The Presenilin 1 P264L Mutation Presenting as non-Fluent/Agrammatic Primary Progressive Aphasia

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Supplementary Table 1
Previously reported clinical phenotypes of the presenilin 1 P264L mutation. *Detailed clinical phenotype lacking but patients said to meet NINDS-ADRSA consensus criteria for a clinical diagnosis of Alzheimer’s disease; -, absent or not described; AAO, average age of onset; FTLD, frontotemporal lobar degeneration; PPA, primary progressive aphasia

<table>
<thead>
<tr>
<th>Publications</th>
<th>Origin</th>
<th>AAO (range)</th>
<th>No. Cases</th>
<th>Clinical diagnosis</th>
<th>Other features</th>
</tr>
</thead>
<tbody>
<tr>
<td>Wasco et al., 1995 [1]</td>
<td>US</td>
<td>50</td>
<td>5</td>
<td>Alzheimer’s disease</td>
<td></td>
</tr>
<tr>
<td>Jacquemont et al., 2002 [4]</td>
<td>France</td>
<td>54</td>
<td>1</td>
<td>Atypical dementia with gait decline</td>
<td>Spastic paraparesis</td>
</tr>
<tr>
<td>Duminet et al., 2006 [6]</td>
<td>France</td>
<td>55</td>
<td>4</td>
<td>Presenile dementia</td>
<td>Abnormal white matter on MRI</td>
</tr>
<tr>
<td>Martikainen et al., 2010 [7]</td>
<td>Finland</td>
<td>46–51</td>
<td>3</td>
<td>Alzheimer’s disease (n = 1)</td>
<td>Spastic paraparesis; Visual hallucination; extrapyramidal signs, delusions</td>
</tr>
<tr>
<td>Ishii et al., 2012 [8]</td>
<td>Japan</td>
<td>43–51</td>
<td>2</td>
<td>Dementia with Lewy bodies (n = 2)</td>
<td>Anomaly, comprehension deficits, personality change, disinhibition</td>
</tr>
<tr>
<td>Lohmann et al., 2012 [9]</td>
<td>Turkey</td>
<td>51</td>
<td>2</td>
<td>Atypical Alzheimer’s/FTLD signs (n = 1)</td>
<td>Change in personality; frontotemporal atrophy on MRI</td>
</tr>
<tr>
<td>Current study</td>
<td>UK</td>
<td>43</td>
<td>1</td>
<td>Non-fluent/agrammatic PPA</td>
<td>Prominent early anxiety</td>
</tr>
</tbody>
</table>

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Supplementary Figure 1. Representative diffusion weighted imaging (A, b = 1000 s/mm² images; B, Apparent diffusion co-efficient images) acquired axially.

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dementia caused by PSEN1 mutation (P264L), responsible for
diagnosis of autosomal dominant early onset Alzheimer’s
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