

A Rare Case of Childhood Progressive Myoclonic Epilepsy

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ABSTRACT

Introduction: Progressive myoclonic epilepsies are characterized by myoclonus, tonic clonic seizures, progressive neurological deterioration, cerebellar signs and dementia. We reported a case of adolescent child with an exceedingly rare disorder.

Case Report: A child presented in our ward with dementia and severe myoclonus with all cerebellar signs. Child was on antiepileptics but no improvement showed in past. Our treatment reduced the myoclonus and improvement in speech and walking.

Conclusion: Progressive myoclonic epilepsy is very debilitating in nature with mostly poor outcome with progressive dementia and myoclonus with fatality at an early age.

Keywords: Myoclonus, Progressive, Genetic, Cerebellar Signs

INTRODUCTION

The approach to a child with progressive myoclonic epilepsy is a vast and challenging task as the establishment of differential diagnosis is very complicated and is a cause of concern for both the parents and the physician.^{1,2} Not only can the symptoms be debilitating but also development of myoclonus may be the harbinger of lifelong disability and can be fatal. Progressive myoclonic epilepsies are a group of symptomatic generalized epilepsies caused by rare disorders. Most of these disorders have a genetic component, a debilitating course and a poor outcome. Progressive myoclonic epilepsies are characterized by myoclonus, tonic clonic seizures, progressive neurological deterioration, cerebellar signs and dementia. Certain specific disorders comprise the most common causes of progressive myoclonic epilepsy:

- 1) Unverricht-Lundborg disease (Baltic myoclonus)
- 2) Myoclonic epilepsy with ragged red fibres (MERRF syndrome)
- 3) Lafora disease
- 4) Neuronal ceroid lipofuscinosis
- 5) Type I sialidosis.

Less common causes amongst specific disorders include dentato-rubro-pallido-luysian atrophy (DRPLA), the noninfantile neuronopathic form of Gaucher disease, and atypical inclusion body disease. PME has also been reported in Niemann-Pick disease type C. Most of the cases have been described from South India.

CASE REPORT

A 12 year old female child who was conscious and oriented, presented to OPD with myoclonic jerks. Clinical history revealed difficulty in walking for last 2 years, history of frequent falls for 1 and a half year and difficulty in speech for last 1 year. On clinical examination the patient had frequent myoclonic jerks with slurring of speech and presence of cer-

ebellar signs. Myoclonic jerks were initially 5-6 episodes per day which at the time of examination were every 5-10 minutes. There was history of frequent falls 1-2 per day but presently the patient could not even stand without support due to frequent myoclonic jerks. Speech was non rhythmic with scanning present but understanding was normal. There was h/o abnormal movements of generalized tonic clonic in nature in the past six months. On examination Tone was decreased in all four limbs and generalized wasting of muscles was seen. Power was 4/5 in all limbs, all the jerks were exaggerated with pendular jerks and knee clonus present. Sensory system was normal. All the cerebellar signs were present and coordination not present. Rest of the CNS examination was normal. All other systemic examination was normal. CSF examination was normal with sepsis screen clear. MRI (Figure-1) was normal but EEG (Figure - 2) showed generalized polyspike wave and high voltage spike and wave discharges reaching anteriorly.

Treatment outline at admission

Child was managed conservatively. Antiepileptic drugs such as sodium valproate, clonazepam, levetiracetam were given to the child. Physiotherapy as well as genetic counseling was provided to the patient. Patient showed gradual improvement and decreased frequency of myoclonic jerks on discharge and follow up.

DISCUSSION

Unverricht-Lundborg disease (ULD), or epilepsy progressive myoclonus type 1, is an autosomal-recessive disorder that was described by Unverricht in 1891², and by Lundborg in 1903³ It is one of the most common cause of PME. The age of symptom onset in ULD is 6–15⁶ years. Symptoms progress insidiously. Stimulus-sensitive myoclonic jerks are an essential feature for the diagnosis of the disease and are the first symptom in half the cases of ULD. Its incidence is 1 in 20,000 births.⁴ It is linked to chromosome 21q22.3. Further linkage disequilibrium and historical recombination breakpoint mapping placed the associated gene as *CSTB*. The main mutation is in *CSTB* gene. The exact pathophysiology of the disease remains unknown. *CSTB* encodes cystatin B, a cysteine protease inhibitor. Presumably, with mutations in cystatin B and loss of inhibition of cysteine proteases, apop-

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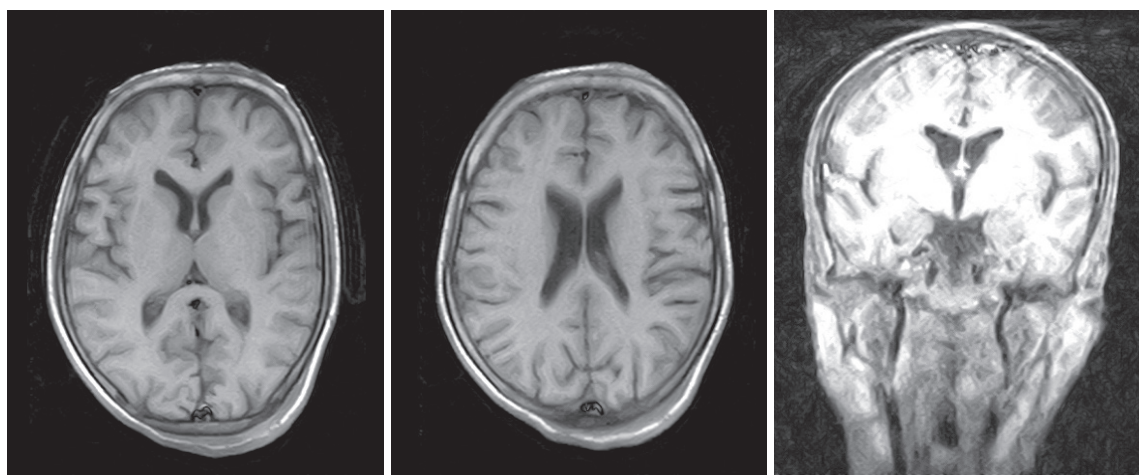


Figure-1: MRI of the patient

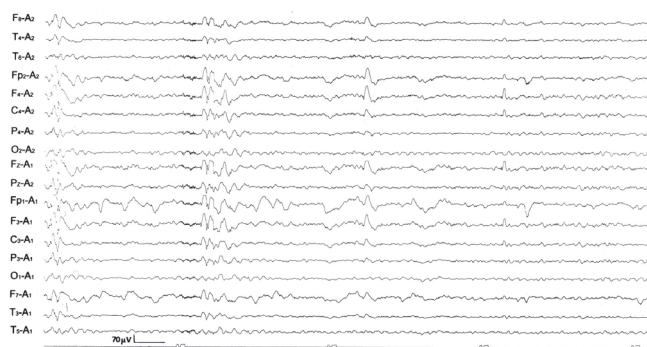


Figure-2: EEG of the patient

tosis proceeds abnormally and neurodegeneration develops. A multiprotein complex has been identified to interact with cystatin B in vitro. Immunofluorescent on focal microscopy showed that the same proteins are present in the granule cells of the developing cerebellum and the purkinje cells of adult rat cerebellum, which raises the possibility that a cystatin B multiprotein complex may have a specific cerebellar function.⁹ For diagnosis, clinical suspicion is mainstay. There is no definitive cure for the disease. Management is symptomatic with antiepileptic drugs and physiotherapy.

CONCLUSION

Based on history, relatively preserved cognition and vision, clinical examination and laboratory findings we came to the diagnosis of Unverricht-Lundborg Disease (ULD). ULD is one of the most common cause of PME (Progressive Myoclonic Epilepsy). Prevalance is 1 in 20,000. Age of onset is between 6 to 15 years. Stimulus sensitive myoclonic jerks are characteristic of this disease. Patient usually has ataxia, incoordination, intention tremor and dysarthria, but normal cognition.

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