

|                             |                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                           |
|-----------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Title                       | Hereditary leukoencephalopathy with axonal spheroids: a spectrum of phenotypes from CNS vasculitis to parkinsonism in an adult onset leukodystrophy series                                                                                                                                                                                                                                                                                                                                                |
| Authors                     | Lynch, David S.;Jaunmuktane, Zane;Sheerin, Una-Marie;Phadke, Rahul;Brandner, Sebastian;Milonas, Ionnis;Dean, Andrew;Bajaj, Nin;McNicholas, Nuala;Costello, Daniel J.;Cronin, Simon;McGuigan, Chris;Rossor, Martin;Fox, Nick;Murphy, Elaine;Chataway, Jeremy;Houlden, Henry                                                                                                                                                                                                                                |
| Publication date            | 2016-04-14                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                |
| Original Citation           | Lynch, D. S., Jaunmuktane, Z., Sheerin, U.-M., Phadke, R., Brandner, S., Milonas, I., Dean, A., Bajaj, N., McNicholas, N., Costello, D., Cronin, S., McGuigan, C., Rossor, M., Fox, N., Murphy, E., Chataway, J. and Houlden, H. (2016) 'Hereditary leukoencephalopathy with axonal spheroids: a spectrum of phenotypes from CNS vasculitis to parkinsonism in an adult onset leukodystrophy series', Journal of Neurology, Neurosurgery and Psychiatry, 87(5), pp. 512-519. doi:10.1136/jnnp-2015-310788 |
| Type of publication         | Article (peer-reviewed)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                   |
| Link to publisher's version | 10.1136/jnnp-2015-310788                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  |
| Rights                      | © 2016, the Authors. This is an Open Access article distributed in accordance with the terms of the Creative Commons Attribution (CC BY 4.0) license, which permits others to distribute, remix, adapt and build upon this work, for commercial use, provided the original work is properly cited. - <a href="http://creativecommons.org/licenses/by/4.0/">http://creativecommons.org/licenses/by/4.0/</a>                                                                                                |
| Download date               | 2024-04-20 08:25:20                                                                                                                                                                                                                                                                                                                                                                                                                                                                                       |
| Item downloaded from        | <a href="https://hdl.handle.net/10468/5178">https://hdl.handle.net/10468/5178</a>                                                                                                                                                                                                                                                                                                                                                                                                                         |



**University College Cork, Ireland**  
Coláiste na hOllscoile Corcaigh

Supplementary Table 2: Summary of the clinical presentations of the 43 patients in whom a *CSF1R* mutation was not found.

| Case      | Sex | Age at onset (y) | Clinical Features |                    |          |            |        | MRI Features | Additional                     |
|-----------|-----|------------------|-------------------|--------------------|----------|------------|--------|--------------|--------------------------------|
|           |     |                  | Cognitive Decline | Behavioural Change | Seizures | Spasticity | Ataxia | WML          |                                |
| <b>1</b>  | m   | 28               | -                 | -                  | -        | +          | -      | +            |                                |
| <b>2</b>  | f   | 25               | -                 | -                  | +        | -          | -      | +            |                                |
| <b>3</b>  | f   | 43               | +                 | +                  | -        | +          | -      | +            |                                |
| <b>4</b>  | f   | 36               | -                 | -                  | -        | +          | -      | +            | Vertigo, migraine              |
| <b>4</b>  | f   | 53               | +                 | +                  | -        | -          | -      | +            |                                |
| <b>5</b>  | m   | 33               | +                 | +                  | -        | -          | -      | +            | AD Family History              |
| <b>6</b>  | m   | 51               | +                 | -                  | -        | -          | -      | +            | AD Family History              |
| <b>7</b>  | f   | 42               | +                 | -                  | -        | -          | -      | +            |                                |
| <b>8</b>  | f   | 55               | +                 | +                  | -        | -          | -      | +            |                                |
| <b>9</b>  | m   | 57               | +                 | +                  | -        | -          | -      | +            | AD Family History              |
| <b>10</b> | m   | 18               | +                 | +                  | -        | +          | -      | +            |                                |
| <b>11</b> | m   | 52               | +                 | +                  | -        | +          | -      | +            | Myoclonus                      |
| <b>12</b> | m   | 35               | -                 | -                  | -        | -          | -      | +            | Migraine                       |
| <b>13</b> | f   | 76               | -                 | -                  | -        | +          | -      | +            | Lower limb sensory disturbance |
| <b>14</b> | m   | 57               | +                 | +                  | -        | -          | +      | +            |                                |
| <b>15</b> | f   | 21               | -                 | -                  | -        | +          | +      | +            |                                |
| <b>16</b> | m   | 18               | -                 | +                  | -        | -          | -      | +            | Upper limb tremor              |
| <b>17</b> | m   | 46               | +                 | -                  | -        | -          | -      | +            |                                |
| <b>18</b> | m   | 64               | +                 | -                  | -        | -          | +      | +            | Myoclonus                      |
| <b>19</b> | m   | 30               | -                 | -                  | -        | +          | +      | +            |                                |

|    |   |    |   |   |   |   |   |   |                              |
|----|---|----|---|---|---|---|---|---|------------------------------|
| 20 | m | 64 | - | - | - | + | + | + |                              |
| 21 | f | 53 | + | - | - | + | - | + | Vertigo                      |
| 22 | m | 18 | + | + | - | - | - | + |                              |
| 23 | m | 50 | + | + | - | - | - | + |                              |
| 24 | f | 47 | - | - | - | - | + | + |                              |
| 25 | f | 42 | + | + | - | - | - | + | AD Family History            |
| 26 | m | 60 | + | - | - | - | + | + |                              |
| 27 | f | 57 | + | + | - | - | + | + |                              |
| 28 | m | 37 | + | + | - | - | - | + |                              |
| 29 | f | 18 | - | - | + | + | - | + |                              |
| 30 | m | 44 | - | - | - | - | - | + | Visual Change,<br>dysarthria |
| 31 | f | 44 | + | - | - | - | + | + | Dysarthria                   |
| 32 | m | 45 | - | - | - | + | + | + |                              |
| 33 | m | 18 | + | + | - | + | - | + | Myoclonus                    |
| 34 | m | 22 | + | + | - | - | - | + |                              |
| 35 | f | 22 | + | - | - | - | - | + |                              |
| 36 | f | 44 | + | - | + | + | - | + |                              |
| 37 | f | 56 | + | - | - | - | - | + |                              |
| 38 | m | 23 | + | - | - | - | - | + |                              |
| 39 | m | 34 | + | - | - | - | - | + |                              |
| 40 | f | 68 | + | - | - | - | - | + |                              |
| 41 | f | 35 | + | - | - | + | - | + |                              |
| 42 | m | 42 | - | - | - | - | - | + |                              |
| 43 | f | 54 | - | - | + | + | - | + |                              |

WML: Symmetric T2 hyperintense white matter lesions, m: male, f: female, +: present, -:absent, AD: Autosomal Dominant