Eosinophilic Fasciitis in Siblings

To the Editor:

Eosinophilic fasciitis (EF) is an uncommon disorder with no documented prevalence and with unknown etiology. We describe a case of siblings with EF and identical HLA types, suggesting a genetic predisposition. The reporting of unique cases such as these is important in furthering our knowledge of rare conditions.

Patient 1 presented at 14 years of age with a 3-month history of upper limb joint stiffness and swelling after prolonged physical exertion with associated general malaise and weight loss. On examination she had skin thickening with digital sparing and venous guttering predominantly of her upper limbs. Decreased range of movement of her wrists and metacarpophalangeal joints was noted. Total white blood cell count (WCC) was normal with an eosinophilia of 1.38 × 10^9/l (normal 0.04–0.44 × 10^9/l), an elevated serum immunoglobulin G (IgG) of 29.4 g/l (normal 5–13 g/l), and erythrocyte sedimentation rate (ESR) 42 mm/h.

Cutaneous manifestations are generally the primary symptoms of EF and include painful swelling, edema, and induration; while any part of the body may be affected, limb involvement is most common, with characteristic sparing of the hands. The grooves seen are because of sparing of the epidermis and superficial dermis, with relative immobility of the connective tissue around deep veins. Joint contractures, inflammatory arthritis, and muscle weakness have been reported.

While both ultrasound and MRI are useful, the diagnosis of EF is primarily clinical; an early and often transient peripheral eosinophilia is present in 63% of patients, elevated ESR in 29%, and hypergammaglobulinemia in 35%.

An inflammatory infiltrate of eosinophils, mast cells, histiocytes, and lymphocytes is classically seen on full-thickness biopsy. Subsequent magnetic resonance imaging (MRI) was normal; however, treatment had been ongoing for several months at the time of imaging.

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Table 1. HLA typing of eosinophilic fasciitis in familial cases.

<table>
<thead>
<tr>
<th>Sibling pair</th>
<th>HLA-A</th>
<th>HLA-B</th>
<th>HLA-C</th>
<th>HLA-DR</th>
</tr>
</thead>
<tbody>
<tr>
<td>Case 1</td>
<td>A2</td>
<td>B7/27</td>
<td>Cw 7/1</td>
<td>DRB1-0103, DRB3-01</td>
</tr>
<tr>
<td>Case 2</td>
<td>A2</td>
<td>B7/27</td>
<td>Cw 7/1</td>
<td>DRB1-0103, DRB3-01</td>
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<tr>
<td>Thomson 1989^9</td>
<td></td>
<td></td>
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</tr>
<tr>
<td>Sibling 1</td>
<td>A2/A11</td>
<td>B7/B35</td>
<td>Cw4</td>
<td>DR2/3</td>
</tr>
<tr>
<td>Sibling 2</td>
<td>A2/A11</td>
<td>B7/B35</td>
<td>Cw4</td>
<td>DR2/3</td>
</tr>
</tbody>
</table>

EF remains poorly understood. This report suggests that family members with similar HLA typing are at increased risk, with a possible link to HLA-A2. It is likely that a second environmental factor contributes to the timing of disease onset. The low disease prevalence makes it difficult to do the case-control studies needed to confirm a correlation with HLA.

CATHERINE SULLIVAN, PHD, MB, BCh; ROBERT COUGHLAN, MB, BCh, Department of Rheumatology, Galway University Hospital, Galway, Ireland. Address correspondence to Dr. Sullivan, 13 Larkfield Way, Lucan, County Dublin, Ireland. E-mail: sullivancat@yahoo.com

REFERENCES