succumbed with MRI suggestive of hyperintensiles in thalamus and basal ganglia, the other responded to aggressive immunotherapy without any complications.

Elizabeth, et al. [4] reported three cases of JDM who developed generalized tonic clonic seizures two weeks after the initiation of immunosuppressive therapy. Out of the three, two patients had cerebral vasculopathy showing prominent fontal blood vessels on CT angiogram. One patient had assymetric perfusions in both hemispheres whereas the other had multiple infarctions in MRI brain.

The vascular pathology in JDM is not a true vasculitis. It is limited to small arterioles and capillaries demonstrating fibrinoid necrosis on biopsy. Seizures in JDM can be due to vasculopathy, true cerebral vasculitis of small to medium sized vessels, hypoxic ischemic encephalopathy, hypertensive encephalopathy, cyclosporine induced encephalopathy or secondary infections. In our child, the cause of seizures was attributable to CNS vasculopathy as supported by MRI changes. Although MR angiography provides supportive evidence of CNS vasculopathy, but it has a low diagnostic detection rate [2]. Hence MRI angiogram being normal in our case does not rule out the possibility of CNS vasculopathy. Though biopsy is the gold standard, but considering the patient's moribund condition, it was not done.

Most centers treat severe JDM initiate treatment using a combination of IV methylprednisolone and methotrexate, with addition of IVIg. However, treatment of refractory disease remains controversial with limited reports on the use of cyclophosphamide [5], and rituximab [6]. Our patient was steroid and IVIg refractory, and finally showed response to a combination of cyclophosphamide pulses along with rituximab, and remains asymptomatic on follow up.

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Congenital Neuronal Ceroid Lipofuscinosis: An Important Cause of Unexplained Seizures in Newborns

Neuronal ceroid lipofuscinoses (NCL or CLN) are the largest group of neurodegenerative diseases in childhood. The overall incidence is 1 in 12,500 live births [1]. There are 13 known types (CLN 1-13) based on age of onset and genetic mutation involved. Among these, CLN type 10 can present as neonatal or juvenile phenotype. It is caused by homozygous or compound heterozygous mutation in the *Cathepsin D* gene (*CTSD*) on chromosome 11p15. Neonatal onset phenotype is rare and has a poor outcome with rapid deterioration. We report a case of neonatal onset CLN 10.

A male baby delivered vaginally at 34-36 weeks of gestation, birth weight of 2200 g, was well till day 26 of life when he developed seizures, lethargy and breathing difficulty. There was history of third degree consanguinity and the previous sibling had microcephaly, who had died at day 15 of life with similar complaints. On examination, our patient was lethargic with dysmorphic facies, microcephaly (head circumference 29 cm) and hepatosplenomegaly. Sepsis workup was normal. The seizures were refractory to multiple antiepileptic drugs like phenobarbitone, phenytoin, leveteracitam, clonazepam and pyridoxine. Arterial blood gas, serum ammonia, uric acid and EEG were normal. Fundus revealed a cherry red spot. Lysosomal storage disease panel (Gauchers, Neimann-Pick type A and B, Krabbe, Pompe, Hurler, Fabry) was sent, which was non-contributory. MRI brain showed cerebral and cerebellar atrophy with diffuse, thinning of cerebral cortex, shallow sulcal spaces suggestive of microcephaly, with simplified gyral pattern. Whole exome sequencing (WES) showed a missense variant NM_001909.5 (*CTSD*-Catepsin D gene):c.299C>T (p.Ser100Phe) causing amino acid substitution from serine to phenylalanine at codon 100 in exon 3 of the *CTSD* gene. The diagnosis of NCL type 10 was made. According to the American College of Medical Genetics and Genomics (ACMG) classification, this mutation was a likely pathogenic variant. The patient was discharged but succumbed to seizures on day 80 day of life. Genetic counseling was offered to the family for subsequent pregnancies.

Till date, five cases of neonatal onset phenotype of CLN 10 with microcephaly have been reported. Three babies carried homozygous mutations in *cathepsin D*, a gene coding for a lysosomal aspartic protease and succumbed within first 10 days of life. Some affected babies also had intrauterine seizures that were not present in the index case. CLN 10 should be kept as a differential diagnosis in neonates with seizures and microcephaly

[2]. The index case and his siblings, both had microcephaly. Two neonates (brother and sister) with CLN 10 presenting with intractable seizures after birth have been similarly reported [3]. A female baby with CLN 10 presented with severe microcephaly and hypertonia, with MRI showing generalized hypoplasia of the cerebral and cerebellar hemispheres and expired on day 2 of life [4]. Postmortem examination revealed a small, firm brain with extensive neuronal loss and gliosis and an identical mutation as the index case. Another term infant with microcephaly, and status epilepticus, who died 36 hours later, is described [5]. At autopsy, atrophic brain with microscopic changes consistent with NCL were reported [5].

To conclude, NCL should be suspected in newborns and infants presenting with unexplained microcephaly and intractable seizures with radiological features of cerebral and cerebellar atrophy. Genetic work up and counseling helps to offer prenatal diagnosis.

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