G20210A homozygosity in antiphospholipid syndrome secondary to systemic lupus erythematosus

We report the first case of systemic lupus erythematosus (SLE)-associated antiphospholipid syndrome in a young female homozygous for the G20210A allele in the prothrombin gene who developed an extensive venous thrombosis while taking oral contraceptives.

Sir,
The risk of deep venous thrombosis (DVT) is increased by conditions that cause hypercoagulability or venous stasis.1 A variant of prothrombin (G20210A) represents the second most common genetic risk factor in Caucasians, after factor V Leiden.2 3 The mechanism of thrombosis is probably related to the high amounts of thrombin generated.2

We report a case of a 28-year-old woman who developed an extensive DVT after having taken oral contraceptives for one year. Venous ultrasonography demonstrated a femoral-iliac thrombosis with proximal extension to the common iliac vein. Past history was positive for oral ulcers and Raynaud’s phenomenon, since she was a teenager. She reported photosensitivity lasting years, with an important episode on the scalp some months earlier: scarring lesions with atrophy and alopecia were still evident. Platelet count, the scalp some months earlier: scarring lesions with atrophy and alopecia were still evident. Platelet count, red blood cell count, white cell count, platelet count, and erythrocyte sedimentation rate and immunoglobulin M were moderately increased and white cell count and erythrocyte sedimentation rate and immunoglobulin M were moderately increased and white cell count were normal. The partial thromboplastin and prothrombin time, antithrombin III and fibrinogen were normal. The patient was treated with continuous intravenous nonfractionated heparin infusions followed by oral warfarin for 7 months (INR = 3.0). During the follow up period, she had a history of thrombosis while taking oral contraceptives. The relative risk in women with factor V Leiden using contraceptives is 34.7, that of carriers of prothrombin variant is unknown.1

Key words
Thrombophilia, prothrombin variant, antiphospholipid syndrome, SLE

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References

Respiratory burst activity in late pregnancy in a carrier of X-linked chronic granulomatous disease

Screening for chronic granulomatous disease (CGD) and carrier status is carried out by the nitroblue tetrazolium test. The diagnosis is confirmed by quantitative tests of respiratory burst activity. Production of reactive oxygen intermediates may be increased in late pregnancy which may compensate for the otherwise low level in a CGD carrier, thus confounding diagnosis for the unwary clinician.

Table 1. NBT slide test. Percentage of NBT non-responding cells.

<table>
<thead>
<tr>
<th>Subjects</th>
<th>Healthy donors</th>
<th>CGD-affected subjects during pregnancy</th>
<th>CGD carrier 5 months after pregnancy</th>
</tr>
</thead>
<tbody>
<tr>
<td>% NBT non-responding cells</td>
<td>0-2</td>
<td>98-100</td>
<td>70-67</td>
</tr>
</tbody>
</table>

Figure 1. Evaluation of oxidative burst activity performed by flow cytometric assay (DCF fluorescence, and SOD inhibitable Cytochrome c reduction).