

A Rare Presentation of Progressive Encephalomyelitis with Rigidity and Myoclonus in a Child: A Case Report

Jessica Beardsley, MD, and Angela Garcia, MD

Case Diagnosis

Progressive Encephalomyelitis with Rigidity and Myoclonus (PERM)

Case Description

A 6-year-old girl presented with four days of progressively worsening whole body spasms and rigidity, confusion, gait difficulties, and hyperekplexia. Labs were notable for elevated transaminases and CPK. CSF studies were notable for lymphocytic pleocytosis, elevated protein, and oligoclonal bands. Glycine receptor antibodies were found in the serum and CSF. EEG, MRI studies, and EMG were unremarkable. She was diagnosed with glycine receptor antibody positive PERM and treated with four doses of IVIG. Symptoms also improved with lorazepam trial; she was subsequently transitioned to clonazepam and baclofen. She showed significant clinical improvement in self-care and mobility at discharge but continued to demonstrate balance and cognitive deficits. She was discharged home with pediatric rehabilitation medicine follow up and outpatient PT, OT, and SLP.

Discussion

- Stiff-person syndrome (SPS) is a rare and progressive neurological autoimmune disorder resulting in muscular rigidity, painful spasms, gait difficulties, and falls.
- PERM is a severe variant of SPS further characterized by encephalopathy, myoclonus, brainstem dysfunction, hyperekplexia, dysautonomia, flare-ups, and remissions.
- PERM is a clinical disease. MRI, CSF, and EMG are typically normal. Continuous motor unit activity can be seen on EMG.
- Associated with anti-GAD antibodies, glycine receptor antibodies, DPPX antibodies, and NMDA receptor antibodies.
- Early diagnosis and treatment with corticosteroids, IVIG, PLEX, and or rituximab can result in dramatic clinical improvement. Benzodiazepines and baclofen are also utilized for symptomatic management.
- Rigidity and stiffness can lead to contractures of the affected joints, resulting in impairments in mobility and self-care.
- Rehabilitation should focus preserving range of motion and contracture prevention through medications, therapies, and bracing.

Conclusion

Children with PERM require prompt diagnosis, medical management, and comprehensive rehabilitation to address self-care, mobility, and cognitive deficits. The Pediatric Rehabilitation Medicine team plays an essential role in managing children with PERM given the disease's chronic, progressive, fluctuating, and recurring nature.

References

1. Baizabal-Carvallo JF and Jankovic J. Stiff-person syndrome: insights into a complex autoimmune disorder: *J Neurol Neurosurg Psychiatry*. 2015; 86:840.
2. Baker RA, Revesz T, Thom M, et al. Review of 23 patients affected by stiff man syndrome: clinical subdivision into stiff trunk (man) syndrome, stiff limb syndrome, and progressive encephalomyelitis with rigidity. *J Neurol Neurosurg Psychiatry*. 1998; 65:633.
3. Clardy SL, Lennon VA, Dalmau J, et al. Childhood onset of stiff-man syndrome. *JAMA Neurol*. 2013; 70:1531.
4. Crisp SJ, Balint B, and Vincent A. Redefining progressive encephalomyelitis with rigidity and myoclonus after the discovery of glycine antibodies. *Curr Opin Neuro*. 2017. 30(3):310-316.
5. Hutchinson M, Waters P, McHugh J, et al. Progressive encephalomyelitis, rigidity, and myoclonus: a novel glycine receptor antibody. *Neurology*. 2008; 71:1291.
6. Jazebi N, Rodrigo S, Gogia B, and Shawagfeh A. Anti-glutamic decarboxylase (GAD) positive cerebellar ataxia with transitioning to progressive encephalitis with rigidity and myoclonus (PERM), responsive to immunotherapy: A case report and review of the literature. *J Neuroimmunol*. 2019. 332:135-137.
7. Khan F, Chevidikunnan MF, Almalki RA, Mirdad MK, Nimatallah KA, Al-Zahrani S, Alshareef AA. Stiff-Person Syndrome Outpatient Rehabilitation: Case Report. *J Neurosci Rural Pract*. 2020 Oct;11(4):651-653. doi: 10.1055/s-0040-1715081. Epub 2020 Aug 20. PMID: 33144807; PMCID: PMC7595795.
8. Shaw PJ. Stiff-man syndrome and its variants. *Lancet*. 1999; 353:86.
9. Sturchio A, Gastaldi M, Princiotta Cariddi L, Biacchi D, Espay AJ, Franciotta D, Versino M, and Mauri M. Levodopa-responsive progressive encephalomyelitis with rigidity and myoclonus associated with glycine-receptor antibodies. *Parkinsonism Relat Disord*. 2021. 82:7-9.
10. Su Y, Cui L, Zhu M, Liang Y, and Zhang Y. Progressive Encephalomyelitis with Rigidity and Myoclonus with Thymoma: A Case Report and Literature Review. *Front Neurol*. 2020. <https://doi.org/10.3389/fneur.2020.01017>
11. Vaiyapuri A, Kashyap P, Kashyap N, Muthusamy H, Unnikrishnan R, Alqahtani M. Is Stiff Person Syndrome Benefited by Physical Therapy Intervention? Summary of Case Reports. *Biomed Res Int*. 2019 Mar 24;2019:5613680. doi: 10.1155/2019/5613680. PMID: 31019968; PMCID: PMC6451805.