**Supplementary Table 1. Clinical information on carriers of observed ALS specific missense variants.**

|  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Mutation** | **Cohort** | **Sex** | **AAO****(yr)** | **Surv****(mo)** | **SOO** | **Myopathy** | **Muscle Biopsy** | **Deafness** | **Ataxia** | **Sensory****deficits** | **FTD** | **Family History** |
| p.Pro80Leu | BE | M | 61 | 54 | LE | Yes | Yes | No | No | No | No | No |
| p.Arg15Leu | NL | M | 73 | 76 | UE | Possible | No | Yes | No | No | No | No |
| p.Arg15Leu | US | F | 42 | - | LE | - | - | - | No | No | No | ALS |
| p.Arg15Leu | US | F | 71 | - | LE | - | - | - | No | No | No | ALS |
| p.Arg11Gly | US | F | 47 | 64 | LE | - | - | - | No | No | No | Myasthenia |

Overview of known clinical data available for carriers of ALS-specific rare *CHCHD10* missense mutations in our study; Country of sample origin (Cohort)(BE = Belgium, NL = The Netherlands, US = The United States of America), Age of onset in years (AAO), Survival after onset in months (Surv) Site of onset (SOO) (LE = lower extremities, UE = upper extremities), Clinical indications for Myopathy (Myopathy).

**Supplementary Table 2. Authors and contributions of Project MinE Sequencing Consortium.**

|  |
| --- |
| **Members of Project MinE Sequencing Consortium** |
| **Author** | **Contribution** |
|  | **Consortium Design** | **Whole Genome****Sequencing** | **Data****preparation** | **Data analysis** | **Manuscript****Writing** | **Manuscript****Revision** |
| Fulya Akçimen1 |  | X | X |  |  |  |
| Ammar Al-Chalabi, MD, PhD2 | X |  |  |  |  | X |
| A. Nazli Basak, PhD1 | X |  |  |  |  | X |
| Leonard H. van den Berg, MD, PhD3 | X |  |  |  |  | X |
| William J. Brands, MSc3 |  | X |  |  |  |  |
| Philip Van Damme, MD, PhD4 | X |  |  |  |  | X |
| Annelot M. Dekker, MD3 |  | X | X |  |  |  |
| Perry T.C. van Doormaal, MD3 |  | X |  |  |  |  |
| Kristel R. van Eijk, PhD3 |  |  | X |  |  |  |
| Michael A. van Es, MD, PhD3 | X |  |  |  |  | X |
| Jonathan D. Glass, MD5,6 | X |  |  |  |  | X |
| Orla Hardiman, PhD7,8 | X |  |  |  |  | X |
| Kevin P. Kenna, PhD3 |  |  | X |  |  | X |
| Cemile Kocoglu1 |  | X | X |  |  |  |
| Maarten Kooyman10 |  |  | X |  |  |  |
| John E. Landers, PhD9 | X |  |  |  |  | X |
| Russell L. McLaughlin, PhD11 | X | X |  |  |  | X |
| Bas Middelkoop3 |  |  | X |  |  |  |
| Matthieu Moisse, PhD4 |  | X |  |  |  | X |
| Jesus S. Mora, MD12 | X |  |  |  |  | X |
| Karen E. Morrison, MD, PhD13 |  |  |  |  |  | X |
| Stephen J. Newhouse, PhD14 | X |  |  |  |  |  |
| Monica Povedano, MD15,16 | X |  |  |  |  | X |
| Sara L. Pulit, PhD3 |  |  | X |  | X | X |
| Wouter van Rheenen, MD3 |  | X | X | X | X | X |
| Wim Robberecht, PhD4 | X |  |  |  |  | X |
| Raymond D. Schellevis, MSc3 |  | X |  |  |  |  |
| Christopher E. Shaw, MD, PhD17 | X |  |  |  |  | X |
| Pamela J. Shaw, MD, PhD17 | X |  |  |  |  | X |
| Rick A.A. van der Spek, MD3 |  | X | X |  |  | X |
| William Sproviero, PhD1 |  |  | X |  |  |  |
| Gijs. H.P. Tazelaar, MD3 |  |  |  | X | X | X |
| Ceren Tunca1 |  | X | X |  |  |  |
| Jan H. Veldink, MD, PhD3 | X |  |  |  | X | X |
| Joke J.F.A. van Vugt, PhD3 |  |  | X |  |  |  |

1Bogazici University, Suna and Inan Kirac Foundation, NDAL and Genomize Inc. TTR, Istanbul, Turkey;

2Maurice Wohl Clinical Neuroscience Institute, Department of Basic and Clinical Neuroscience, King’s College London, London, UK

3Department of Neurology, Brain Center Rudolf Magnus, University Medical Center Utrecht, Utrecht, the Netherlands;

4KU Leuven – University of Leuven, Department of Neurosciences,  Experimental Neurology; VIB Center for Brain & Disease Research; University Hospitals Leuven, Department of Neurology, Leuven, Belgium;

5Department of Neurology, Emory University School of Medicine, Atlanta, Georgia, USA;

6Emory ALS Center, Emory University School of Medicine, Atlanta, Georgia, USA;

7Academic Unit of Neurology, Trinity College Dublin, Trinity Biomedical Sciences Institute, Dublin, Ireland;

8Department of Neurology, Beaumont Hospital, Dublin, Ireland.

9Department of Neurology, University of Massachusetts Medical School, Worcester, Massachusetts, USA;

10SURFsara, Amsterdam, the Netherlands.

11Population Genetics Laboratory, Smurfit Institute of Genetics, Trinity College Dublin, Dublin, Ireland;

12Department of Neurology, Hospital San Rafael, Madrid, Spain;

13Faculty of Medicine, University of Southampton, Southampton, UK;

14Department of Clinical Neuroscience, King's College London, MRC Centre for Neurodegeneration Research, Institute of Psychiatry, London, UK

15Biomedical Network Research Center on Neurodegenerative Diseases (CIBERNED), Institute Carlos III, Hospitalet de Llobregat, Spain.

16Functional Unit of Amyotrophic Lateral Sclerosis (UFELA), Service of Neurology, Bellvitge University Hospital, Hospitalet de Llobregat, Spain.

17Sheffield Institute for Translational Neuroscience (SITraN), University of Sheffield, Sheffield, UK;