Familial Dercum’s Disease (Adiposis Dolorosa)

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Dercum’s disease consists of multiple, painful lipomata seen mostly in postmenopausal, obese women. Its exact aetiology is not known. A family with six members in two generations affected with Dercum’s disease is presented supporting the hereditary aetiology of this condition.

Dercum’s disease (adiposis dolorosa) is characterized by multiple painful lipomata, most commonly seen in obese, postmenopausal women, classically associated with asthenia and emotional disturbances.¹ It may appear at any site, mostly affecting the trunk, shoulders, arms, thighs, forearms and legs. The face and head are usually spared.

The exact aetiology of the disease is unknown, and no studies have documented its incidence. Several hypotheses suggested a possible endocrine aetiology,²⁻³ but now there is some evidence that Dercum’s disease is a hereditary disorder, transmitted as an autosomal dominant with incomplete penetrance.⁴⁻⁻⁷

This report describes Dercum’s disease in six members of a family in two generations supporting the hereditary aetiology of this condition.

Case Report

A 38-year-old male presented to the hospital complaining of multiple subcutaneous swellings since the age of 15 years. The swellings appeared in both upper limbs simultaneously and then in other parts of the body. The swellings increased gradually in size and were not painful to start with, but then he started to complain of pain intermittently on pressure. There was no history of other problems.

Physical examination revealed a healthy young man (height 184 cm, weight 79 kg). There were multiple, well circumscribed subcutaneous masses involving upper limbs (arms and forearms), thighs and abdominal wall. Those in the upper limbs were tender on pressure. They varied from 0.5 to 4 cm in diameter. The rest of the physical examination was normal and his investigations (CBC, electrolytes, urine analysis, liver function tests, blood cholesterol and thyroid function tests) were normal. The largest of the swellings which was in the right thigh was excised and sent for histopathology; it revealed a simple lipoma without evidence of inflammation or infarction. Chromosome banding studies were not available.

The family history was obtained from the patient who was a physician and who had examined his relatives; it revealed five other members of his family to be affected by the disease as shown in the constructed pedigree. The dates of births of the family members were not obtained, only the ages as recalled by the proband, including the age at death of two siblings. In those affected the lipomata appeared after puberty and were small when they first appeared but increased gradually in size and number (Table 1).

Discussion

The name adiposis dolorosa was proposed by Dercum in 1882 when he described three cases of fatty swellings with pain, asthenia and emotional disturbance.¹

Dercum’s disease is characterized by the presence of painful, multiple fatty tumours seen
Table 1

<table>
<thead>
<tr>
<th>Patient no.</th>
<th>Age when examined by the proband (years)</th>
<th>Age at onset (years)</th>
<th>Personally examined by the authors</th>
<th>Symptoms and signs</th>
<th>Histology findings</th>
</tr>
</thead>
<tbody>
<tr>
<td>I 2</td>
<td>76</td>
<td>20</td>
<td>—</td>
<td>Generalized lipomata affecting mostly upper limbs</td>
<td>—</td>
</tr>
<tr>
<td>I 2</td>
<td>79</td>
<td>Not affected</td>
<td>17</td>
<td>Generalized lipomata affecting mostly upper limbs, obesity and emotional disturbances</td>
<td>—</td>
</tr>
<tr>
<td>II 1</td>
<td>65</td>
<td>—</td>
<td>—</td>
<td>—</td>
<td>—</td>
</tr>
<tr>
<td>II 2</td>
<td>52</td>
<td>Not affected</td>
<td>18</td>
<td>Generalized lipomata affecting mostly upper limbs</td>
<td>—</td>
</tr>
<tr>
<td>II 3</td>
<td>48</td>
<td>—</td>
<td>—</td>
<td>Generalized lipomata affecting mostly upper limbs</td>
<td>—</td>
</tr>
<tr>
<td>II 4</td>
<td>44</td>
<td>—</td>
<td>—</td>
<td>Generalized lipomata affecting mostly upper limbs and asthenia</td>
<td>—</td>
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<tr>
<td>II 5</td>
<td>45</td>
<td>—</td>
<td>—</td>
<td>Generalized lipomata affecting mostly lower limbs</td>
<td>—</td>
</tr>
<tr>
<td>II 6</td>
<td>38</td>
<td>—</td>
<td>Yes</td>
<td>Generalized lipomata affecting mostly upper limbs</td>
<td>Simple lipomata</td>
</tr>
<tr>
<td>II 7</td>
<td>35</td>
<td>—</td>
<td>—</td>
<td>—</td>
<td>—</td>
</tr>
</tbody>
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*a Died at the age of 76 and 48 years.

Figure 1. Pedigree of the family showing six members affected in two generations. The proband is indicated by the arrow.

mostly in postmenopausal women. The distribution of these lipomata is variable and have been described affecting any part of the body except face and head. Although the classic description of Dercum's disease is that of painful lipomata, there are several cases described in which the lipomata remained painless. This probably is a variant of its clinical presentation. The pain may vary from tenderness on pressure to violent attacks of spontaneous pain, and the exact cause of pain in these lesions is not known. It has been suggested that the pain is due to pressure to nearby peripheral nerves by the enlarging lipoma, but this has not been confirmed on pathological examination. A condition that closely resembles Dercum's disease is multiple lipomatosis, in which the lipomata are asymptomatic and numerous, affecting the forearms and thighs predominantly, showing a symmetrical distribution. This condition occurs predominantly in males and appears in the third decade of life. Rarely multiple lipomatosis is familial inherited as autosomal dominant and its separation from Dercum's disease is arbitrary.

The lipomatous lesions of Dercum's disease have been classified into three types. These are: (1) the circumscribed diffuse type, the most common; (2) the generalized diffuse type, the next most common; (3) the nodular type, the least common. Our patient belonged to the third, least common type.

The aetiology of the disease is still not known. As a result of its occurrence mainly in postmenopausal females, the possibility of a specific endocrine disorder of aetiological importance was considered. There have been four previous reports of families with adiposis dolorosa suggesting that Dercum's disease is a hereditary condition transmitted as an autosomal dominant trait with incomplete penetrance. This report describes the fifth family with adiposis dolorosa supporting the hereditary aetiology of this condition. The variations in its clinical features suggest variations in the expression of the gene.

Its main occurrence in females (a female to male ratio of 5:1) and the high incidence of obesity in affected females suggest a possible endocrine factor which is sex-determined that enhances the expression of the gene and could account for the apparent sex disparity as males are frequently mildly affected by the disease compared with females.
References


