

# A rare occurrence of myoclonus-fixed dystonia syndrome

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Myoclonus   Dystonia   Ketamine

## INTRODUCTION

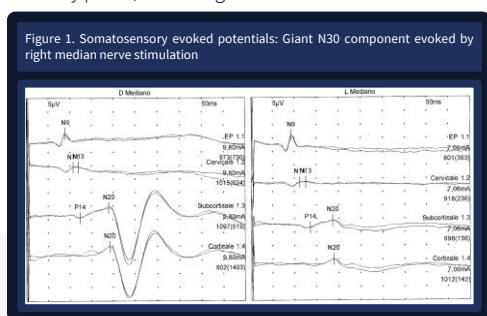
Myoclonus–dystonia syndrome is a rare genetic movement disorder of childhood–early adult onset, defined by myoclonic jerks with dystonic postures.<sup>1</sup> Cortical/cortico-subcortical myoclonus shows giant somatosensory evoked potentials (SEPs), and increased cortico-muscular coherence, though with limited spatial specificity.<sup>2</sup> Fixed dystonia may reflect sensorimotor network dysfunction rather than a purely functional disorder.<sup>3</sup>

## PATIENT

A 54-year-old woman with a history of hormone receptor–positive breast cancer (2016; disease-free until May 2023). In February 2023, she experienced a **subacute onset of stimulus-induced myoclonus** of right hand and forearm with **fixed dystonic flexion-posture** of 4th–5th fingers → rapid spread to the whole hand → later involved ipsilateral lower face/tongue with dysarthria and severe chronic pain.

## INVESTIGATIONS

- Neurophysiology: giant SEPs (prominent N30); MEP: CMCT markedly prolonged; EEG unremarkable.
- CSF analyses shows oligoclonal bands; negative neuronal-antibody panel, neurodegeneration markers.
- MRI/DAT-SPECT/autoimmunity were unremarkable.
- 18F-FDG PET-CT: **parasternal nodal recurrence** → possible seronegative paraneoplastic syndrome
- February 2025: inconclusive reassessment with **persistent oligoclonal bands**; negative 3T brain PET–MRI and whole-body PET-CT; negative immunofluorescence on fixed cerebellum; genetic analysis pending.



## Treatments tested → Outcome

Immunotherapy: high-dose steroids **X**; IVIG **X**; PEX **X**

Symptomatic therapy included benzodiazepines, gabapentinoids, baclofen → **X**; anti-seizure medications (levetiracetam, piracetam, valproate, brivaracetam, perampanel) → transient and mild; zonisamide (ongoing). Levodopa, anticholinergics, tetrabenazine, risperidone → **X**

Oncology: endocrine therapy + palbociclib → metabolic remission over months but neurological progression

## NMDAR MODULATION

IV ketamine 0.5 mg/kg bolus (≈30 mg) → continuous infusion (0.3 mg/Kg/h) → marked reduction of myoclonus, discontinued due to adverse effects (nausea)

IN esketamine (ESK-IN) 84 mg (bioavailability ~45–50%, but higher NMDA affinity than racemate) → minimal benefit and practical constraints (once-/twice-weekly, supervised setting).

Watch the video



## DISCUSSION

The negative result with ESK-IN likely stems from route-specific pharmacokinetics. If the antimyoclonic effect is contingent on **dissociation-related network modulation**, then peak-dependent dynamics are critical. Despite comparable AUC, the reduced and delayed C<sub>max</sub> with IN administration likely blunted the dissociative state—and, consequently, the therapeutic effect—observed with IV ketamine.

## REFERENCES

1. Roze E, Apartis E, Clot F, et al. Myoclonus-dystonia: clinical and electrophysiologic pattern related to SGCE mutations. *Neurology*. 2008;70(13):1010-1016. doi:10.1212/01.wnl.00002297516.98574.c0
2. Latorre A, Belvisi D, Rothwell JC, Bhatia KP, Rocchi L. Rethinking the neurophysiological concept of cortical myoclonus. *Clin Neurophysiol*. 2023;156:125-139. doi:10.1016/j.clinph.2023.10.007
3. Quartarone A, Rizzo V, Terranova C, et al. Abnormal sensorimotor plasticity in organic but not in psychogenic dystonia. *Brain*. 2009;132(Pt 10):2871-2877. doi:10.1093/brain/awp213

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