Introduction:
Vanishing white matter disease (VWM) is one of the most prevalent inherited childhood leucoencephalopathies. The disease is variably called Myelinopathia Centrallis Diffusa. Childhood ataxia and diffuse central nervous system hypomyelination are the common findings. The disease is characterized by chronic progressive and episodic deterioration with ataxia, spasticity and optic atrophy.\(^1\) VWM is caused by mutation in any of the five genes encoding the subunits of eukaryotic translation initiation factor eIF2B.\(^2,3\) The disease has an autosomal recessive mode of inheritance. The cause of the disease is unknown. Previously it was known that there is no biochemical marker for this disease,\(^4\) but recently analysis of body fluids has revealed only a few biochemical markers for VWM. The first marker found was a consistent elevation of cerebrospinal fluid glycine concentrations with an elevated ratio of cerebrospinal fluid to plasma glycine concentrations.\(^5\) A decreased cerebrospinal fluid concentration of asialotransferrin is a recently identified biomarker for VWM.\(^6,7\)

Case Report:
Rudru, eight months old male child, first issue of non-consanguineous parents, hailing from Dhaka presented with sudden jerky movement of single limb associated with stiffness of whole body for one month and fifteen days which had been gradually increasing and there was developmental deterioration for three months. He had two episodes of fever and maculo-papular rash four months back. But there was no history of head trauma or contact with TB patients. His birth history was uneventful and he was immunized as per EPI schedule. He was on complementary feeding.

On examination the child was ill looking, mildly pale, BCG mark was present. His weight was 8 kg, supine length was 73 cm (within normal limit), and OFC was 42 cm (below 5\(^{th}\) percentile). His motor functions, bulk,

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power was normal but tone was increased. All reflexes of both upper and lower limbs were exaggerated, plantar was extensor and ankle clonus was also present bilaterally. Coordination and gait could not be elicited. His sensory functions and cranial nerve revealed intact. He was developmentally delayed as he was not able to sit. Other system revealed normal findings. His blood picture was normal. MRI of brain showed an abnormal signal of cerebral white matter, U fibers were spared, rarefaction and cystic degeneration of white matter. Considering history, clinical examinations and investigation reports this case was diagnosed as a case of Vanishing white matter (VWM) disease. There is no specific treatment for VWM; counseling was done with the parents regarding the etiology, progression and outcome of the disease. Advice was given to avoid stress situations and head trauma and to take liberal use of antibiotics, antipyretics and vaccination. He was also advised regular follow-up for progression of the condition.

Discussion:
The classical and most common variant of Vanishing white matter disease has its onset in childhood, at age 2-6 years, though this disease may have an early infantile or antenatal onset that had happened in our patient. The first time this disease was documented in 1962 when Eickle studied a 36 year old woman. In 1993-94, Dr. Hanefeld and Dr. Schiffmann and their colleagues identified the disease as childhood-onset progressive leukoencephalopathy. It is characterized by chronic progressive neurological deterioration with cerebellar ataxia, usually less prominent spasticity and relatively mild mental decline. Epilepsy is common. Characteristically, there are additional episodes of major and rapid deterioration following minor head trauma and especially febrile infections. In our case the patient had a chronic progressive neurological abnormal sign as a jerky movement, but he had no epilepsy. His mental function and ataxia could not be elicited due to too younger age. Optic atrophy with loss of vision may occur, but that was not present in our patient. The baby had not a history of acute frightening that recently has been reported as another provoking factor. MRI of the brain is usually diagnostic in VWM. It shows an abnormal signal of all or almost all cerebral white matter with relatively spared U-fibers in some cases and progressive rarefaction and cystic degeneration of the affected white matter that is replaced by fluid. These features are almost consistent with the MRI of our case. No test was done to identify biomedical marker relevant to this disease.

Conclusion:
Vanishing white matter disease is an intriguing disease, both from a clinical and molecular perspective. There is no specific treatment for VWM. Avoidance of stress situations known to provoke deterioration in VWM patients is essential. Liberal use of antibiotics and antipyretics, vaccinations, and abstinence of contact sports are simple but important measures. However, they are not sufficient to prevent onset or progression of the disease. The most important consequence of research findings of the last 5 years probably is that prenatal diagnosis has become available for families as soon as the disease-causing mutations in the index patient have been identified.

References:


