

neck muscles and upper torso) may be attributable to early manifestations of MND. These findings were also supported by EMG which also showed decreased recruitment and presence of spontaneous fibrillation potentials and fasciculations. These findings were not present in other affected members.

Although, the exact incidence of amyotrophy is not known but few case reports has shown association of MND with SCA2.⁴ The pathophysiological basis for this association was pointed out by Elden et al.⁵ who concluded that ATXN2 gene as a relatively common suitability gene to Amyotrophic lateral sclerosis (ALS). They demonstrated that ATXN2 is a potent modifier of TAR DNA binding protein (TDP-43) toxicity in animal and cellular models. In addition, 6 patients with ALS were evaluated and disclosed different ATXN2 localization in spinal cord. In 2006, the 43-kDa (TDP-43) was identified as the major disease protein in ALS and frontotemporal lobar degeneration with ubiquitinated inclusions⁶. Thus our case report highlights the possibility of association of other neurodegenerative disorders like MND in the patients of SCA2. So, genetic testing for dominant ataxias should be included in the evaluation of patients with ataxia, especially in cases with a positive family history for spinocerebellar ataxia. New gene locuses which are linked to SCA's, are discovered every other day. Genetic study is significant in defining SCA types which are common in our country.

CONCLUSIONS

Although SCA 2 is the most common SCA in India, due to the multi ethnicity and multi religious pattern of society other uncommon SCAs may also be detected in presently unreported populations of the country. The clinical phenotype suggestive of a particular SCA type may sometimes vary even in a same family despite having same genotype. Even though it is not possible to establish a definite correlation between amyotrophy suggestive of MND and SCA2, it seems important to monitor the occurrence of these rare cases for better understanding of this combined multiple system neurodegenerative disorder.

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Recurrent Acute Disseminated Encephalomyelitis in a Child.

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Abstract : Acute disseminated encephalomyelitis (ADEM) is a non-vasculitic inflammatory demyelinating condition, which is usually monophasic. Recurrent ADEM is a much less characterized entity and its differentiation from multiple sclerosis (MS) poses a diagnostic challenge. We report a five year old girl with recurrence of ADEM after 15 months and discuss the diagnostic issues involved.

INTRODUCTION

Acute disseminated encephalomyelitis (ADEM) is an uncommon multifocal idiopathic inflammatory myelinopathy of CNS of pre pubertal children, typically occurring one to three weeks after a clearly identifiable febrile prodromal illness of viral infections and occasionally bacterial infections or immunization¹⁻³. It is usually Monophasic, Multiphasic or Recurrent Acute Disseminated Encephalomyelitis (RDEM). Recurrent ADEM is a much less characterized entity and its differentiation from multiple sclerosis (MS) is a diagnostic challenge²⁻⁵. The distinction between these two entities is important

because ADEM has good long-term prognosis over MS Most patients of ADEM recover fully following treatment with steroids or immunoglobulins⁴. The diagnosis is difficult due to insensitivity of CT imaging and lack of pathognomic clinical and laboratory features. We present a case of 5years old female child who presented to us with complaints of vomiting followed by weakness of right side of the body, difficulty in walking and swallowing after an episode of chickenpox. Child also had similar complaints one year back for which she was admitted in hospital.

CASE REPORT

A 5 year old girl belonging to average socioeconomic status presented in July 2014 with history of fever, cough, malaise and occasional vomiting. There was also history of difficulty in walking and swallowing. There was no history of seizures, rash or recent vaccination. The child had similar history in past for which she was hospitalized in April 2013. When we reviewed her old records this girl had presented with fever, cough, headache, fatigue and vague body ache of 5 days duration followed by progressively increasing drowsiness. At that time also there was no history of seizures, rash or recent vaccination. The sensorium was altered (GCS 10/15); cranial nerve examination was normal and there were no focal motor or sensory signs. Deep tendon reflexes were depressed with bilateral extensor planter

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response. There were no meningeal signs. The systemic examination was unremarkable. Blood counts and biochemical parameters were within normal range. Her cerebrospinal fluid (CSF) profile was normal. MRI brain revealed hyperintense signals on T2 and FLAIR sequences over bilateral subcortical white matter and cerebral cortices; they also involved basal ganglia, pons, midbrain, thalami, cerebellar peduncles and cerebellar white matter. She was treated with intravenous methyl prednisolone for 5 days. She had complete neurological recovery within a week. Her follow-up MRI done four months later showed complete disappearance of the abnormalities seen in the first MRI.

This time on examination vitals were maintained. General physical examination did not reveal anything abnormal. Her neurological examination revealed altered sensorium with Glasgow Coma Scale of 9/15, left gaze palsy, right hemiparesis, bilateral brisk deep tendon reflexes and extensor plantar response. The papillary response and fundi were normal. There were no meningeal signs. Other systems revealed no abnormality. Her hemogram showed WBC count of 11,000 with 65% polymorphs. The CSF was normal. Investigations for systemic diseases including LFT, RFT, blood and urine cultures, malarial parasite, Montoux, ECG, chest X-ray were within normal limits. CT scan brain was normal. MRI brain revealed multiple focal hyperintensities in left inferior and middle cerebellar peduncle, dorsal medulla, Pons, ventromedial thalami (L>R) and bilateral sub cortical and deep white matter suggestive of demyelination as depicted in figures 1 and 2.



Figure 1



Figure 2

Fig 1,2 showing MRI brain revealed multiple focal hyper intensities in left inferior and middle cerebellar peduncle, dorsal medulla, Pons, ventromedial thalami (L>R) and bilateral sub cortical and deep white matter suggestive of Demyelination.

MRI brain revealed hyperintense lesions on T2 and FLAIR at the same location as were observed in the first episode. She was investigated for causes of recurrent encephalopathy. Her CSF and blood lactate levels were normal as was urine and plasma amino-acidogram. Her tests for autoimmune diseases (LE cell, RA factor, ANA, dsDNA and lupus anticoagulant) were negative. Serological tests for the presence of HSV 1 & 2 and Japanese B in CSF yielded negative results.

She was diagnosed as having recurrence of ADEM. Patient was given symptomatic treatment. She was also given intravenous methyl prednisolone in dose of 20 mg/kg/day for 5 days followed by 2 mg/kg /day for another 5 days. Child showed improvement after a weeks time. She was discharged on oral methyl prednisolone in dose of 2 mg/ kg /day for 3 weeks and was advised to taper the steroids after one week.

On follow up examination after 1month child was asymptomatic. There was no weakness of right upper and lower limb and no difficulty in swallowing and walking. The patient was asked for regular follow up.

Her follow-up MRI three months later showed significant reduction in the number and size of the lesions.

DISCUSSION

The clinical features in our case were indicative of recurrent encephalitis with extensive lesions involving subcortical white matter, cerebral cortex, basal ganglia, midbrain, pons, cerebellum and thalami. The characteristic features of this case were the reappearance of symptoms and the lesions in the MRI that were at the same location after 15 months of interval with normal imaging in between. Such recurrent CNS symptoms raise the possibilities of MS or systemic disease with CNS involvement such as collagen vascular disease. Our case neither had any systemic features nor autoantibodies that suggested collagen vascular disease. MS becomes the second differential to ADEM and at times it is difficult to distinguish these two diseases,

particularly in relapsing cases ADEM is marked by an acute multifocal inflammatory myelinopathy of CNS, which is generally but not exclusively monophasic^{4,8}. Typically it is antedated by an infectious illness, most commonly measles, mumps, influenza A or B, hepatitis A or B or infection with herpes simplex, human herpes virus 6, varicella, rubella, mycoplasma, chlamydia, legionella or streptococci, rabies, diphtheria or Japanese B encephalitis; rarely malaria or following vaccination over days, weeks or months, punctuated by an acute worsening. Perivenular inflammation (with mainly of T lymphocytes) and demyelination is the hallmark of the disease^{4,7}.

The diagnosis of ADEM is difficult to confirm, because definite laboratory method, or even markers suggesting ADEM have not been established. The predominant white matter involvement suggests demyelination. Characteristic clinical features include sudden onset of multifocal neurological disturbances such as aphasia, motor and sensory deficits, signs of meningoencephalitis and depressed level of consciousness⁸⁻¹⁰. MRI abnormalities are most frequently identified on T2-weighted and FLAIR sequences as patchy, poorly marginated areas of increased signal intensity. Lesions in ADEM are typically large, multiple, and asymmetric. They typically involve the sub cortical and central white matter and cortical gray-white junction of cerebral hemispheres, cerebellum, brainstem, and spinal cord. The gray matter of the thalami and basal ganglia are frequently involved, typically in a symmetric pattern. The periventricular white matter is also frequently involved. Lesions confined to the corpus callosum are less common⁸. Though MRI is the most widely applied diagnostic tool for ADEM, but no MRI criteria have been identified specific for ADEM⁸⁻¹⁰. The MRI findings that strongly suggest ADEM over MS are bilateral, asymmetric, involvement of cerebral white matter along with involvement of basal ganglia, thalamus, cerebellum and sparing of corpus callosum and periventricular region⁶. However, the confusion between recurrent ADEM and MS still persists despite various published reports on recurrent ADEM illustrating characteristic MRI findings¹⁰. Based on its immune mediated etiology, ADEM is commonly treated with high-dose steroids and intravenous immunoglobulin. Plasmapheresis and cytostatic drugs are alternative treatment options in patients who do not respond to steroid and/or to intravenous immunoglobulin^{5,7}.

ADEM can mimic many other neurological diseases. Multifocal white matter demyelination which is best visualized on MRI is characteristic of this disorder. Methylprednisolone may improve outcome though some cases may remit spontaneously.

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