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Case Report

Rare presentation of mucopolysaccharidosis with neuro-leptospirosis

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ABSTRACT

In this case report, we present a 10-year-old boy who was a known case of Hunters disease who presented with complaints of fever, vomiting, altered sensorium for 2 days. On examination he was febrile and had moderate hepatosplenomegaly. Lab investigation showed anaemia with thrombocytopenia and fever workup turned positive for leptospirosis. In view of altered sensorium, MRI brain was done which revealed a large subdural hematoma with midline shift which required parietal craniectomy with evacuation. From this case study, we reported that in a patient with underlying lysosomal storage disorder, vasculitis secondary to endothelial damage by leptospirosis along with thrombocytopenia can cause severe intracranial bleeding even with higher platelet counts.

Keywords: Mucopolysaccharidosis, Neuro-leptospirosis, Hunters disease

INTRODUCTION

Leptospirosis is a zoonotic infection with worldwide distribution which is mostly prevalent in tropical and sub-tropical countries like India with disease burden disproportionately affecting resource-poor population.[1] Leptospires survive for days to weeks in warm and damp environmental conditions including water and moist soil. Leptospires infect many species of animals including rats, livestock and domestic dogs.

Leptospires enter human hosts through mucous membranes primarily eyes, nose, mouth and trans-dermally through abraded skin or by ingestion of contaminated water. After penetration they circulate in bloodstream causing endothelial damage of small blood vessel affecting the end organs. Transient thrombocytopenia occurs in more than 50% cases but haemorrhagic manifestation occurs rarely.[2] Here we reported a rare case of leptospirosis with mucopolysaccharidosis who presented with severe intracranial bleed.

CASE REPORT

A 10-year-old boy, second born of non-consanguineous marriage who is a known case of Hunter syndrome (type II mucopolysaccharidosis), presented with fever, vomiting and altered sensorium for 2 days. On admission child was irritable, febrile with GCS score of 11/15. On clinical examination he had dysmorphic facies, protuberant abdomen with umbilical hernia, short stature, grade II clubbing, joint contractures in both elbow joints. He had moderate hepatosplenomegaly with no signs of neck stiffness or meningeal irritation. Laboratory investigation revealed anemia, leucocytosis, thrombocytopenia with normal liver function test, renal function test and coagulation profile. Fever workup turned positive for leptospirosis. During the course of hospitalization, he had one episode of seizure in form of generalized tonic-clonic movements. In view of altered sensorium and seizures, magnetic resonance imaging of brain was done which showed large subdural hematoma with midline shift. Patient was treated with IV ceftriaxone, doxycycline and antiepileptics along with

parietal craniectomy and evacuation of subdural hematoma. His laboratory values are given in Table 1.

Table 1: Laboratory values.

Test	Patient value	Normal value
Haemoglobin (gm%)	9.1	11.5-15.5
Total count (cells/cu.mm)	6400	5000-15000
Differential count	55/38/7	P 38-68, L 25-54, E 2-8
PCV (%)	28	38-40
Platelet (cells/cu.mm)	25000	1.5-4 lakh
Total bilirubin (mg/dl)	0.4	0-1.2
Direct bilirubin (mg/dl)	0.1	0-0.3
Indirect bilirubin (mg/dl)	0.3	0-0.9
SGOT (U/l)	29	0-40
SGPT (U/l)	30	0-41
Alkaline phosphatase (U/l)	191	40-129
Total protein (g/dl)	7.1	6.6-8.7
Albumin (g/dl)	3.3	3.2-4.6
Globulin (g/dl)	3.8	2.5-3.5
Prothrombin time (s)	13.9	11.5-14.6
INR	1.1	0.9-1.1
Urea (mg/dl)	13	16-48
Creatinine (mg/dl)	0.6	0.7-1.2

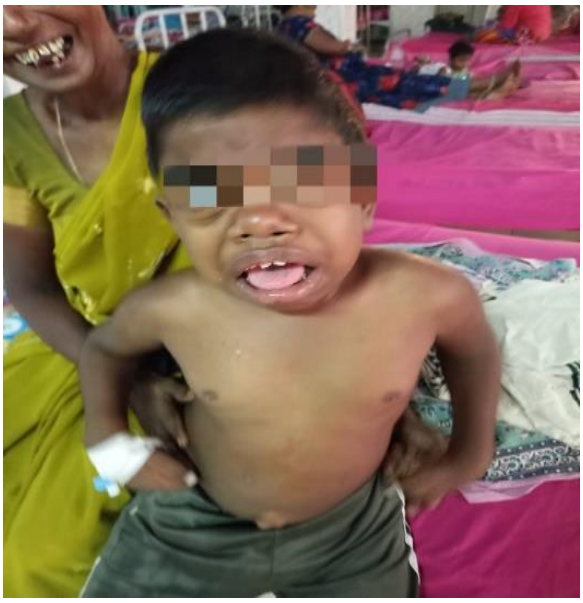

Figure 1: General appearance of the child at presentation.

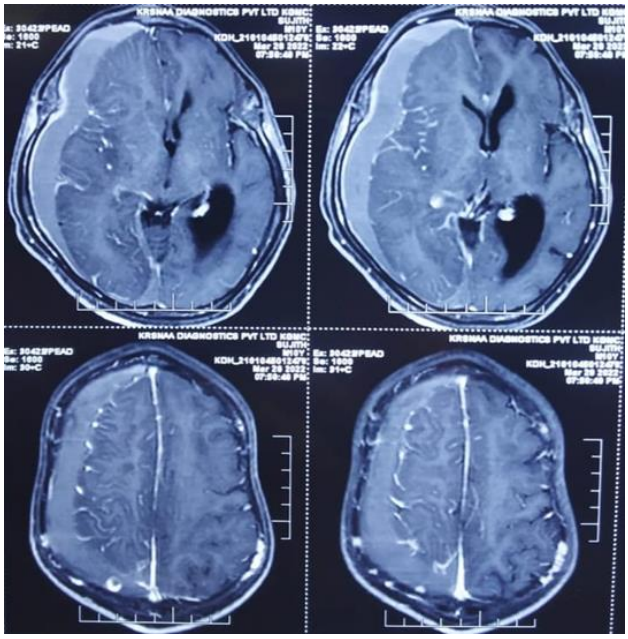

Figure 2: MRI brain showing large subdural hematoma along the cerebral convexity with mass effect, midline shift and herniation.

DISCUSSION

Hunter syndrome (mucopolysaccharidosis II) is an X-linked disorder caused by deficiency iduronate 2-sulfatase (IDS) which is mapped to chromosome Xq28. Point mutation, deletion or rearrangement of IDS have been detected in these patients.[3]

It manifests exclusively in male patients. Marked molecular heterogeneity explains wide clinical spectrum of hunter syndrome. Leptospirosis is a potentially fatal disease with bleeding and multiorgan failure as its clinical hallmark although majority of cases present with mild sudden onset of febrile illness. In this case report of Hunter syndrome after the fever workup, child had score of 26 according to Faine's criteria with definitive diagnosis of leptospirosis.[4] Unlike other infectious diseases, the bleeding tendency in leptospirosis is the result of imbalance in the haemostatic equilibrium. Leptospiral infection is self-limiting in 90% of cases but can cause potentially fatal illness like renal failure, liver failure, pneumonitis and haemorrhagic diathesis.[5] In this case report, we presented a large subdural haematoma which was rare complication due to thrombocytopenia and vasculitis occurring in leptospirosis even with a platelet count of 25,000 cells/mm^3. In rare case reports of otherwise normal healthy patients, it was reported that visceral haemorrhage occurred with a platelet count less than 20,000 cells/mm^3.[6] But in this case report, we found that visceral hemorrhage can occur even with platelet counts more than 25,000 cells/mm^3 when there was a underlying comorbid condition like mucopolysaccharidosis.

CONCLUSION

Through this case report, we present a case of leptospirosis in a known case of Hunters syndrome who presented with large intracranial hematoma which was actively intervened at the early stage of illness with parietal craniectomy and evacuation thereby reducing the mortality with good clinical outcomes.

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