Eosinophilic Fasciitis in Siblings

CATHERINE SULLIVAN and ROBERT COUGHLAN

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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 age 14 years 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. C-reactive protein (CRP) and creatinine kinase (CK) levels were within normal limits. A biopsy demonstrated a diffuse inflammatory cell infiltrate in the fascia, with eosinophils, lymphocytes, and plasma cells. She responded well to prednisolone and is maintained on oral methotrexate.

Patient 2, the younger brother and only sibling of Patient 1, also presented at age 14 years with a 3-week history of skin thickening on his upper limbs, with venous guttering and digital sparing. Total WCC and eosinophils were within normal limits, as were CRP, ESR, and CK levels. Serum IgG was elevated at 22.8 g/l. A clinical diagnosis of EF was made and he responded to oral prednisolone and methotrexate. There was a nonspecific inflammatory infiltrate on full-thickness biopsy. Subsequent magnetic resonance imaging (MRI) was normal; however, treatment had been ongoing for several months at the time of imaging.

Cutaneous manifestations are generally the primary symptoms of EF and include painful swelling, edema, and induration; while any part 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 primarily clinical; an early and often transient peripheral eosinophilia is generally seen 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.

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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>
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<tbody>
<tr>
<td>Case 1</td>
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<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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<td>Thomson 1989⁹</td>
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<td>B7/B35</td>
<td>Cw4</td>
<td>DR2/3</td>
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<tr>
<td>Sibling 1</td>
<td>A2/11</td>
<td>B7/B35</td>
<td>Cw4</td>
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