23.8)</td>
<td>0.761</td>
</tr>
<tr>
<td>Triple aPL profile</td>
<td>103/250</td>
<td>9/15 (60.0)</td>
<td>94/235 (40.0)</td>
<td>0.127</td>
</tr>
</tbody>
</table>

Conclusions

The present study highlights the importance of conducting appropriate renal assessment and, when necessary, renal biopsy in PAPS patients, even in the presence of mild urinary alterations. On the other hand, from the nephrologists’ point of view, it may be relevant to consider the routine research of aPL at time of glomerulopathies evaluation, in particular because of the aPL prognostic role in the development of subsequent related events.
Discussion and Conclusion Secondary events in APS are often triggered by precipitating factors, which can be identified in approximately 55% of patients. In this case, infection and the discontinuation of anticoagulation therapy were identified as contributing factors. A noteworthy characteristic of this patient's case is the co-occurrence of CAPS and vasculitis, which is likely associated with multiorgan failure and a higher mortality rate. The scarcity of reported cases combining these phenomena may be attributed to the infrequency of biopsies being performed due to the severity of such patients.

Clinicians should be aware of this entity, given its challenging differential diagnosis with other thrombotic entities, including COVID-19. Timely identification is imperative, as it necessitates urgent intervention as a potential life-saving measure. Despite prompt diagnosis and treatment, this disease continues to exhibit a higher mortality rate compared to other manifestations of APS.

Abstract P2 Figure 1

Discussion and Conclusion Secondary events in APS are often triggered by precipitating factors, which can be identified in approximately 55% of patients. In this case, infection and the discontinuation of anticoagulation therapy were identified as contributing factors. A noteworthy characteristic of this patient’s case is the co-occurrence of CAPS and vasculitis, which is likely associated with multiorgan failure and a higher mortality rate. The scarcity of reported cases combining these phenomena may be attributed to the infrequency of biopsies being performed due to the severity of such patients.

Clinicians should be aware of this entity, given its challenging differential diagnosis with other thrombotic entities, including COVID-19. Timely identification is imperative, as it necessitates urgent intervention as a potential life-saving measure. Despite prompt diagnosis and treatment, this disease continues to exhibit a higher mortality rate compared to other manifestations of APS.

Abstract P3

Background Pediatric Antiphospholipid Syndrome (ped APS) is the most common acquired condition of increased blood clotting in developmental age.

Objectives This case report describes a 13-year-old patient with primary ped APS.

Methods The patient presented with a history of transient visual disturbance in 2019, exhibiting a limited visual field in the left eye. MR brain imaging and EEG showed no abnormalities.

In 2021, the patient experienced headaches and swelling of the face and neck. Physical examination revealed dilated collateral circulation vessels on the chest.

Angio CT of the head, neck and chest revealed extensive thrombosis of the neck vessels involving both internal, external, subclavian, brachiocephalic veins up to superior vena cava. On the right side, the thrombus extends higher, visible in the lumen of the sigmoid sinus. On the left side, the lesion started in the extracranial segment of the internal jugular vein with extensive collateral circulation.

Abdominal ultrasound excluded thrombosis while ECHO yielded normal results.

Laboratory tests ESR, CRP with normal limits, PLT 108 thousand/mm³. Coagulation system tests revealed elevated D-dimers, APTT, INR.

Tests for thrombophilia showed no evidence of Leiden mutation of factor V gene, but the abnormal result was obtained for the presence of variant F2:c."97G>A in a heterozygous pattern mutation G20210A.

Additionally, ANA - 1: 640, high levels of anti-cardiolipin antibodies (aCL) IgM, IgG, anti-B2GPI IgM, LAC were detected, anti - dsDNA, anti-Sm were not detected. Component components C3, C4 were with normal limits, BTA test results were negative.

Results According to Revised APS Criteria (2006) and ACR/EULAR APS Criteria - ped APS was diagnosed. The patient did not meet the criteria of SLE or thrombosis associated thrombophilia.

Treatment with low-molecular-weight heparin in a therapeutic dose was initiated, followed by the use of warfarin.

Follow-up tests after 2 years revealed low titer of anti-cardiolipin antibodies IgM, IgG, positive LAC, ANA 1:320.INR 3.6, persistence of anti-B2GPI IgM antibodies.

Conclusion In the diagnosis, consideration was given to SLE-like syndrome, but the significant improvement with only anticoagulants speaks in favor of APS. Further follow-up tests are necessary for SLE due to the presence of ANA.