Isolated visual symptoms at onset in sporadic Creutzfeldt-Jakob disease: the clinical phenotype of the ‘‘Heidenhain variant’’

S A Cooper, K L Murray, C A Heath, R G Will, R S G Knight

**Background:** The Heidenhain variant of sporadic Creutzfeldt-Jakob disease (sCJD) is commonly understood to represent cases with early, prominent visual complaints. The term is clarified to represent those who present with isolated visual symptoms. This group may pose diagnostic difficulties and often present to ophthalmologists where they may undergo needless invasive procedures.

**Method:** A retrospective review of 594 pathologically proved sCJD cases referred to the UK National CJD Surveillance Unit over a 15 year period to identify Heidenhain cases.

**Results:** Twenty-two cases had isolated visual symptoms at onset with a mean illness duration of 4 months. The mean age at disease onset was 67 years. Most displayed myoclonus, pyramidal signs, and a delay in the onset of dementia for some weeks. 17 (77%) were referred initially to ophthalmology. Two underwent cataract extraction before diagnosis. All tested cases were homozygous for methionine at codon 129 of the prion protein gene.

**Conclusions:** This rare, but clinically distinct, group of patients with sCJD may cause diagnostic difficulties. Because ocular intervention carries with it the risk of onward transmission awareness of this condition among ophthalmologists is important.
and extrapyramidal signs in one (5%). None had documented seizures. A rapidly progressive dementia was observed in all after the initial period of cognitive preservation which lasted from 2–6 weeks.

Case 1
A 73 year old man complained of difficulty reading, with blank spaces appearing in words. He also complained of colours appearing abnormally enhanced. He was assessed by an ophthalmologist when there was normal visual acuity but dense scotomata lying to the right of fixation bilaterally. A provisional diagnosis of an occipital infarct was made. Six weeks after onset he developed myoclonus, followed by ataxia and ultimately dementia. His vision deteriorated with oculomotor apraxia and cortical blindness. He died 3 months after disease onset.

Case 2
A 62 year old woman presented with deteriorating visual acuity. She felt that her vision was “fogging up” and complained of tunnel vision. She attended an optician but no abnormality was identified. A week later she complained that everything appeared green. An MRI brain scan was ordered following referral to the ophthalmology department but no diagnosis made. Over the next month her gait became unsteady and she was increasingly forgetful. By the time she developed myoclonus she could only perceive light. She died in an akinetic and mute state 4 months after onset.

Investigation results
Twenty patients had at least one EEG. These were considered typical for sCJD after review at the NCJDSU in seven cases (35%). CSF 14-3-3 was analysed in five patients (positive in 35%). CSF 14-3-3 was analysed in five patients (positive in 35%).

Table 1  Visual symptoms at onset (n = 22)

<table>
<thead>
<tr>
<th>Visual symptom</th>
<th>Number of patients (n = 22)*</th>
</tr>
</thead>
<tbody>
<tr>
<td>Decreased visual acuity</td>
<td>8</td>
</tr>
<tr>
<td>Blurred vision</td>
<td>6</td>
</tr>
<tr>
<td>Peripheral visual field defect</td>
<td>2</td>
</tr>
<tr>
<td>Visual distortions</td>
<td>3</td>
</tr>
<tr>
<td>Impaired colour vision</td>
<td>2</td>
</tr>
<tr>
<td>Palinopsia</td>
<td>1</td>
</tr>
<tr>
<td>Tunnel vision</td>
<td>1</td>
</tr>
</tbody>
</table>

*One patient experienced both impaired colour vision and visual distortions at onset.

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