A rare case of Langerhans cell histiocytosis of the skull in an adult: a systematic review

Corinna Chiong,1 Shruti Jayachandra,1 Guy D. Eslick,1 Darweesh Al-Khawaja,2 Vidhyasagar Casikar2
1Whiteley-Martin Research Centre, Discipline of Surgery, University of Sydney, Nepean Hospital, Sydney Medical School, Nepean, Penrith, New South Wales; 2Department of Neurosurgery, Nepean Hospital, Penrith, New South Wales, Australia

Abstract

We report a 41-year old male who presented to the Emergency Department after falling while water-skiing. He had a previous medical history included chronic headaches, which had persisted for the last 2-3 months prior to presentation. Computed tomography of the head showed a small hypersensitivity with a small extra axial collection with a maximum thickness of 1mm. Differential diagnoses included an arachnoid cyst, haemangioma, meningioma or a secondary lesion. A diagnosis of Langerhans Histiocytosis was made based on the histopathology examination and the immunoperoxidase staining.

Introduction

Langerhans cell histiocytosis (LCH) is a rare condition that usually affects the pediatric population. The condition can affect almost any organ in the body. It is a rare condition with an estimated annual incidence between 1-7 cases per million of the population.1 More than 50% of patients are less than two-years of age have disseminated LCH with organ dysfunction and die of the disease.2 The prevalence of LCH seems to be higher among whites than other races. The incidence of LCH is greater in males than in females, with a male-to-female ratio of 2:1. Recent early data from the U.K. suggest that just as many cases of LCH present in adult life as in childhood.1 However, it should be noted that at present it remains unclear if these adult cases are de novo or if they are occult cases from childhood.2 We report a case of adult LCH with his presentation, medical history, clinical characteristics, and radiological findings as well as the success of the surgical intervention.

Case Report

A 41-year old male who presented to the Emergency Department after falling while water-skiing at 40-50 km/hr. He landed face first onto the water but did not lose consciousness and remembered that he was in trouble.
Langerhans cell histiocytosis is a clonal proliferative disorder of the antigen-presenting cell lineage, histiocytes. It is characterized by the accumulation of Langerhans cells, which are dendritic cells derived from the bone marrow. These cells are involved in the immune response and play a role in the regulation of the inflammatory response. LCH affects patients under 10 years of age, however, this case represents an uncommon presentation of LCH, occurring in an adult population.

The patient was a 57-year-old female with a history of diabetes insipidus and hypertension. She presented with a sudden onset of headache and swelling of the right side of her head. On examination, she had a raised intracranial pressure and reduced visual acuity. Imaging studies revealed a lesion in the right parietal bone, with a characteristic imaging pattern suggestive of LCH. Despite initial improvements with medical therapy, the lesion recurred and surgical intervention was required. The patient underwent excision of the bone lesion and post-operative chemotherapy with VLB, PSL, and mercaptopurine. The patient has been disease-free for two years post-treatment.

<table>
<thead>
<tr>
<th>Study (ref)</th>
<th>Age/sex</th>
<th>Symptoms and signs</th>
<th>Investigations</th>
<th>Lesions</th>
<th>Histological findings</th>
<th>Management</th>
<th>Outcome</th>
</tr>
</thead>
<tbody>
<tr>
<td>Suzuki et al., 2008</td>
<td>47/F</td>
<td>Hip pain, diabetes insipidus, forehead bone defect</td>
<td>Skull XR, whole body CT, bone scintigram scan</td>
<td>Multifocal</td>
<td>Accumulation of Langerhans cells expressing CD1a and S100 antigens, with scattered eosinophils and lymphocytes</td>
<td>Chemotherapy: 6-weeks of PSL + VLB, then 6-mercaptopurine, PSL + VLB for 1 year</td>
<td>Clinical improvement, traced for over 12 months</td>
</tr>
<tr>
<td>Fung et al., 2002</td>
<td>29/F</td>
<td>Progressive non-tender left frontal head swelling over 2 months</td>
<td>CT head</td>
<td>Unifocal</td>
<td>Round cells expressing CD1a and S100 admixed with osteoclast-like giant cells expressing CD68, Birbeck granules and inflammatory cells</td>
<td>Craniotomy and resection of lesion</td>
<td></td>
</tr>
<tr>
<td>Suzuki et al., 2010</td>
<td>56/F</td>
<td>Sudden sensorineural hearing loss and vertigo</td>
<td>CT head, MRI head</td>
<td>Unifocal</td>
<td>Histiocytic cells expressing CD1a and S100 with eosinophilic cytoplasm, inflammatory and giant cells</td>
<td>Excision of lesion and post-operative chemotherapy with VLB, PSL, 6-mercaptopurine</td>
<td>Disease-free 2 years post-surgery although hearing not fully recovered</td>
</tr>
<tr>
<td>Kobayashi et al., 2007</td>
<td>25M</td>
<td>Raised intracranial pressure and reduced visual acuity</td>
<td>CT head, MRI head, squash preparation from lesion specimens</td>
<td>Unifocal</td>
<td>Admixture of small, mature lymphocytes, eosinophils and Langerhans cells expressing CD1a &amp; S100</td>
<td>Surgical resection</td>
<td></td>
</tr>
<tr>
<td>Murayama et al., 1988</td>
<td>39M</td>
<td>Tenderness in right posterior parietal region</td>
<td>CT head and T2-weighted MRI head</td>
<td>Unifocal</td>
<td>Characteristics Langerhans cells with prominent nuclear groove exhibiting positive immunochemical staining to S-100 protein and CD1a antibody</td>
<td>Local excision and chemotherapy with methotrexate and azathioprine</td>
<td>Improvement reported at 9 months post-treatment initiation</td>
</tr>
<tr>
<td>Makras et al., 2004</td>
<td>57/F</td>
<td>Right scalp pain, nodular lesion in right parietal bone, diabetes insipidus 3 months post commencement of treatment</td>
<td>MRI head</td>
<td>Multifocal</td>
<td>Characteristic Langerhans cells with prominent nuclear groove exhibiting positive immunochemical staining to S-100 protein and CD1a antibody</td>
<td>Local excision and chemotherapy with methotrexate and azathioprine</td>
<td></td>
</tr>
</tbody>
</table>

CT, computed tomography; MRI, magnetic resonance imaging; PSL, prednisolone; VLB, vinblastine.
in adults. Osteolytic bone lesions are a common manifestation of single system LCH in adults. These lesions tend to be unifocal rather than multifocal, often involving the skull or axial skeleton, such as in our patient. Our review of skull vault lesions in adults (Table 1) has confirmed this observation with 4 out of 6 cases of LCH involving the skull having unifocal lesions. Calvarial lesions are normally found incidentally as was the case with our patient. However they may also present with bone pain, soft-tissue swelling, hearing loss, vertigo and visual disturbances. Histologically, proliferation of Langerhans cells expressing CD1a and S100 admixed with acute and chronic inflammatory cells are consistently reported. Immunohistochemical findings in our patient were consistent with these features. This report is interesting and adds valuable information to limited literature available on unifocal skull vault lesions in adults with LCH. Local treatment with excision, systemic chemotherapy and corticosteroid injection is highly successful in treating this disease and patients have excellent prognosis. Over 90% of patients survive 3 to 5 years post diagnosis. Age at diagnosis and initial response to therapy affect the prognosis and rate of recurrence of disease.

**References**