

Supplementary Table S1. Common CPA6 variants in JME patients.

	Control n (%)	JME n (%)
rs10957393 (c.133 T>C, p.Phe45Leu)		
MAF: C=0.2322		
Allele	476	248
T	394 (82.8)	206 (83.1)
C	82 (17.2)	42 (16.9)
p_u		0.921
Genotype	238	124
TT	156 (65.5)	82 (66.1)
CT	82 (34.5)	42 (33.9)
CC	0 (0.0)	0 (0.0)
p_u		0.912
rs17853192 (c.518 C>G, p.Ser173Cys)		
MAF: G = 0.0891		
Allele	474	243
C	439 (92.6)	230 (93.5)
G	35 (7.4)	16 (6.5)
p_u		0.661
Genotype	237	123
CC	202 (85.2)	107 (87.0)
CG	35 (14.8)	16 (13.0)
GG	0 (0.0)	0 (0.0)
p_u		0.999
rs17343819 (c.746 A>G, p.Asn249Ser)		
MAF: G = 0.0791		
Allele	476	248
A	411 (86.3)	201 (83.0)
G	65 (13.7)	41 (17.0)
p_u		0.245
Genotype	238	121
AA	176 (74.0)	82 (67.8)
AG	59 (24.8)	37 (30.6)
GG	3 (1.2)	2 (1.6)
p_u		0.767

p_u = uncorrected p -value

MAF = minor allele frequency (source: 1000 genomes, accessed Oct 2014)