



Supplemental Fig. 1 Box plots showing regional TDP-43 burden differences and burden distribution between *GRN* and *C9ORF72* cases. The line in box plots represents the median, the box represents interquartile range and the whiskers correspond to minimum and maximum values. *P*-values are from Wilcoxon rank sum test

Supplemental Table 1 Demographic and neuropathologic characteristics by mutation: *GRN* vs *C9ORF72*

Characteristic	No. (% or interquartile range)		p-value
	<i>GRN</i> n = 6	<i>C9ORF72</i> n=9	
Female	3 (50%)	3 (33%)	0.62
White/Caucasian	6 (100%)	8 (89%)	>0.99
Age at onset, years	66 (64 – 69)	69 (64 – 72)	0.37
Disease duration, years	12 (10 – 19)	10 (8 – 15)	0.55
Age at death, years	80 (76 – 86)	79 (76 – 83)	>0.99
Family history	4 (67%)	6 (67%)	>0.99
Education, years	16 (10 – 18)	13 (12 – 16)	>0.99
Last MMSE/30	13 (3 – 26)	17 (5 – 25)	0.79 ^a
Clinical diagnosis			
FTLD clinical spectrum diagnosis	3 (43%)	2 (22%)	
<i>Frontotemporal dementia</i>	2 (29%)	1 (11%)	
<i>Primary progressive aphasia</i>	1 (14%)	0 (0%)	
<i>Progressive supranuclear palsy</i>	0 (0%)	1 (13%)	0.28
Alzheimer's dementia	3 (43%)	6 (67%)	
Dementia with Lewy bodies	0 (0%)	1 (11%)	
Vascular dementia	1 (14%)	0 (0%)	
Pathologic characteristics			
Time to autopsy, hours	3 (3 -3)	5 (3 – 9)	0.28
Brain weight, grams	737 (615 – 1055)	920 (880 – 1070)	0.17
Tau (Braak NFT 0-VI)			
Median Braak NFT stage	1 (0 – 2)	3 (2 – 4)	0.012
Braak 0	2 (33%)	0 (0%)	
Braak I	2 (33%)	0 (0%)	
Braak II	1 (17%)	2 (22%)	
Braak III	1 (17%)	4 (44%)	0.11
Braak IV	0 (0%)	1 (11%)	
Braak V	0 (0%)	2 (22%)	
Braak VI	0 (0%)	0 (0%)	
Amyloid-β (Thal phases 0 -5)			
Median CERAD	0 (0 – 0)	0 (0 – 0)	0.50
Median Thal phase	1 (0 – 2)	1 (0 – 3)	0.58
0 (no amyloid-β)	2 (33%)	3 (33%)	
1 (neocortical)	3 (50%)	3 (33%)	
2 (allocortex)	1 (17%)	1 (11%)	>0.99
3 (thalamus, striatum)	0 (0%)	1 (11%)	
4 (brainstem)	0 (0%)	1 (11%)	
5 (cerebellum)	0 (0%)	0 (0%)	

Alzheimer's disease neuropathologic changes			
High likelihood	0 (0%)	0 (0%)	
Intermediate likelihood	0 (0%)	3 (33%)	0.23
Low likelihood/none	6 (100%)	6 (67%)	
Hippocampal sclerosis	4 (67%)	6 (67%)	>0.99
<i>moderate/severe</i>	4 (67%)	5 (56%)	>0.99
TDP-43 type			
Type A	6 (100%)	7 (78%)	0.49
Type B	0 (0%)	2 (22%)	
Cerebrovascular disease			
Vascular score (0 – 4)	2 (0 – 3)	0 (0 – 2)	0.26
0 (no lesions)	2 (33%)	5 (56%)	
1 (mild CAA/arteriolosclerosis)	0 (0%)	1 (11%)	
2 (≥moderate CAA/arteriolosclerosis)	2 (33%)	2 (22%)	0.75
3 (micro-infarcts, no large infarcts)	1 (17%)	1 (11%)	
4 (lacunar/large infarcts)	1 (17%)	0 (0%)	
Argyrophilic grains disease	1 (17%)	1 (11%)	>0.99

Data are represented as median (range) for continuous variables or number (per cent) for categorical variables. *P*-values are from Wilcoxon rank sum test or Fisher's exact test

^a adjusted for disease duration at time of last MMSE