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VANISHING WHITE MATTER DISEASE WITH CENTRAL HYPOMYELINATION

Pediatrics

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ABSTRACT

Vanishing white matter disease (VWM) is the most prevalent inherited leucoencephalopathies in children with central hypomyelination. The classical phenotype is characterized by early onset of chronic neurological deterioration, dominated by cerebellar ataxia. The phenotypic variation is extremely wide from antenatal in onset type with early demise to adult in onset with slowly progressive disease. The basic defect is in one of the five sub-units of eIF2Binitiation factor. Here we report a case of 80 days old male infant with decreased weight gain, stiffness of all four limbs, seizures following head trauma in whom MRI brain identified T2 hyperintensities in white matter of fronto-temporo-parieto-occipital regions and on DWI, it showed abnormal white matter with increased diffusivity reflecting the rarefaction and cystic degenerations. Hence diagnosis of VWM disease was established.

KEYWORDS

Vanishing white matter disease,Cerebellar ataxia, Central hypomyelination, Leucoencephalopathy.

INTRODUCTION:

VWM disease is also called as childhood ataxia with central hypomyelination. In the recent years, there is evidence that this disease has extremely wide phenotypic variation and affects all ages. Characteristic feature of this disease is rapid and major neurological deterioration provoked by stress such as fever, minor head trauma and acute fright.Recently, undue activation of the unfolded-protein response has emerged as important entity in the pathophysiology of VWM.

CASE REPORT:

An 80 days old male infant, 1st born to non-consanguineous parentage, presented with decreased weight gain and stiffness of all four limbs since birth with two episodes of seizures following a minor head injury. Antenatally there is a history of decreased fetal movements and oligohydramnios.Clinical examination and anthropometryrevealed his weight, length andhead circumference were less than 3rd percentile as per WHO growth charts. Child has not attained social smile, hypertonia in all four limbs with scissoring of lower limbs, fundoscopy showed Bilateral optic atrophy. Child was treated with antiepileptics, neuro imaging was done which revealed bilateral fronto-temporo-occipito-parietal T_2 hyperintensities with T_1hypointensities which were diffuse in nature associated with cystic degeneration which establishes the diagnosis of VWM disease.

DISCUSSION:

The classical phenotype is more common in 2-6 years of age. The disease mayhas it's onset in antenatal periodor early infancy period. Cree encephalopathy is a severe variant of VWM disease. More severe variants may present antenatally with decreased fetal movements, oligohydramnios in 3rd trimester of pregnancy,in the postnatal period with growth failure, microcephaly, feeding problems, failure to thrive, limb hypertonia, seizures as observed in this case. Respiratory failure may also occur which is the leading cause of deathin severe cases. MRI brain is usually diagnostic in VWM disease. In some cases, biochemical markers like cerebrospinal fluid glycine is elevated while asialotransferrin is decreased, but it lacks sensitivity and specificity.

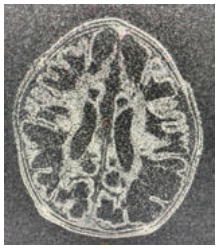

Figure 1: T1 Axial section image showing diffuse hypointensities

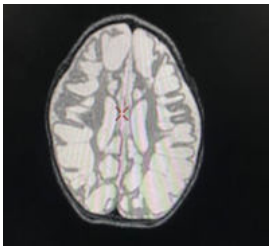

Figure 2: T2 Axial section image showing diffuse hyperintensities

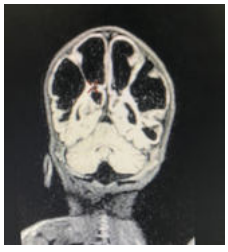

Figure 3: Flair image of coronal section showing diffuse cystic degeneration of white matter

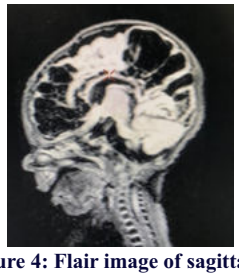

Figure 4: Flair image of sagittal section showing diffuse cystic degeneration of white matter

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