Supplementary Table 1: CMTNS scores for rare mutations. The diseases and genes are named according to OMIM (http://www.ncbi.nlm.nih.gov/Omim/) and HUGO (http://www.genenames.org/), respectively.

CMT subtype (mutation)	Median CMTNS (n)	Range	First/third quartile
CMT4A (*GDAP1*)	9 (3)	8-32	8.5/20.5
CMT1F (*NEFL*)	16 (7)	5-29	11/21.5
CMT1E (*PMP22)*	18 (5)	11-20	15/18
CMT4C (*SH3TC2*)	22 (7)	2-32	6.5/16
HSAN1 (*SPTLC1*)	13 (22)	2-34	15/27

Supplementary Table 2: CMTNS scores for rare mutations with N<3**

CMT subtype (mutation)	CMTNS		
	patient 1	patient 2	patient 3
CMT1C (*LITAF*)	6	20	
CMT1D (*EGR2*)	16		
CMT2C (*TRPV4*)	8	2	14
CMT2D (*GARS*)	2	5	
CMT4B1 (*MTMR2*)	32	15	
CMT4F (*PRX*)	17		
AR CMT2A (*LMNA*)	26		
CMT4J (*FIG4*)	3	17	29
HMN5A (*BSCL2*)	3	7	

**Due to low numbers summary statistics were not performed for the above mutations.

Supplementary Table 3: Age and gender adjusted mean CMTNS scores for individual sites*.

site	Age and gender adjusted Mean CMTNS			
	CMT1A	CMT1B	CMT2A	CMT1X
London	15.8	15.7	14.4	16.8
Milan	9.2	9.2	7.9	10.3
Detroit	14	13.9	12.6	15.0
Iowa	13.2	13.2	11.9	14.3
all others	13.3	13.3	12.0	14.4

***These are model predicted means ,which have been adjusted for age and gender.**

Supplementary Table 4: Mean CMTES scores for individual sites.

site	Mean CMTNS +/- SD (n)			
	CMT1A	CMT1B	CMT2A	CMT1X
London	10.2 +/- 4.3(104)	14 +/- 5.6(9)	14.1 +/- 7.5(11)	10.5 +/- 3.2(37)
Milan	6.6+/- 4.2(123)	10.9 +/- 7.7(13)	9.2 +/- 6.6(13)	7.2 +/- 4.9(24)
Detroit	8.3+/- 5.6(144)	8.4 +/- 5.7(19)	10.8 +/- 7.8(17)	10.2 +/- 6.0(23)
Iowa	7.3+/- 4.8(39)	8.7 +/- 6.8(3)	11.8 +/- 5.5(6)	5.8 +/- 6.3(4)
all others	10.1 +/- 5.3(98)	9.4 +/- 2.3(9)	7.8 +/- 4.8(9)	7.8 +/- 3.5(8)

Supplementary Table 5: Previously reported CMT series.

The table summarizes the proportion of different types and subtypes of CMT in selected papers that describe CMT patients who were analyzed for the causative mutation. Both the numerator (the genes that were tested) and the denominator (the population of CMT patients who were analyzed) differ between each paper. The table indicates the genes that were tested, and whether HNPP, CMTDI, HSN/HSAN, and/or HMN were excluded. ND: not determined. Of note, the OMIM classification of CMT subtypes used in this table reflects that employed by the authors and differs from that used in the current study.

	mutations	[30] Boerkoel, et al., 2002	[15]Latour et al. 2006	[10]Saporta et al. 2011	[21]Abe et al. 2011	[19] Braathan et al. 2011	[18]Karadima et al. 2011	[14]Murphy et al. 2012- seen in clinic	[14]Murphy et al. 2012- not seen in clinic	[20] Sivera et al. 2013	[16] Gess et al., 2013
Clinical diagnosis											
HNPP		excluded	excluded	6% (48/787)	0.6% (2/354)	excluded	excluded	8% (46/601)	4% (56/1493)	excluded	14% (83/589)
CMT1		94% (145/153)	100% (968/968)	55% (434/787)	62% (111/354)	49% (92/187)	100% (243/243)	40.% (240/601)	30% (446/1493)	63% (275/438)	60% (355/589)
CMT2		4% (6/153)	excluded	12% (96/787)	36% (127/354)	47% (88/187)	excluded	19% (115/601)	22% (335/1493)	37% (163/438)	26% (151/589)
CMTDI		excluded	excluded	4% (31/787)	excluded	3% (7/187)	excluded	10.% (62/601)	2% (23/1493)	excluded	excluded
HSN/HSAN		excluded	excluded	2% (17/787)	excluded	excluded	excluded	11% (69/601)	9% (129/1493)	excluded	excluded
HMN		excluded	excluded	1% (7/787)	excluded	excluded	excluded	10.% (61/601)	8% (126/1493)	excluded	excluded
other/ unknown		3% (2/153)	excluded	20.% (154/787)	1% (5/354)	excluded	excluded	1% (8/601)	25% (378/1493)	excluded	excluded
Total (all		153	968	787	354	187	243	601	1493	438	589

forms reported in each paper)	% found	67% 103/153	95% (921/968)	67% (527/787)	40.% (143/354)	29% (54/187)	31% (76/243)	44% (266/601)	33% (497/1493)	83% (365/438)	58% (339/589)
"CMT1" (+/- DSN, CHN, CMT4)	% total reported	96% (147/153)	100% (968/968)	55% (434/787)	64% (225/354)	49% (92/187)	100% (243/243)	40.% (240/601)	30.% (446/1493)	63% (275/438)	60.% (355/589)
	% found	68% (100/147)	95% (921/968)	98% (481/489)	51% (116/227)	52% (48/92)	32% (76/243)	80.% (193/240)	60% (269/446)	96% (263/275)	48% (111/233)
"CMT2" (+/- recessive)	% total reported	4% (6/153)	ND	12% (96/787)	36% (127/354)	47% (88/187)	ND	19% (115/601)	22% (335/1493)	37% (163/438)	26% (151/589)
	% found	33% (2/6)	ND	34% (33/96)	21% (27/127)	7% (6/88)	ND	25% (29/115)	13% (44/335)	63% (102/163)	35% (53/151)
CMTDI	% total reported	ND	ND	4% (31/787)	ND	4% (7/187)	ND	10.% (62/601)	2% (23/1493)	ND	ND
	% found	ND	ND	ND	ND	ND	ND	60.% (37/62)	22% (5/23)	ND	ND
HNPP	% total reported	ND	ND	6% (48/787)	0.6% (2/354)	ND	ND	8% (46/601)	4% (56/1493)	ND	14% (83/589)
	% found	ND	ND	ND	ND	ND	ND	67% (31/46)	38% (21/56)	ND	64% (53/83)
other/ unknown	% total reported	ND	ND	22% (171/787)	ND	ND	ND	1% (8/601)	25% (378/1493)	ND	ND
	% found	ND	ND	ND	ND	ND	ND	ND	34% (128/378)	ND	ND

Supplementary Table 6: Previously reported CMT series.

The table summarizes the proportion of different types and subtypes of CMT in selected papers that describe CMT patients who were analyzed for the causative mutation. Both the numerator (the genes that were tested) and the denominator (the population of CMT patients who were analyzed) differ between each paper. The table indicates the genes that were tested, and whether HNPP, CMTDI, HSN/HSAN, and/or HMN were excluded. ND: not determined. Of note, the OMIM classification of CMT subtypes used in this table reflects that employed by the authors and differs from that used in the current study.

	mutations	[30] Boerkoel, et al., 2002	[15]Latour et al. 2006	[10]Saporta et al. 2011	[21] Abe et a. 2011	[19] Braathan et al. 2011	[18]Karadima et al. 2011	[14]Murphy et al. 2012- seen in clinic	[14]Murphy et al- not seen in clinic	[20] Sivera et al. 2013	[16] Gess et al., 2013
Demyelinating - HNPP, CMT1, and autosomal recessive											
HNPP	*PMP22* mutation	excluded	excluded	6% (48/787)	0.6% (2/354)	excluded	excluded	5% (31/601)	1% (21/1493)	excluded	9% (53/589)
CMT1A	*PMP22* duplication	52% (79/153)	77% (741/968)	55% (290/787)	14% (50/354)	20.% (37/187)	26% (63/243)	28% (168/601)	17% (247/1493)	49% (184/438)	31% (180/589)
CMT1B	*MPZ*	3% (5/153)	6% (59/968)	9% (45/787)	6% (20/354)	1% (2/187)	0.4% (1/243)	2% (13/601)	1% (18/1493)	4% (19/438)	3% (19/589)
CMT1C	*LITAF*	ND	0.6% (6/968)	1% (5/787)	0%	0%	ND	0.7% (4/601)	0.1% (2/1493)	0%	0%
CMT1D	*EGR2*	0.7% (1/153)	0.1% (1/968)	0.2% (1/787)	0.3% (1/354)	0%	ND	0%	0.2% (4/1493)	0%	0%
CMT1E	*PMP22* other	3% (5/153)	0.3% (3/968)	1% (5/787)	4% (10/354)	0%	ND	1% (6/601)	0.3% (5/1493)	0.5% (2/438)	0.7% (4/589)
CMT1F	*NEFL*	0%	0.5% (5/968)	0% (0/787)	2% (8/354)	0%	ND	0%	0.1% (2/1493)	0.3% (1/438)	0%
CMT1X	*GJB1*	7% (11/153)	11% (106/968)	15% (80/787)	7% (25/354)	5% (9/187)	5% (12/243)	8% (46/601)	7% (101/1493)	15% (56/438)	8% (47/589)
	FBLN5	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND
	ARHGEF1	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND
CMT4A	*GDAP1*	ND	ND	0.2% (1/787)	0%	ND	ND	0.3% (2/601)	0.7% (10/1493	11% (42/438)	0%
CMT4B1	*MTMR2*	ND	ND	ND	0%	ND	ND	0.2%	0.1%	0%	0%

								(1/601)	(1/1493)		
CMT4B2	*MTMR13*	ND	ND	ND	0%	ND	ND	ND	ND	0%	ND
CMT4B3	*SBF1*	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND
CMT4C	*SH3TC2*	ND	ND	0.6% (3/787)	ND	ND	ND	1% (5/601)	0.3% (4/1493)	6% (28/438)	0%
CMT4D	*NDRG1*	ND	ND	ND	ND	ND	ND	ND	ND	1.9% (7/438)	0.3% (2/589)
CMT4E	*EGR2*	0%	ND	ND	0%	0%	ND	0%	0%	0%	0%
CMT4F	*PRX*	0.7% (1/153)	ND	0.2% (1/787)	1% (5/354)	ND	ND	0.2% (1/601)	0%	1.1% (4/438)	0%
CMT4G	*HK1*	ND	ND	ND	ND	ND	ND	ND	ND	1% (6/438)	ND
CMT4H	*FGD4*	ND	ND	ND	ND	ND	ND	ND	ND	0.5% (2/438)	0.2% (1/589)
CMT4J	*FIG4*	ND	ND	0.4% (2/787)	ND	ND	ND	ND	ND	0%	ND
<u>CMTDI</u>		excluded	excluded		excluded		excluded			excluded	excluded
CMTDIA											
CMTDIB	*DNM2*	ND	ND	ND	0%	ND	ND	ND	ND	0%	ND
CMTDIC	*YARS*	ND	ND	ND	0%	ND	ND	ND	ND	0.2% (1/438)	ND
CMTDID	*MPZ*	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND
CMTDIE	*IFN2*	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND
CMTDIF	*GNB4*	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND
<u>Axonal</u>			excluded				excluded				
CMT2A	*MFN2*	ND	ND	4% (21/787)	4% (14/354)	3% (6/187)	ND	2% (12/601)	3% (48/1493)	2% (6/438)	2% (12/589)
CMT2B	*RAB7*	ND	ND	ND	0%	ND	ND	ND	ND	0%	ND
CMT2C	*TRPV4*	ND	ND	ND	ND	ND	ND	0.5% (3/601)	0.1% (1/1493)	0.3% (1/438)	0%
CMT2D	*GARS*	ND	ND	0.6% (3/787)	0.3% (1/354)	ND	ND	ND	ND	0.9% (4/438)	0.3% (2/589)
CMT2E	*NEFL*	0.7%	ND	0.8%	0%	ND	ND	0.3%	(2/1493)	0.9%	0%

		(1/153)		(4/787)				(2/601)		(4/438)	
CMT2F	*HSPB1*	ND	ND	ND	0%	ND	ND	0.3% (2/601)	0.1% (1/1493)	2% (7/438)	0%
CMT2G											
CMT2I&J	*MPZ*	ND	ND	ND	1.0% (5/354)	ND	ND	0%	0%	4% (10/438)	0.3% (2/589)
CMT2K	*GDAP1*	ND	ND	1% (5/787)	0%	ND	ND	0%	0%	5% (24/438)	0%
CMT2L	*HSPB8*	ND	ND	ND	0%	ND	ND	0%	0%	0.7% (3/438)	0%
CMT2M	*DNM2*	ND	ND	ND	0%	ND	ND	ND	ND	0%	ND
CMT2N	*AARS*	ND	ND	ND	ND	ND	ND	ND	ND	0%	ND
CMT2O	*DYNC1HI*	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND
CMT2P	*LRSAM1*	ND	ND	ND	ND	ND	ND	ND	ND	0%	ND
CMT2Q	*DHTKD1*	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND
HMSNP	*TFG*	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND
CMT2	*TUBA8*	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND
CMT2	*HARS*	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND
CMT2	*MARS*	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND
CFEOM1	*TUBB3*	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND
CMT2N	*DCAF8*	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND
HNA	*SEPT9*	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND
	MTATP6	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND
CMTX5	*PDK3*	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND
ARCMT2B1	*LMNA*	ND	ND	ND	ND	ND	ND	ND	ND	0%	ND
ARCMT2B2	*MED25*	ND	ND	ND	ND	ND	ND	ND	ND	0%	ND
ARCMT2	*GDAP1*	ND	ND	ND	0.3% (1/354)	ND	ND	0.3% (2/601)	0.7% (10/1493)	4% (18/438)	0%
ARCMT2	*MFN2*	ND	ND	0%	0%	0%	ND	0%	0%	0%	0%
ARCMT2	*NEFL*	ND	ND	0%	0%	ND	ND	0%	0%	0%	0%
ARCMT2	*LRSAM1*	ND	ND	ND	ND	ND	ND	ND	ND	0%	ND
ARCMT2	*KARS*	ND	ND	ND	ND	ND	ND	ND	ND	0.5% (1/438)	ND
ARCMT2	*HINT1*	ND	ND	ND	ND	ND	ND	ND	ND	0%	ND
ARCMT2	*TRIM2*	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND

ARCMT2	*PLEKHG5*	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND
GAN	*GAN*	ND	ND	ND	ND	ND	ND	0.2% (1/601)	0%	0%	2% (9/589)
HSAN and HSN		excluded	excluded		excluded	excluded	excluded			excluded	excluded
HSN reflux											ND
HSN1A	*SPTLC1*	ND	ND	ND	ND		ND	2% (14/601)	0%	ND	ND
HSN1B											
HSN1C	*SPTLC2*	ND	ND	ND	ND		ND	1% (5/601)	0%	ND	ND
HSN1D	*ATL1*	ND	ND	ND	ND		ND	ND	ND	ND	ND
HSN1E	*DNMT1*	ND	ND	ND	ND		ND	ND	ND	ND	ND
HSAN2A	*WNK1*	ND	ND	ND	ND		ND	ND	ND	ND	ND
HSN2B	*FAM134*	ND	ND	ND	ND		ND	0.2% (1/601)	0%	ND	ND
HSN2C	*KIF1A*	ND	ND	ND	ND		ND	ND	ND	ND	ND
HSN2D	*SCN9A*	ND	ND	ND	ND		ND	ND	ND	ND	ND
HSAN3	*IKBKAP*	ND	ND	ND	ND		ND	ND	ND	ND	ND
HSAN4	*NTRKA*	ND	ND	ND	ND		ND	ND	ND	ND	ND
HSAN5	*NGFB*	ND	ND	ND	ND		ND	0.2% (1/601)	0%	ND	ND
HSAN6	*DST*	ND	ND	ND	ND		ND	ND	ND	ND	ND
HMN		excluded	excluded		excluded	excluded	excluded			excluded	excluded
HMN1											
HMN2A	*HSPB8*	ND	ND	ND	ND		ND	0.2% (1/601)	0%	0%	0%
HMN2B	*HSPB1*	ND	ND	ND	ND		ND	1% (5/601)	0.1% (1/1493)	1.9% (7/438)	0%
HMN2C	*HSPB3*	ND	ND	ND	ND		ND	ND	ND	ND	ND
HMN2	*FBLN5*	ND	ND	ND	ND		ND	ND	ND	ND	ND
HMN2	*HARS*	ND	ND	ND	ND		ND	ND	ND	ND	ND
HMN5A	GARS	ND	ND	ND	ND		ND		ND	1.1% (4/438)	0.3% (2/589)

HMN5A	*BSCL2*	ND	ND	ND	ND		ND	0.2% (1/601)	0.1% (1/1493)	0%	0%
HMN2C	*REEP1*	ND	ND	ND	ND		ND	ND	ND	ND	ND
HMN2	*SPAST*	ND	ND	ND	ND		ND	ND	ND	ND	ND
HMN2	*SETX*	ND	ND	ND	ND		ND	ND	ND	ND	ND
DSMA1	*IGHMBP2*	ND	ND	ND	ND		ND	ND	ND	ND	ND
DSMA2											
DSMA3											
DSMA4	*PLEKHG5*	ND	ND	ND	ND		ND	ND	ND	ND	ND
DSMA5	*HSJ1*	ND	ND	ND	ND		ND	ND	ND	ND	ND
DSMA6	*FBXO38*	ND	ND	ND	ND		ND	ND	ND	ND	ND
SMA	*SMN*	ND	ND	ND	ND		ND	0.2% (1/601)	ND	ND	ND
HMN7A	*SLC5A7*	ND	ND	ND	ND		ND	ND	ND	ND	ND
HMN7B	*DCTN1*	ND	ND	ND	ND		ND	ND	ND	ND	ND
SMAX3	*ATP7A*	ND	ND	ND	ND		ND	ND	ND	ND	ND
scapulo-peroneal SMA	*TRPV4*	ND	ND	ND	ND		ND	ND	ND	0%	0%
congenital distal SMA	*TRPV4*	ND	ND	ND	ND		ND	ND	ND	0%	0%
SMALED1	*DYNC1HI*	ND	ND	ND	ND		ND	ND	ND	ND	ND
SMALED2	*BICD2*	ND	ND	ND	ND		ND	ND	ND	ND	ND