

Table 1. Diagnostic data

	Pathogenic Variants	AMN Pre-treatment (% of total fatty acids)							AMN Post-treatment (% of total fatty acids)						
		C22:0	C24:0	C26:0	C28:0	C30:0	C24:1(n-9)	C26:1(n-9)	C22:0	C24:0	C26:0	C28:0	C30:0	C24:1(n-9)	C26:1(n-9)
EC-001	p.Arg401Gln	0.67%	1.12%	0.052%	0.0063%	0.0013%	1.38%	0.024%	0.70%	1.13%	0.049%	0.0065%	0.0016%	1.57%	0.028%
EC-002	p.Arg554His	0.48%	0.87%	0.042%	0.0048%	0.0011%	1.10%	0.026%	0.49%	0.83%	0.043%	0.0045%	0.0015%	1.05%	0.026%
EC-003	p.Val457fs*	0.71%	1.20%	0.049%	0.0054%	0.0018%	0.75%	0.014%	0.73%	1.16%	0.050%	0.0064%	0.0017%	0.65%	0.013%
EC-004	p.Tyr174Cys	0.75%	1.25%	0.057%	0.0080%	0.0021%	0.95%	0.019%	0.75%	1.22%	0.056%	0.0074%	0.0011%	0.98%	0.020%
EC-005	p.Tyr181Cys	0.78%	1.24%	0.043%	0.0057%	0.0016%	0.86%	0.012%	0.63%	0.98%	0.042%	0.0054%	0.0018%	0.55%	0.011%
EC-006	p.Val102Glu	0.62%	1.02%	0.039%	0.0039%	0.0009%	0.62%	0.013%	0.63%	1.09%	0.045%	0.0047%	0.0008%	0.62%	0.014%
EC-007	p.Ser108Ter	0.79%	1.52%	0.059%	0.0071%	0.0010%	0.96%	0.020%	0.74%	1.35%	0.054%	0.0054%	0.0015%	0.79%	0.016%
EC-008	p.Gly277Arg	0.73%	1.08%	0.045%	0.0111%	0.0049%	1.31%	0.022%	0.86%	1.37%	0.054%	0.0095%	0.0028%	1.04%	0.017%
EC-009	p.Leu628Pro	0.44%	0.86%	0.048%	0.0048%	0.0011%	0.63%	0.014%	0.72%	1.34%	0.059%	0.0062%	0.0012%	0.68%	0.017%
EC-010	p.Arg617Leu	0.58%	1.23%	0.080%	0.0119%	0.0024%	1.25%	0.036%	0.77%	1.45%	0.073%	0.0101%	0.0023%	1.46%	0.037%
EC-011	p.Arg389Gly	0.75%	1.24%	0.043%	0.0047%	0.0013%	0.88%	0.014%	0.89%	1.51%	0.052%	0.0044%	0.0013%	0.86%	0.013%
EC-012	p.Arg389Gly	0.91%	1.17%	0.037%	0.0044%	0.0010%	0.86%	0.012%	0.89%	1.18%	0.030%	0.0037%	0.0014%	0.97%	0.014%
EC-013	p.Tyr181Ter	0.72%	1.21%	0.057%	0.0071%	0.0016%	1.07%	0.018%	0.78%	1.41%	0.065%	0.0076%	0.0021%	0.95%	0.022%
CONTROLS															
media (n=13)		0.873%	0.818%	0.010%	0.0010%	0.00003%	1.05%	0.006%	0.873%	0.818%	0.010%	0.0010%	0.00003%	1.05%	0.006%
SD		0.054	0.051	0.000350	0.000094	0.000031	0.042	0.00036	0.054	0.051	0.000350	0.000094	0.000031	0.042	0.00036

*Novel pathogenic variant

Table 2a. Evoked potential data. BAEP=Brainstem Auditory Evoked Potential; SEP=Somatosensory Evoked Potential; MEP=Motor Evoked Potential; LEP=Laser Evoked Potential ;pre=pre- treatment; post=post-treatment; R=right; L=left; NR=no response; in bold: abnormal values; CMCT=central motor conduction time; ND: not done; in bold= abnormal values

ID	AGE	SIDE	MEP (ms)		MEP (ms)		BAEP (ms)						LEP (ms)				VEP (70°)					
			CMCT	CMCT	CMCT	CMCT	I-III		III-V		I-V		TRIG N2/P2 (ms)		UL N2/P2 (ms)		LL N2/P2 (ms)		P100 lat (ms)		P100 ampl (µV)	
			pre	post	pre	post	pre	post	pre	post	pre	post	pre	post	pre	post	pre	post	pre	post	pre	post
1	24	R	25.8	34.1	10.9	10.9	3.2	2.94	2.84	2.9	6.04	5.84	181	171	216	220	336	324	96.5	98.5	15.2	14
		L	29.7	32.8	9.42	9.2	2.78	2.68	3.16	2.94	5.94	5.62	171	176	222	218	301	350	92.5	99	14	15.3
2	48	R	22.05	20	7.6	5.6	2.56	2.68	2.02	2	4.58	4.68	176	162	220	222	260	278	104.5	104.5	18.9	19.6
		L	20.05	17	7.9	6.82	2.64	2.5	1.92	2	4.56	4.5	178	176	213	216	250	258	101.5	102	20.1	24.5
3	35	R	22.02	18.62	7.3	8.52	2.9	3.1	2.46	2.58	5.36	5.68	173	164	222	230	232	252	101	101.5	5	7.4
		L	20.5	20.05	5.77	7.25	2.94	3.02	2.5	2.4	5.44	5.42	178	163	228	215	251	248	101	99.5	8.2	5.8
4	44	R	20.7	17.25	8.2	6.5	2.54	2.68	3.14	2.94	5.68	5.62	156	155	202	214	254	251	108.5	108	24.3	24.1
		L	19.9	18.8	8.3	6.1	3.08	3.02	2.92	2.88	6	5.9	155	153	210	208	259	247	108	104.5	21	24.6
5	45	R	22.5	23.8	8.7	9.8	3.04	3	2.58	2.66	5.62	5.66	198	189	233	263	NR	NR	98	102	13.7	17.2
		L	23.8	19.6	12.2	10.5	2.7	3	2.5	2.42	5.2	5.4	198	190	229	267	NR	NR	99.5	101	10	17
6	37	R	NR	28.7	11.8	11.5	3.12	3.12	2.96	2.8	6.08	5.92	246	191	280	264	NR	NR	118	115	2.3	1.7
		L	NR	23.1	9.7	8.22	3.1	3.18	3.04	2.82	6.14	6	245	205	277	259	NR	NR	114.5	118	3.9	2.1
7	50	R	23.9	20.45	8.52	8.25	2.4	2.4	2.32	2.38	4.72	4.78	197	197	265	268	NR	NR	102	104	14.8	10.4
		L	21.6	21.6	8.25	7.6	2.78	2.52	2.2	2.28	5	4.8	185	186	273	252	NR	NR	104	103.5	13.8	10.4
8	41	R	27.4	30.12	10.35	13.35	3	3.04	2.82	2.82	5.82	5.86	209	198	268	237	389	345	105	108.5	8.9	17.4
		L	27.6	31.52	9.75	8.65	3.22	2.92	2.88	2.98	6.1	5.9	199	196	262	245	333	388	102	107.5	20.7	16.9
9	28	R	29.4	26.65	8.9	8.85	2.8	2.82	2.54	2.5	5.34	5.32	159	158	198	194	361	347	131	115.9	2.5	3.9
		L	26.25	25.8	9.02	9.3	2.9	3	2.62	2.46	5.52	5.46	167	179	187	194	357	349	121	121.5	6.8	6.3
10	38	R	24.6	20.15	9.85	8.35	2.24	2.92	2.92	2.64	5.88	5.56	232	201	275	225	318	277	107	110.5	6.3	5.4
		L	25.4	22.32	11.6	9.82	2.86	2.86	2.6	2.64	5.46	5.5	237	199	256	225	290	255	106	110	8.1	9.6
11	37	R	25.65	24.1	9.55	9.5	2.76	2.81	2.7	2.9	5.46	5.71	176	171	231	220	NR	NR	106.5	109.5	12.3	14.4
		L	NR	28.9	8.45	8.12	2.8	2.85	2.52	2.98	5.32	5.83	190	197	219	213	NR	NR	107	105.5	12.7	13
12	64	R	17.07	19.8	7.95	8.8	2.72	2.78	2.42	2.24	5.14	5.02	180	177	245	246	NR	NR	111	112	18.6	15.6
		L	17.07	17.8	8.1	8.2	2.42	2.58	2.44	2.14	4.86	4.72	162	176	236	238	NR	NR	114.5	116.5	18.3	17.4
13	31	R	28	24.85	12.45	10.1	3.26	3.4	2.96	2.86	6.22	6.26	182	182	233	229	280	254	101.5	108	7.5	5.4
		L	27	26.05	9.3	8.65	3.34	3.3	2.6	2.58	6	5.88	183	181	243	220	293	273	104.5	106	5.3	7.7
Upper limit of normative values (mean + 2.5 SD)			15,7		7		2.47		2.35		4.57		206		255		283		110		26.4	

Table 2b. Evoked potential data. SEP=Somatosensory Evoked Potential; pre=pre-treatment; post=post-treatment; R=right; L=Left; NR=no response; in bold: abnormal values; ND: not done

ID	AGE	SIDE	SEP LL (ms)								SEP UL (ms)									
			N8		N22		P40		N22-P40		N9		N13		N20		N9-N13		N13-N20	
			pre	post	pre	post	pre	post	pre	post	pre	post	pre	post	pre	post	pre	post	pre	post
1	24	R	8	8.2	22.8	23.1	NR	NR	ND	ND	10.6	11	15.75	15.7	24.15	25	5.15	4.7	8.4	9.3
		L	8.2	8.2	23.1	23.1	NR	NR	ND	ND	10.45	10.65	14.8	14.75	24.3	24.4	4.35	4.1	9.5	9.7
2	48	R	8.5	8.2	23.9	23.9	49.8	48.5	25.9	24.6	10.7	10.5	15.15	14.8	22.05	21.4	4.45	4.3	6.9	6.6
		L	8.8	8.4	23.7	23.9	49.8	49	26.1	25.1	10.5	10.25	15.5	14.45	21.4	21.3	5	4.2	5.9	6.85
3	35	R	10.8	10.5	29.8	29.8	47	48.3	17.2	18.5	12.6	12.2	16.75	17.25	23	23.6	4.15	5.05	6.25	6.35
		L	10.2	10.5	29	29.4	46.8	47.7	17.8	18.3	12.5	12.05	16.9	17.2	23	23	4.4	5.15	6.1	5.8
4	44	R	10.1	9.7	25.6	25.4	58.7	55.3	33.1	29.9	11	10.04	15.7	14.6	22.65	22.4	4.7	4.56	6.95	7.8
		L	9.8	9.3	26.2	25.1	58.5	55.4	32.3	30.3	11	10.03	15.45	15.1	22.25	21.7	4.45	5.07	6.8	6.6
5	45	R	NR	NR	NR	NR	NR	NR	ND	ND	10.8	11	NR	NR	23.9	24.2	ND	ND	ND	ND
		L	NR	NR	NR	NR	NR	NR	ND	ND	11.1	11.25	NR	NR	24.2	24.3	ND	ND	ND	ND
6	37	R	9.7	9.9	26	25.9	NR	NR	ND	ND	11.25	11.5	NR	NR	24.5	24.6	ND	ND	ND	ND
		L	9.7	NR	26.1	24.8	NR	NR	ND	ND	11.15	11.4	NR	NR	24.8	24.6	ND	ND	ND	ND
7	50	R	NR	NR	NR	30.4	60.2	60.9	ND	30.5	12.8	12.85	NR	NR	26.45	26.2	ND	ND	ND	ND
		L	NR	NR	30.6	30.2	60	60	29.4	29.8	12.6	12.25	NR	17.25	25.5	25.9	ND	5.0	ND	8.65
8	41	R	NR	NR	28.3	27.6	NR	58.13	ND	30.53	11.3	11.3	16.05	15.52	25.1	24.95	4.75	4.22	9.05	9.43
		L	NR	NR	28.2	27.29	NR	58.33	ND	31.04	11	11.2	14.65	15.05	24.7	24.74	3.65	3.85	10.05	9.69
9	28	R	9.6	9	25.6	24.6	NR	NR	ND	ND	10.7	10.5	14.05	15.25	23.9	22.8	3.35	4.75	9.85	7.55
		L	10	9.6	25.5	24.4	NR	NR	ND	ND	10.25	10.1	14.85	14.25	22.15	22.1	4.6	4.15	7.3	7.85
10	38	R	9.6	9.1	26.3	25.3	57.2	57.6	30.9	32.3	10.65	10.5	15.65	14.8	22.8	23.2	5	4.3	7.15	8.4
		L	9.6	9.2	26	25.6	58.8	58.4	32.8	32.8	10.65	10.6	15.4	15.65	23	23.25	4.75	5.05	7.6	7.6
11	37	R	8.5	8.23	24.5	24.17	NR	NR	ND	ND	9.75	9.74	14.9	15.1	22.2	22.3	5.15	5.36	7.3	7.2
		L	9.3	8.96	25.2	24.7	NR	NR	ND	ND	9.8	9.6	14.4	14.6	22.8	22.4	4.6	5	8.4	7.8
12	64	R	9.4	8.3	22.2	22.9	49.4	50.9	27.2	28	10.05	10.4	14.15	13.2	20.95	21.2	4.1	2.8	6.8	8
		L	8	7.9	21.5	22.2	48.5	48.5	27	26.3	9.5	9.75	13.9	14.2	20.95	21.2	4.4	4.45	7.05	7
13	31	R	NR	NR	NR	NR	NR	NR	ND	ND	12.1	12.2	16.3	16.5	25.8	26.45	4.2	4.3	9.5	9.95
		L	NR	NR	NR	NR	NR	NR	ND	ND	12.1	11.9	16.8	16.5	25.9	26.3	4.7	4.6	9.1	9.8
Upper limit of normative values (mean + 2.5 SD)			10.16		24.63		42.91		20.16		11.21		14.3		22.46		4.33		6.95	

Table 3. Nerve conduction data. pre=pre-treatment; post=post-treatment; R= Right; L= Left; SENS NCS= Sensory Nerve Conduction Studies; MOT NCS= Motor Nerve Conduction Studies; Lat= Latency; AMP= Amplitude; NCV= Nerve Conduction Velocity; NR= No Response; in bold =abnormal values

D	AGE	SIDE	SENS NCS (Sural Nerve)						MOT NCS (Peroneal Nerve)					
			LAT (ms)		AMP(μV)		NCV (m/s)		LAT (MS)		AMP (μV)		NCV (m/s)	
			pre	post	pre	post	pre	post	pre	post	pre	post	pre	post
1	24	R	2.4	2.65	23.7	23.5	58.3	52.8	3.8	3.65	29.9	25.1	43.1	49.2
		L	2.45	2.75	25.3	19.6	57.1	50.9	3.9	3.3	26.1	24.1	44.6	43.5
2	48	R	2.65	2.9	22	21.7	52.8	49.3	5.65		8.2		47.1	
		L	2.7	2.55	20.3	21	51.9	48.3	5.15	5.4	8.9	4.3	54.5	49.6
3	35	R	2.6	2.65	11.4	6.6	53.8	48.3	5.75	3.65	6.2	7.4	42	38
		L	3.1	2.95	9	6	45.2	44.5	4.85		7.3		41.8	
4	44	R	2.5	2.35	8	4.4	56	62.2	3.8	4.2	6.4	3.8	44	42
		L	2.4	2.35	9.8	11.3	58.3	59.6	4.9		2.6		43.7	
5	45	R	2.8	2.3	9.9	17.7	51.8	51.2	3.55	4.3	4.2	1.1	42.4	40.7
		L	2.7	2.35	10	19.9	53.8	50.1		4.15		1.9		42.2
6	37	R	2.85	2.65	14.6	14.3	49.1	47.1	4.55	4.75	2.2	8.2	45.9	43.7
		L	2.5		22.9		56							
7	50	R	2.85	2.55	15.3	15.3	41.5	42	4.55	4.85	8.5	6	33.3	32.9
		L	2.5	2.4	11.7	15.9	48	46.2		4.85		6		33.1
8	41	R	2.75	3.35	9.3	8.2	54.5	41.8	3.95	4.75	5.3	7.1	41.9	38.8
		L	2.9	3.3	10.3	8.1	50	42.4		4.9		4.1		37.5
9	28	R	2.3	2.2	15.7	15	60.9	63.6	4.95	3.35	5.5	7.9	43.1	45.6
		L	2.6		9.9		53.7							
10	38	R	2.75		17.6		50.9		6.3	5.3	10.6	5	42.2	43.5
		L	2.15	2.6	21.4	14.7	60.5	53.8						
11	37	R	2.35	2.85	9.8	11.1	59.6	49.1	4.55	4.45	7.8	3.9	43.8	44.6
		L	2.45		7.3		57.1							
12	64	R	NR	NR	NR	NR	NR	NR	4.2	4.8	1.2	1.3	48.8	48
		L	NR	NR	NR	NR	NR	NR						
13	31	R	2.75	2.95	15.5	20.8	50.9	47.5	5.85	6.25	2.4	3.5	24.3	22.5
		L	2.55	3	19.3	24.8	54.9	46.7	6.75	6.5	2.3	3.4	25.2	24.6

NORMATIVE VALUES

	Amp	Lat	NCV
Peroneal Nerve			
Upper limit (mean + 2SD)			
Range 23-35 y-o	5.4 +/- 3	3.7 +/- 1.8	49.5 +/- 10.8
Upper limit (mean + 2SD)			
Range 44-65 y-o	5.0 +/- 2.6	3.7 +/- 1.4	48.3 +/- 9.2
Sural nerve			
Upper limit (mean + 1SD)			
Range 23-35 y-o	20.9 +/- 8.0	2.7 +/- 0.3	52.5 +/- 5.6
Upper limit (mean + 1SD)			
Range 44-65 y-o	17.2 +/- 6.7	2.8 +/- 0.3	51.1 +/- 1.3
Tibial Nerve			
Upper limit (mean + 2SD)	5.8 +/- 3.8	3.96 +/- 2.00	48.5 +/- 7.2
Superficial Peroneal Nerve			
Upper limit (mean + 1SD)			
Age over 15 y-o	13.9 +/- 4.6	2.24 +/- 0.49	47.3 +/- 3.4

Table 4. Spectroscopic and Diffusion Tensor Imaging data

ID	AGE	Spectroscopy								Difusion Tensor Imaging	
		Myo/Cr		NAA/Cho		NAA/Cr		Cho/Cr		FA	
		PO		PO		PO		PO		CST	
		pre	post	pre	post	pre	post	pre	post	pre	post
1	24	0.8838	0.9046	2.10	2.65	1.6879	1.3367	0.8041	0.5043	0.515269	0.494690
2	48	0.8387	0.7861	2.32	2.41	1.7097	1.3668	0.7359	0.5661	0.529420	0.528411
3	35	1.0382	0.8109	1.31	1.74	1.6279	1.6218	1.2452	0.9345	0.554577	0.545326
4	44	0.7874	0.8153	3.54	1.73	1.6556	1.8241	0.4677	1.0514	0.512509	0.479226
5	45	0.9500	0.8596	1.52	1.89	2.0600	1.2682	1.3596	0.6696	0.522925	0.513929
6	37	0.9652	1.1509	1.76	1.45	1.4299	0.9899	0.8133	0.6818	0.497866	0.4635
7	50	0.9858	0.9693	2.16	1.20	1.4871	1.4559	0.6897	1.2145	0.492247	0.477074
8	41	0.7809	0.7233	1.65	2.18	1.5551	1.3841	0.9416	0.6360	0.530123	0.526146
9	28	0.7474	0.8401	6.79	2.37	1.6173	1.6132	0.2382	0.6808	0.518413	0.512709
10	38	0.8376	1.0453	1.73	3.58	1.8619	1.4057	1.0754	0.3928	0.485787	0.491679
11	37	0.7669	1.2308	2.68	4.66	1.6889	1.0735	0.6296	0.2306	0.554720	0.534809
12	64	1.0243	0.9385	3.25	3.14	1.5255	1.3483	0.4699	0.4298	0.531937	0.506943
13	31	0.9693	0.9111	1.20	1.70	1.4559	1.5715	1.2145	0.9258	not done	not done
mean		0.8904	0.9220	2.4614	2.3611	1.6433	1.4046	0.8219	0.6860	0.5205	0.5062
SD		0.10	0.15	1.48	0.96	0.17	0.22	0.34	0.28	0.02	0.03
Normative values											
mean		0.5700		2.2000		1.4400		0.6600		0.5700	
(mean +/- 3 SD)		< 0.81		2.95-1.45		> 1.800		0.42-0.90		> 0.51	

FA=Fractional anisotropy; CST= Corticospinal Tract; PO=Parieto-occipital white matter; in bold: abnormal values; in red: withdrawn from treatment; Myo: myo-inositol; Cre: creatine; Cho: choline; NAA: N-acetylaspartate

Normative values (spectroscopy): Garnett et al; Brain 2000;123 (Pt 7):1403-1409.

Normative values (DTI): Dubey et al; Ann Neurol 2005;58:758-766.