

Table e-1:

Case, Age, Sex	Gene Involved; Allele 1/Allele 2	VA RE:LE	Color vision (test method)	Retinal Examination	FAF	SD-OCT	Full-field ERG
P1, 21, M	<i>EPM2A</i> ; deletion of Exons 1 & 2/ deletion of Exons 1 & 2	20/40: 20/25	Normal RG and BY (HRR charts)	Optic Disc: N, Macula: N, Vessels: N, Background: N, Retinal Periphery: N	N	CRT: N, retinal lamination: N	Rod system: selective bipolar involvement; Cone system: mild dysfunction
P2, 22, F	<i>EPM2A</i> ; p. Asn163Asp/ p. Ala254Metfs*33	20/40: 20/40	Normal RG and BY (HRR charts)	Optic Disc: N, Macula: N, Vessels: N, Background: N, Retinal Periphery: N	N	CRT: N, retinal lamination: N	Rod system: normal; Cone system: mild dysfunction
P3, 21, M	<i>EPM2B</i> ; p.Pro69Ala/ p.Pro69Ala	20/25: 20/25	Normal RG (Ishihara)	Optic Disc: N, Macula: N, Vessels: N, Background: N, Retinal Periphery: N	N	CRT: N, retinal lamination: N	Rod system: photoreceptor & bipolar involvement; Cone system: moderate dysfunction
P4, 31, M	<i>EPM2B</i> ; p.Asp308Val/ p.Asp308Val	20/25: 20/25	Normal RG (Ishihara)	Optic Disc: N, Macula: N, Vessels: N, Background: N, Retinal Periphery: N	N	CRT: N, retinal lamination: N	Rod system: photoreceptor & bipolar involvement; Cone system: moderate dysfunction

Abbreviations: VA = distance visual acuity, RE = right eye, LE = left eye, P = patient, M = male, F = female, FAF = fundus autofluorescence, SD-OCT = spectral domain optical coherence tomography, ERG = electroretinography, RG = red-green, BY = blue-yellow, HRR = Hardy Rand Rittler charts, N = normal, CRT = central retinal

Table e-2:

Case, Age at testing, Sex	Age of Disease Onset	Simplified Myoclonus rating scale score ¹ at the time of testing	Medications at the time of testing
P1, 21, M	14	3	Devalproex sodium 1000 mg twice daily Levetiracetam 500 mg twice daily Zonisamide 100 mg thrice daily Rufinamide 200mg twice daily Clonazepam 1 mg twice daily
P2, 22, F	14	4	Devalproex sodium 2500 mg twice daily Levetiracetam 1500 mg twice daily Zonisamide 200 mg twice daily Rufinamide 1600mg twice daily Clonazepam 1 mg twice daily Lorazepam 4mg twice daily Perampanel 10 mg at night
P3, 21, M	14	3	Levetiracetam 1500 mg twice daily Clonazepam 2 mg twice daily
P4, 31, M	16	3	Valproic acid 2000 mg daily Levetiracetam 1500 mg twice daily Zonisamide 100 mg thrice daily Clonazepam 2mg twice daily Metformin 500mg twice daily

Abbreviations: M = male, F = female.

References:

1. Magaouda A, Gelisse P, Genton P. Antimyoclonic effect of levetiracetam in 13 patients with Unverricht-Lundborg disease: clinical observations. *Epilepsia* 2004;45:678-681.