

Supplementary Table 1. Summary of all published *UBQLN2* mutations.

Publication	Mutant	ExAc Freq	Sex	AOO	Survival	Site of onset	FTD/Cognitive	Inheritance	Ethnicity
Daoud	S155N	2/43191	M	51	alive	bulbar	No	SALS	French-French Canadian
Daoud	P189T	Novel	F	46	60	spinal	No	SALS	French-French Canadian
Daoud	P189T	Novel	M	59	48	spinal	No	SALS	French-French Canadian
Synofzik	A282V	Novel	M	54	alive	cognitive	Yes	FTD	German
Synofzik	A283T	1/43660	M	72	48	limb	No	FALS	German
Kim HJ	D314E	4/42845	F	60	N/A	limb	N/A	SALS	Korean
Kim HJ	D314E	4/42845	M	30	N/A	limb	N/A	SALS	Korean
Lattante	T336A	Novel	F	52	96	cognitive	FTLD	N/A	French
Ozoguz	S340I	Novel	F	58	alive	N/A	Dementia	FALS	Turkish
Ozoguz	S340I	Novel	M	12	alive	N/A	No	FALS	Turkish
Ozoguz	S340I	Novel	M	20	alive	N/A	No	FALS	Turkish
Dillen	S346C	2/41441	F	77	30	N/A	Yes	N/A	Belgian
Ozoguz	M392I	Novel	M	14	N/A	limb	No	SALS	Turkish
Ozoguz	M392I	Novel	F	16	N/A	limb	No	SALS	Turkish
Huang	M392V	Novel	M	59	alive	limb	No	SALS	Chinese
Dillen	S400G	1/43029	F	78	6	N/A	No	FALS	Belgian
Synofzik	Q425R	Novel	F	51	alive	limb	No	FALS	German
Dillen	P440L	5/43442	F	30	56	N/A	No	SALS	Bulgarian Gypsy
Gellera	M446R	Novel	F	43	36	spinal	N/A	FALS	Italian
Ugwu	T467I	Novel	F	70	N/A	N/A	Yes	FTD	UK
Williams	T487I	Novel	F	58	alive	limb	No	FALS	European
Williams	T487I	Novel	F	30	132	limb	No	FALS	European
McCann	T487I	Novel	N/A	N/A	N/A	N/A	N/A	FALS	Australian
Teyssou	A488T	Novel	M	47	51	limb	No	SALS	French
Teyssou	<b>P494L</b>	Novel	F	55	alive	limb	No	FALS	French
Deng	<b>P497H</b>	Novel	F	53	47	N/A	No	FALS	NA
<b>This study</b>	<b>P497H</b>	Novel	M	39	108	limb	No	FALS	UK
Gellera	<b>P497H</b>	Novel	M	33	25	bulbar	FTD	FALS	Italian
Teyssou	<b>P497S</b>	Novel	F	48	63	bulbar	FTD/ALS	FALS	French
Deng	<b>P497S</b>	Novel	M	29	19	N/A	No	FALS	NA
Teyssou	<b>P500S</b>	Novel	F	62	16	limb	No	FALS	French
Huang	<b>P500_G502d</b>	Novel	M	61	alive	limb	No	SALS	Chinese
Millecamps	<b>G502_I504d</b>	Novel	F	52	24	bulbar	N/A	FALS	French
Teyssou	<b>P506A</b>	Novel	M	35	alive	limb	No	FALS	French
Ozoguz	<b>P506S</b>	Novel	M	26	alive	limb	No	FALS	Turkish

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Gellera	P506S	Novel	M	30	60	spinal	N/A	FALS	Italian
Vengoechea	P506S	Novel	F	34	alive	cognitive	FTD	FALS	NA
<b>This study</b>	<b>P506S</b>	Novel	F	54	36	cognitive	FTD/ALS	FALS	UK
Deng	P509S	2/20965	F	53	36	N/A	No	FALS	NA
Deng	P525S	18/1918 1	F	71	58	N/A	No	FALS	NA
Ozoguz	P525S	18/1918 1	M	22	alive	limb	No	FALS	Turkish
Gellera	P533L	Novel	F	34	6	spinal	N/A	FALS	Italian
Gellera	V538L	1/22231	F	57	24	bulbar	N/A	SALS	Italian

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