Niemann-Pick Disease Type A Presenting as Unilateral Tremors

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Niemann-Pick group of diseases are rare lysosomal storage disorders. The clinical phenotype is variable. We report a child who first time presented with tremors of tongue and tremors of one side of the body. On examination child had hemiparesis and hepatosplenomegaly. Bone marrow examination shows storage cells suggestive of Niemann-Pick cells and enzyme assay confirmed the diagnosis.

Key words: *Cherry red spot, Niemann-Pick disease type A, Unilateral tremors.*

Niemann-Pick disease is a heterogeneous group of autosomal recessive lysosomal lipid storage disorders, characterized by hepatosplenomegaly and sphingomyelin storage in reticuloendothelial and parenchymal tissues, with or without neurological involvement [1].

CASE REPORT

We report a 9-month-old girl born to a second degree consanguineously married couple at term by normal delivery. She presented with tremors of the tongue and tremors of right upper and lower limbs of 35 days duration, followed by shaking of head and trunk of 20 days duration. Parents noticed paucity of movements of right upper and lower limbs since the onset of shaking. Child has also lost acquired milestones. She used to sit with support, transfer objects from one hand to other, attained stranger anxiety, used to respond for calling her name, and used to speak bisyllables. After the onset of tremors child lost head control, sitting, holding objects and social smile. Child had exaggerated startle response.

On examination, child's weight, length and head circumference were within normal limits. There was protrusion of tongue, upward deviation of eyeballs and rigidity with weakness of right upper and lower limbs. Resting tremors of the tongue, right upper and lower limbs were present. Head titubation and truncal ataxia were noted. Cherry red spot was not present. Liver was palpable 4 cm below the right costal margin and span of 8 cm with firm consistency. Spleen was palpable 4 cm below the costal margin and was firm in consistency.

Investigations showed, hemoglobin - 9.9 g/dL, total leucocyte count - 2400 cells/μL, platelets -2,43,000/μL. Liver functions, renal functions, serum electrolytes and lipid profile were normal. Ultrasonography of abdomen shows moderately enlarged liver and spleen with normal echo-texture, and multiple mesenteric lymph nodes. Bone marrow aspiration showed normocellular marrow, with storage cells suggestive of Niemann-Pick disease. MRI of brain was not done as bone marrow aspiration was suggestive of Niemann-Pick cells. Acid sphingomyelinase activity in leucocytes showed less than 10% of mean normal activity.

DISCUSSION

Niemann-Pick disease type A initially presents with non-neurological manifestations. The first symptom is hepatosplenomegaly, usually noted by the age of three months; over time the liver and spleen become massive [2]. Usually neurological examination at the time of presentation can be normal. Psychomotor development progresses no further than 12-month level, after which neurological deterioration is relentless. The neurological findings can include cerebellar signs, nystagmus, extra pyramidal involvement, intellectual disability, and psychiatric disorders [2]. A classic cherry-red spot, which may not be present in the first few months, is eventually present in all affected children. Interstitial lung disease results in frequent respiratory infections and often respiratory failure can be present. Most children succumb before the third year.

This child presented for the first time with neurological features in the form of tremors of one side of the body followed by weakness of same side, with regression of milestones. Neurologically, child had

rigidity of right side with tremors of tongue and right half of the body with weakness. Initially we suspected space occupying lesion in the brain, as child had predominant unilateral signs. Investigations shows decreased total counts with mesenteric lymph nodes with hepato-splenomegaly on ultrasonography of abdomen. CT scan of brain was normal. Bone marrow examination shows classic Niemann-Pick cells. Acid sphingomyelinase assay confirms our diagnosis of Niemann-Pick disease Type A. Prenatal diagnosis can be made by assay of acid sphingomyelinase activity in cultured chorionic villi or amniocytes [3].

Unilateral involvement with tremors in the initial stage, absence of cherry red spot despite neurological involvement, absence of lung involvement are unusual presentations. There is no literature available on this kind of unusual presentation.

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Diabetic Ketoacidosis Following Mumps

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A-13-year-old girl presented with diabetic ketoacidosis with convincing clinical signs of parotitis (fever, drooling of saliva, inability to swallow with development of bilateral parotid swelling) and pancreatitis (fever, abdominal pain and vomiting), along with high serum amylase and positive mumps IgM titer. This suggests that mumps virus may have been the causative factor, probably as a result of concomitant involvement of the pancreas.

Key words: *Diabetic ketoacidosis, Mumps.*

The common complications of mumps are meningitis with or without encephalitis, gonadal involvement and myocarditis while pancreatitis is an uncommon complication of mumps. Epidemiological studies have suggested that mumps pancreatitis may be associated with subsequent development of diabetes mellitus, but a causal link has not been established. The following is a report of a case of diabetic ketoacidosis (DKA), apparently due to mumps pancreatitis, with involvement of parotid glands.

CASE REPORT

A-13-year-old girl, developed low-grade fever, drooling of saliva and vague abdominal pain. Next day, it progressed to inability to swallow and speak, and vomiting. Two days later, she had twitching of face and generalized convulsions and became unconscious. She was then admitted to our institution.

She was drowsy but oriented and following verbal commands. Moderate dehydration was present. Her temperature was 101ºF, pupils were bilateral normal size and reacting and Glasgow coma score was 15. Her cardiac examination was normal. The abdomen was soft and non-tender. Liver was 2 cm below costal margin, soft, smooth and non-tender. Spleen was not palpable. On central nervous system examination, there was no focal neurological deficit or signs of meningeal irritation.

Investigations revealed high random blood sugar values (462 mg/100mL), urine was postive for sugar and ketone, and blood gas analysis revealed metabolic acidosis. She was diagnosed as a case of diabetic ketoacidosis and treated with intravenous fluids, regular insulin and other supportive treatment. Hyperglycemia was controlled within 24 hours. On the third day of admission, she developed swelling of right parotid gland followed by involvement of the left parotid gland following day. Serum amylase was 918 U/L. USG abdomen showed. hepatomegaly and bilateral enlarged