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Sub acute Sclerosing Pan encephalitis in a child with early onset Huntington's Disease

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Abstract: Onset of Huntington's disease (HD) before 10 years constitutes less than 1% of all patients with HD. We report 5 year old boy with complex presentation having features of both Sub acute Sclerosing Pan encephalitis (SSPE) and early onset HD evaluated by history, clinical and neurologic examination, electroencephalography, cerebrospinal fluid examination and neuro imaging. The genetic workup confirmed the clinical diagnosis of HD, which makes this case as first case with SSPE and early onset HD reported in literature.
Keywords: Huntington's chorea Childhood Onset West phal Variant, Sub acute Sclerosing Pan encephalitis

INTRODUCTION

Huntington disease (HD) is a fatal neurodegenerative disorder usually presenting in the 4th–5th decade of life with motor dysfunction, cognitive decline, mood and personality changes, and is pathologically characterized by diffuse cerebral neuronal loss [1]. The critical mutation represents an expansion of a polymorphic $(CAG)^{n}$ repeat in the coding region of the huntingtin gene on chromosome 4. Early onset Huntington disease (EOHD), with an onset before 20 years of age, is estimated to account for only approximately 7% of all patients with HD, and patients for whom the age of onset is before 10 years constitute less than 1% of all patients with HD [2].

Sub acute Sclerosing pan encephalitis (SSPE) is a progressive inflammatory disorder of the central nervous system with both poor prognosis and high mortality. The disease has been related to a persistent and aberrant measles virus infection. We report a case of SSPE with atypical features including seizures at onset and a fulminant course in a 5 years-old boy with EOHD**.**

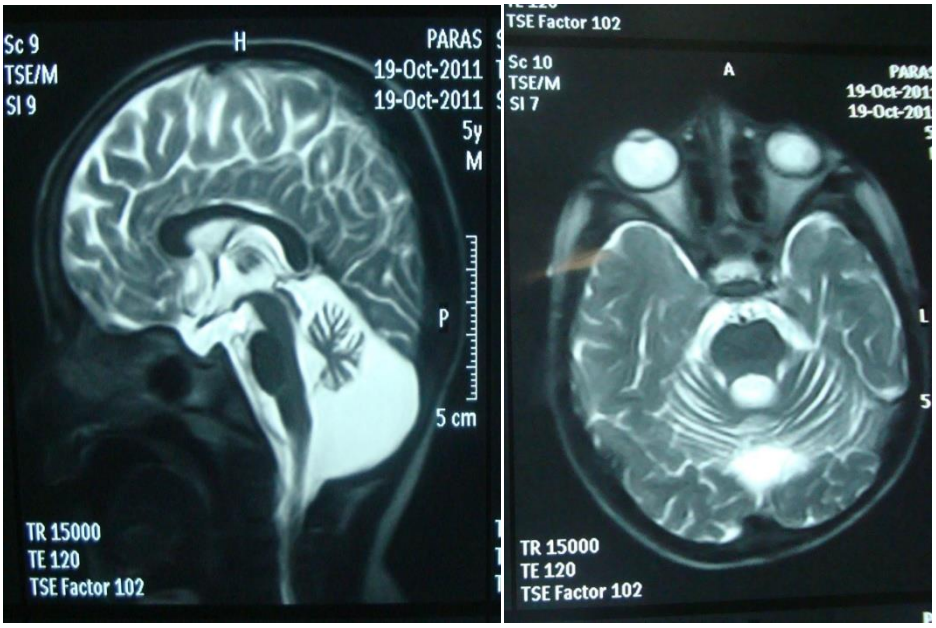
CASE REPORT

P, a 5 year old boy was referred to pediatric neurology clinic of our hospital with chief complaints of seizures, altered speech and involuntary movements noted for last 4 months. His birth and development was normal until 4 months back when he had multiple episodes of unprovoked generalized tonic seizures. He was started on antiepileptic therapy (phenytoin) by a local doctor with no relief in symptoms. Gradually the child was also noted to have developed jerky movements of neck and limbs with frequent falls along with speech impairment. In addition the child also developed involuntary movements which the mother recognized as being similar to that in his father and late grandmother.

His father had been evaluated in neurology clinic of another tertiary care hospital in the city 8 years back and was suspected to have Huntington's chorea. Genetic testing was not performed due to financial restraints. His condition had further worsened and was bed-ridden for last 1 year. Grandmother of this child also had similar involuntary movements and had expired at the age of 52 years. Similar presentation in the child and that too at such an early age forced the mother to seek medical attention.

On examination, the child was noted to be alert interictally. He knew his name, would greet the doctors but could follow only simple commands. Subtle cognitive deterioration was evident on detailed history taking. He had dystonic posturing of neck along with frequent myoclonic jerks in addition to choreiform movements. Hearing and vision were essentially normal. No focal neurological deficit was present and rest of systemic examination including fundus examination was normal. Provisional diagnosis of grey matter degenerative brain disease was considered and child evaluated further. Based on strongly positive family history, a possibility of early onset Huntington's chorea was considered, sample for molecular analysis for HD sent. Routine hematological and biochemistry parameters were all normal. Peripheral blood film for vacuolated lymphocytes and acanthocytes didn't reveal

any abnormality. Skin biopsy was normal and didn't reveal any inclusions. Visual evoked potential depicted normal amplitude potentials. BERA evaluation and chest X-ray were also normal. Electroencephalogram (EEG) revealed stereotyped, generalized and synchronous high amplitude periodic (every 6-8 minutes) bursts of polyspikes and spike and wave complexes. MRI brain revealed cerebellar atrophy (Figure 1). Possibility of SSPE was also considered due to rapid clinical deterioration along with myoclonic jerks, characteristic EEG pattern and history of measles at 2 years of age. CSF analysis done revealed clear fluid with protein 30 mg/dL and sugar 56 mg/dL. The value of serum measles-specific IgG antibody CSQ rel (relative CSF/serum quotient) was 5. Molecular genetic analysis of the pathogenic $(CAG)^{n}$ repeat region of the Huntington Disease gene revealed one normal and one abnormal sized (> 80 repeats) allele, characteristic of Huntington's disease. Presence of expanded allele was confirmed by triplet repeat primed PCR (TP PCR). Seizures and myoclonic jerks responded to carbamazepine and involuntary movements decreased with haloperidol. No therapy for SSPE could be started due to financial restrictions. Child expired after 6 months.



A B

Fig 1: Sagittal (A) and coronal (B) magnetic resonance imaging showing severely reduced cerebellar volume in T2-weighted sequences at the age of 5 years.

DISCUSSION

To our knowledge, this is the first case report of SSPE in child with EOHD. We wish to emphasize that despite clear-cut evidence of familial neurodegenerative disorder with autosomal dominant inheritance pattern, holistic work up needs to be carried out to look for associated conditions as seen in this case.

This child presented with generalized tonic seizures to begin with followed by progressive myoclonic epilepsy along with cognitive decline. In addition, he had extra pyramidal involvement as well manifesting as dystonia and choreiform movements. Differentials to be considered in such a case include SSPE, early onset Lafora body disease, late-infantile neuronal ceroid lipo fuscinoses, Unver richt-Lundborg disease, myoclonic epilepsy with ragged-red fiber (MERRF) syndrome, early onset huntington disease, wilson disease, sialidoses, acantocytosis and dentato-rubro-pallidal atrophy.

This child had symptoms common to both EOHD as well as SSPE. Clinical features EOHD remarkably differ from those of adult-onset HD and are characterized by rigidity, oral motor dysfunction, ataxic gait, behavioral disturbance and seizures [3, 4]. Juvenile Huntington's disease has also been reported to present as progressive myoclonic epilepsy [5]**.** The usual initial symptoms of SSPE include intellectual decline and personality changes. Myoclonic jerks usually follow the mental decline. Atypical forms of presentation of SSPE such as generalized seizures as initial symptoms have been reported rarely [6]. MRI brain showed cerebellar atrophy which has been reported in EOHD as well as SSPE [7, 8].

Despite a clear-cut evidence of familial neurodegenerative disorder with autosomal dominant inheritance pattern in this child pointing towards possibility of EOHD in this child, detailed evaluation revealed presence of SSPE, a potentially treatable condition, based on strong clinical suspicion and later

confirmation by characteristic EEG and CSF measles antibody titer analysis. To summarize, complete evaluation of any familial neurodegenerative disorder is a must to diagnose other associated and at times, treatable conditions.

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