

## **Genetic Spectrum and Variability in Chinese Patients with Amyotrophic Lateral Sclerosis**

**Zhi-Jun Liu<sup>1#</sup>, Hui-Xia Lin<sup>1,2#</sup>, Qiao Wei<sup>1#</sup>, Qi-Jie Zhang<sup>2#</sup>, Cong-Xin Chen<sup>2</sup>, Qing-Qing Tao<sup>1</sup>,  
Gong-Lu Liu<sup>1</sup>, Wang Ni<sup>1</sup>, Aaron D. Gitler<sup>3</sup>, Hong-Fu Li<sup>1\*</sup>, Zhi-Ying Wu<sup>1\*</sup>**

<sup>1</sup>Department of Neurology and Research Center of Neurology in Second Affiliated Hospital, and Key Laboratory of Medical Neurobiology of Zhejiang Province, Zhejiang University School of Medicine, Hangzhou, China

<sup>2</sup>Department of Neurology and Institute of Neurology, First Affiliated Hospital, Fujian Medical University, Fuzhou, China

<sup>3</sup>Department of Genetics, Stanford University School of Medicine, Stanford, CA, USA

# SUPPLEMENTARY DATA

**Supplementary Table 1.** List of ALS-related genes targeted in the panel.

Gene	Inheritance	Locus	Refseq NM
<i>SOD1</i>	AD/AR	21q22.11	NM_000454.4
<i>TARDBP</i>	AD	1p36.22	NM_007375.3
<i>FUS/TLS</i>	AD	16p11.2	NM_001170634.1
<i>OPTN</i>	AD	10p13	NM_001008211.1
<i>PFN1</i>	AD	17p13.3	NM_005022.3
<i>VCP</i>	AD	9p13.3	NM_007126.3
<i>ANG</i>	AD	14q11.2	NM_001097577.2
<i>UBQLN2</i>	AD	Xp11.21	NM_013444.3
<i>TAF15</i>	AD	17q12	NM_003487.3
<i>EWSR1</i>	AD	22q12.2	NM_001163285.1
<i>HNRNPA1</i>	AR	12q13.13	NM_002136.3
<i>SETX</i>	AD	9q34.13	NM_015046.5
<i>CREST</i>	AD	20q13.33	NM_001301778.1
<i>MATR3</i>	AD	5q31.2	NM_001194954.1
<i>SQSTM1</i>	AD	5q35	NM_001142298.1
<i>CHMP2B</i>	AD	3p11.2	NM_001244644.1
<i>ALS2</i>	AR	2q33.1	NM_001135745.1
<i>VAPB</i>	AD	20q13.33	NM_001195677.1
<i>SIGMAR1</i>	AR	9p13.3	NM_001282205.1
<i>DCTN1</i>	AD	2p13	NM_001135040.2
<i>SPG11</i>	AR	15q14	NM_001160227.1
<i>NEFH</i>	AD/AR	22q12.2	NM_021076.3
<i>PRPH</i>	AD/AR	12q13.12	NM_006262.3
<i>ERBB4</i>	AD	2q34	NM_001042599.1
<i>FIG4</i>	AD	6q21	NM_014845.5
<i>CHCHD10</i>	AD	22q11.23	NM_001301339.1
<i>NEK1</i>	AD	4q33	NM_001199397.1

# SUPPLEMENTARY DATA

**Supplemental Table 2.** The primers for Quantitative Real-Time PCR were listed as followed

Year	Reference	FALS	SALS	SOD1	FUS	TARDBP
2005	Zhang et al. <sup>1</sup>	1	0	S106L (1F)	NA	NA
2006	Fong et al. <sup>2</sup>	1	0	I150T (1F)	NA	NA
2010	Xiong et al. <sup>3</sup>	5	71	NA	NA	S292N (1F)
2011	Zhao et al. <sup>4</sup>	2	0	C7F (1F), L85F (1F)	NA	NA
2011	Tsai et al. <sup>5</sup>	15	0	G86R (1F), T138R (1F), G139E (1F)	H517D (1F), R521H (1F)	M337V (2F), N378D (1F)
2011	Niu et al. <sup>6</sup>	8	0	C112Y (2F); G148D (1F)	NA	NA
2012	Zou et al. <sup>7</sup>	10	210	NA	R521L (1F), R521H (2S)	NA
2012	Zou et al. <sup>8</sup>	13	312	NA	NA	S292N (2S), G348V (1S)
2012	Hu et al. <sup>9</sup>	1	0	V30Dfs*8 (1F)	NA	NA
2012	Wu et al. <sup>10</sup>	1	0	C147X (1F)	NA	NA
2012	Zhang et al. <sup>11</sup>	43	0	G7S (1F), G17A (2F), G17S (1F), E22G (1F), G42D (1F), H47R (1F), L85F (1F), G94R (1F), G130Wfs*3 (1F)	NA	NA
2012	Huang et al. <sup>12</sup>	0	165	NA	NA	N378S (1S)
2013	Cui et al. <sup>13</sup>	0	51	S135T (1S)	NA	NA
2013	Ye et al. <sup>14</sup>	0	207	NA	NA	0
2013	Xu et al. <sup>15</sup>	1	0	UTR variant (138278 A>G)	NA	NA
2013	Zou et al. <sup>16</sup>	6	110	0	P525L (1S), G504Wfs*12 (1S), R495X (1S), R521G (1F)	NA
2014	Song et al. <sup>17</sup>	30	131	L9V (1F), G11A (1F), G17S (1S), G38R (1S), D84N (1F), G86R (1F), L107F (1F), C112Y(1S), T138R (2F+1S), G139E (1F)	H517D (1F+1S), R521H (1F+1S)	M337V (4F), G348V (1F), N378D (1F), I383V (1F)
2014	Wang et al. <sup>18</sup>	1	0	C7W (1F)	NA	NA
2014	Liu et al. <sup>19</sup>	8	0	F21C (1F), G148D (1F), C147R (1F)	0	0
2014	Yang et al. <sup>20</sup>	4	0	G17A (1F), E134V (1F), G148D (1F), L39R (1F)	NA	NA
2015	Zou et al. <sup>21</sup>	0	2	L85F (1S)	P525L (1S)	NA
2015	Zou et al. <sup>22</sup>	20	324	C7S (1F), G42D (1F), H47R(1F+1S), L85F (1S), D91A (1S), L107V (1F), G142A (1F)	R495X (1S), G504Wfs*12 (1S), R521H (2S), R521G (1F), R521L (1F), P525L (2S)	S292N(2S), G348V (1S)
2016	Chen et al. <sup>23</sup>	1	0	N132Qfs*5 (1F)	NA	NA
2016	Zou et al. <sup>24</sup>	1	1	H47R (1F+1S)	NA	NA
2016	Liu et al. <sup>25</sup>	20	234	G42D (1F), H47R (1F), P67R (1S), P67S (1F), S69P (1F), G86R (1F), D102E (1S), L107F (1S), I114T (1F), S135T (1S), E134V (2S)	R495X (1S), P525L (1S), G229S (1S)	NA
2016	Zou et al. <sup>26</sup>	0	16	L85F (1S)	R495X (1S), G504Wfs*12 (1S), P525L (2S)	NA
2016	Hou et al. <sup>27</sup>	15	158	D102N (1F), D102G (1F), C112Y (1F+1S), N87S (1S), V88A (1S)	R521H (1F), P525_Y526insY (1F)	NA
2016	Niu et al. <sup>28</sup>	1	0	G42D (1F)	NA	NA
2016	Ju et al. <sup>29</sup>	18	200	NA	NA	M337V (1F), S393L (1S)
2017	Wei et al. <sup>30</sup>	12	487	L39V (1F), G42D (1F), H44R (1S), P67S (1S), G73D (1S), C112Y (1F), I114T (1S), V120F (1S)	NA	NA

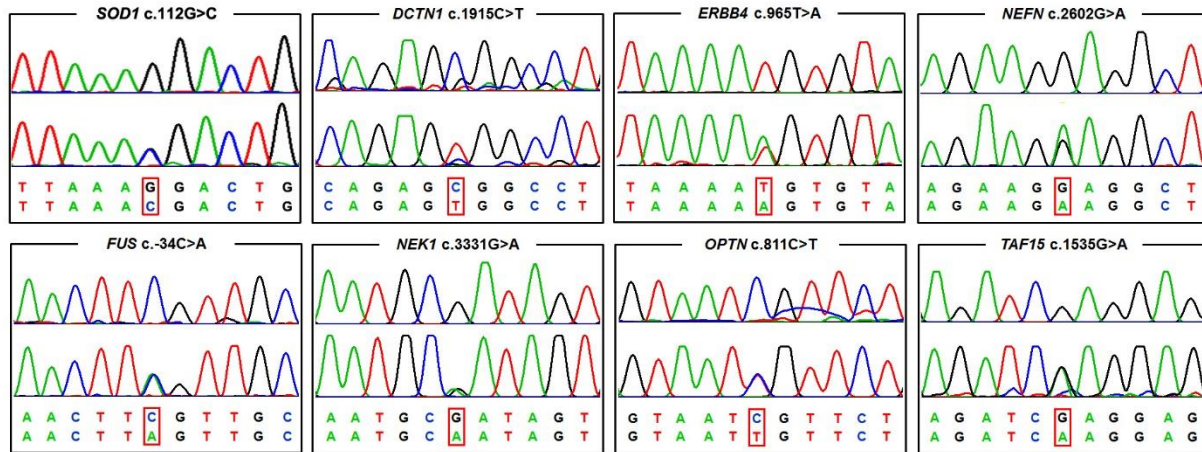
## SUPPLEMENTARY DATA

2017	Liu et al. <sup>31</sup>	2	10	0		G515Vfs*14 (1S), Q519Ifs*9 (1S), G486Pfs*30 (1S), R498Afs*32 (1F)	G348V(1F)
2018	Li et al. <sup>32</sup>	1	0	D91_K92insN (1F)	NA		NA
2018	Tang et al. <sup>33</sup>	1	0	A5V (1F)	NA		NA
2018	Xu et al. <sup>34</sup>	7	0	0	0		M337V(4F), G348C(1F) A315T(1S)
2018	Zhang et al. <sup>35</sup>	0	311	G38V (1S), H47R (1S), G86R (1S), E101K (1S), C112Y (1S), N140K (1S), G142R (1S)	0		
2019	Tang et al. <sup>36</sup>	159	923	A5S (1F), A5V (1F), C7S (1F), V15M (2F), G17C (1F), G17A (2F), F21V (1F), E22G (1F), W33G (1S), G38R (1F), G38V (1F), L39R (1F), E41G (1F), G42D (3F), G42S (2F+1S), H44R (2F+1S), H47R (6F+2S), V48A (2S), P67S (1F), R80S (1F), L85F (2F), G86C (1F), N87S (1S), A90F (1F), G94R (1F), V95G (1S), E101G (1F), S106L (1S), L107F (2F+1S), L107V (1F), C112Y (1F+1S), I114T (1F), V119L (1S), G130Wfs*3 (1F), N132QfsTer5 (1F), E133Ter (1S), E134V(1F), N140K (1S), G142A (3F), A146T (1S), G148D (1F), I150V (2S), I150T (1S)	NA		NA
2019	Current	24	21	A5S (1F), A5V (1F),G11V (1F), G17C (1F), G38R (1F), H47R (2F), D84G (1F), L107V (1F), H121Q (2F), I150T (1F)	R495X (1F), K510E (1S), R521C (1S), R521H (1F), P525L(2S)		M337V (1F), G357S (1S)

Abbreviations: NA =not available, FALS= familial ALS, SALS= sporadic ALS.

Note: Mutations reported in reference 7, 8, 16, 21, 24, 26 were included in reference 22. As also, mutation reported in reference 11, 20 and 33 were included in reference 36.

# SUPPLEMENTARY DATA



Supplementary Figure 1. **Title and description here**

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