

Supplementary Table 3: Clinical characteristics of pure small fiber neuropathy patients with a (potentially) pathogenic *SCN11A* variant

c position	Gender	Age	Age debut	EMFD (5th percentile)	EMFD	TTT	Associated condition(s)	Erythromalgia	Ich	Cramps	Family history	Actual VAS	VASmin	VASmax	NPS Pain Severity	Sharpness	Hottness	Dullness	Coldness	Sensitivity	Itchiness	Unpleasantness	Intensity deep pain	Intensity surface pain	SFN-SQ No. autonomic	SFN-SQ No. somatic	Neuropathic pain medication at moment hospital visit	
c.93C>T	F	55	54	6.8 (4.3)	Normal	Abnormal	Autoimmune disease	No	No	No	Negative	NA	NA	NA	6	6	0	6	0	8	0	NA	6	8	7	3	Amtripyline 30mg daily, pregabalin 75mg BID	
c.155C>T	F	47	44	5.4 (5.7)	Abnormal	Normal	Autoimmune disease, IGT	Yes	No	No	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	None	
c.151C>T	F	54	52	2.9 (4.3)	Abnormal	Abnormal	Autoimmune disease	Yes	No	No	Negative	91	71	98	10	9	9	10	9	5	0	10	10	8	7	3	Amtripyline 50mg daily	
c.1090C>T	F	52	51	9 (4.3)	Normal	Abnormal	Idiopathic	No	No	No	Negative	77	48	87	7	8	7	3	6	2	2	7	8	5	4	4	None	
c.1142T>C	F	55	50	8.1 (4.3)	Normal	Abnormal	Idiopathic	No	Yes	No	Positive	48	12	90	6	8	7	3	0	8	7	6	7	7	5	3	None	
c.1257G>T	F	55	52	1.7 (4.3)	Abnormal	Normal	Autoimmune disease	Yes	No	No	Negative	64	16	81	8	8	7	7	7	8	2	NA	8	NA	7	4	Amtripyline	
c.1257G>T	F	33	22	6.1 (7.1)	Abnormal	Normal	Idiopathic	No	No	No	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	9	4	Gabapentin 300mg TID	
c.1257G>T	F	28	26	7.1 (8.4)	Abnormal	Abnormal	Idiopathic	No	No	No	Negative	77	27	88	8	NA	10	7	9	NA	0	8	8	7	8	4	Tramadol 100mg daily	
c.1257G>T	F	57	49	4.3 (4.3)	Normal	Abnormal	Idiopathic	No	Yes	No	Negative	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	Capzacin creme	
c.1257G>T	F	34	30	9 (1.1)	Normal	Abnormal	Idiopathic	Yes	No	Yes	Negative	NA	NA	NA	8	9	10	6	7	8	8	9	9	7	9	4	None	
c.1744G>A	F	70	66	3.1 (2.2)	Normal	Abnormal	Idiopathic	No	No	No	Negative	52	16	59	6	5	7	6	4	7	4	7	7	6	8	4	Amtripyline 25mg TID, buprenorphine transdermal	
c.1744G>A	F	51	48	9.6 (4.3)	Normal	Abnormal	Idiopathic	Yes	No	Yes	Positive	23	56	89	7	NA	NA	NA	NA	NA	NA	NA	NA	NA	7	4	None	
c.1744G>A	F	69	49	7.4 (3.7)	Normal	Abnormal	Autoimmune disease	No	No	No	Positive	NA	NA	NA	5	4	4	4	5	5	5	6	5	5	8	4	None	
c.1744G>A	F	27	24	9.4 (8.4)	Normal	Abnormal	Idiopathic	No	Yes	Yes	Negative	75	35	84	6	7	4	7	6	7	7	8	8	2	9	4	Dicyclonone on occasion	
c.1744G>A	M	56	53	5.1 (3.5)	Normal	Abnormal	Idiopathic	No	No	No	Negative	20	3	62	3	3	6	3	6	1	1	NA	4	4	3	2	Amtripyline 10mg and 25mg daily	
c.1927A>G	M	70	67	8.1 (2.1)	Normal	Abnormal	Idiopathic	No	No	No	Negative	75	72	87	8	7	9	6	7	7	7	2	8	8	8	3	4	None
c.2042C>A	M	74	65	4.4 (2.1)	Normal	Abnormal	Autoimmune disease	No	No	Yes	Negative	73	68	100	8	9	8	8	8	7	6	8	9	8	7	4	Gabapentin 400mg BID, amtripyline	
c.2095G>A	M	69	64	5.7 (2.8)	Normal	Abnormal	Idiopathic	Yes	No	No	Positive	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	None	
c.2419A>G	F	59	57	11.2 (4.3)	Normal	Abnormal	DM	Yes	No	Yes	Positive	36	23	70	3	5	6	2	3	4	4	3	2	3	9	4	None	
c.2419A>G	F	59	57	5.7 (4.3)	Normal	Abnormal	Idiopathic	No	No	No	Negative	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	None	
c.2513G>A	M	59	56	0.5 (3.5)	Abnormal	Abnormal	IGT	No	No	No	Negative	68	49	91	8	9	8	8	7	7	8	7	8	7	7	4	Amtripyline 10mg daily	
c.2524G>C	F	76	66	2.6 (2.2)	Normal	Abnormal	Idiopathic	Yes	No	No	Positive	22	10	65	7	8	7	9	7	5	4	8	7	9	6	4	Amtripyline	
c.3222A>G	M	39	38	7.7 (5.2)	Normal	Abnormal	IGT	No	No	No	Negative	22	20	30	3	4	2	4	0	1	0	4	3	1	7	0	Carbamazepine 200mg QID, pregabalin 75mg BID	
c.3473T>C	F	61	59	4.8 (3.2)	Normal	Abnormal	Idiopathic	No	Yes	No	Positive	NA	NA	NA	7	7	10	10	8	3	9	7	8	6	9	4	None	
c.3473T>C	M	73	72	3.1 (2.1)	Normal	Abnormal	Idiopathic	No	No	Yes	Negative	71	33	82	8	8	2	8	2	8	8	8	8	8	8	6	4	Pregabalin 75 BID
c.3473T>C**	F	63	60	1.0 (3.7)	Abnormal	Abnormal	Idiopathic	Yes	No	No	NA	16	21	65	5	6	7	5	6	5	3	5	3	3	6	4	Gabapentin 300mg daily	
c.3745G>A	M	51	50	6.1 (3.5)	Normal	Abnormal	Idiopathic	Yes	No	Yes	Negative	65	38	86	7	8	8	6	0	4	0	7	8	7	7	4	None	
c.4049G>A	F	46	30	5.8 (5.7)	Abnormal	Abnormal	Idiopathic	No	No	Yes	Positive	62	16	88	6	3	7	5	3	6	7	8	7	4	9	4	None	
c.4049G>A	F	32	30	11.4 (7.1)	Normal	Abnormal	Idiopathic	No	No	No	Negative	65	22	91	6	8	8	5	9	6	7	4	8	5	7	2	Pregabalin 150mg and 300mg	
c.4957T>G>A	F	45	42	8 (1.7)	Normal	Abnormal	Idiopathic	No	No	Yes	Positive	5	0	30	3	6	1	6	0	8	6	5	NA	NA	8	4	None	
c.4382G>A	M	59	55	0.4 (3.5)	Abnormal	Normal	Idiopathic	Yes	No	No	Negative	36	11	65	3	7	7	6	0	1	4	10	2	9	0	3	Lamotrigine 100mg BID	
c.4309G>A	M	44	42	6.3 (4.4)	Normal	Abnormal	Idiopathic	No	No	No	Negative	84	NA	93	9	10	8	4	4	4	8	4	10	9	9	8	4	None
c.5067C>G	F	20	19	7.3 (8.4)	Abnormal	Abnormal	Autoimmune disease	No	No	No	Negative	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	Nortriptyline 30mg daily, gabapentine 300mg daily

F: female; M: male; EMFD: intraepidermal nerve fiber density; TTT: temperature threshold testing; VAS: visual analogue scale (VAS is ranging from 0-100 (0 = no pain and 100 = the worst possible pain)); min: minimum; max: maximum; NPS: neuropathic pain scale (0 = not applicable to the experienced pain and 10 = in the most severe form applicable to the experienced pain); SFN-SQ: small fiber neuropathy inventory questionnaire; No. autonomic: number of positive autonomic symptoms; No. somatic: number of positive somatic symptoms; NA: not available; DM: diabetes mellitus; IGT: impaired glucose tolerance; BID: twice daily; TID: three times daily; QID: four times daily.
 **Also *SCN11A* variant c.1555G>A; p.(Glu518Val).