

Priapism associated with Niemann–Pick disease in a 15-year-old boy

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ABSTRACT

A 15-year-old boy presented with priapism of 46 h duration. There was no known cause of priapism detected in him. During evaluation, biopsy of the lymph node and bone marrow aspiration detected patient to be having Niemann–Pick disease. He was managed with cavernous aspiration, saline irrigation, and intracavernosal phenylephrine injection. Although priapism is associated with many known diseases, about half of the cases are of idiopathic origin. We are reporting the first case of priapism associated with Niemann–Pick disease in the literature.

Key words: Niemann–Pick disease, priapism, sphingomyelin

INTRODUCTION

Priapism constitutes a urologic emergency in which persistent erection occurs that is not accompanied by sexual desire, usually lasting for more than 6 h and typically involving only the corpora cavernosa.^[1] Priapism has been associated with many known different disease states and some situations that constitute risk associations for the condition. It is estimated that idiopathic category accounts for as many as half of all documented cases. Niemann–Pick disease is a lysosomal storage disease caused by the deficiency of sphingomyelinase enzyme leading to the accumulation of sphingomyelin in the cells of the macrophage–monocyte system.^[2] To our knowledge this unique case of priapism associated with Niemann–Pick disease (sphingolipid accumulation) represents the first such case reported in the literature.

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CASE REPORT

A 15-year-old boy presented with persistent painful erection of 46 h duration. The patient had woken up in the early morning with pain. He had experienced a similar episode of painful erection 1 month back, which resolved spontaneously within 4 h. There was no history of perineal trauma or sickle cell anemia. He was not on any medications. There was a history of delayed developmental milestones.

Physical examination revealed a poorly nourished adolescent male with pallor, clubbing, and generalized lymphadenopathy (bilateral cervical, axillary, and inguinal). Liver was enlarged but spleen was not palpable. No xanthelesma observed. Genitourinary examination showed an erect viable penis, edema of proximal shaft of penis, and adjacent scrotum [Figure 1]. Blood sample aspirated from the penis was dark red and consistent with venous blood.

Laboratory evaluation revealed a white blood count of $13.9 \times 10^9/L$, hematocrit 43.6%, and platelet count $356 \times 10^9/L$, which were within normal limits. Sickle cell test was negative. Corporeal blood gas analysis showed a pH of 6.86, $paCO_2$ 88 mmHg, and paO_2 26 mmHg. Ultrasonography revealed mild hepatosplenomegaly. Biopsy of the lymph node from the cervical area showed infiltration of paracortical and cortical areas by foam cells, which are large cells with abundant pale pink vacuolated cytoplasm with small pyknotic nucleolus placed centrally as well as eccentrically [Figure 2]. Bone marrow aspiration showed large histiocytes with abundant vacuolated cytoplasm

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(sea-blue histiocytes) characteristic of Niemann–Pick cells suggestive of metabolic storage disorder [Figure 3]. These cells were PAS negative.

He was managed with cavernous aspiration and irrigation with saline followed by 100 µg phenylephrine injected into corpus cavernosum for a total of 4 doses, 10–15 min apart. One unit of blood transfusion was given with supplemental oxygen and sedation. Complete detumescence was achieved with this treatment. At 6 months follow-up, the patient denied spontaneous erection.

DISCUSSION

Various etiologies of priapism have been described, including sickle cell disease, hematologic malignancies, penile metastases, perineal trauma, total parenteral nutrition, intracavernous injection therapy for erectile dysfunction, and oral medications. Nevertheless, many cases of priapism are classified as idiopathic.^[3]

Niemann–Pick disease is an inherited lysosomal metabolic storage disease resulting from the deficient activity of the lysosomal hydrolase acid sphingomyelinase leading to lysosomal accumulation of sphingomyelin.^[2] Sphingomyelin is the major lipid that accumulates constituting about 70% of total phospholipid fraction (normal 5%–20%). The pathologic hallmark of Niemann–Pick disease is the characteristic lipid-laden foam cell often referred to as the Niemann–Pick cell in the bone marrow.^[2] Clinical presentation is with hepatosplenomegaly, moderate lymphadenopathy and psychomotor retardation. Enzymatic evaluation by measuring the acid sphingomyelinase activity level in peripheral leukocytes, cultured fibroblasts, and/or lymphoblasts will confirm the diagnosis by showing markedly decreased enzymatic activity.

There are few reports in the literature describing priapism associated with total parenteral nutrition. It is hypothesized that the lipid emulsion is the precipitating factor.^[3] Although the exact pathogenesis of the priapism associated with Niemann–Pick disease is not clear, possible mechanism could be accumulation of foam cells causing sluggish blood flow, hypercoagulability and venous outlet obstruction of corpora cavernosa.^[4] Since many cases of priapism have unidentifiable etiology, we consider that this could be one of the causes. This could be a coincidental and a random association. Occurrence of priapism led to the diagnosis of Niemann–Pick disease in this patient.

This boy had erection lasting 46 h long in duration, usually aspiration fails in such prolonged priapism and a shunt is required. Perhaps success of aspiration in this case could be attributed to the young age of the patient having protective action and hence irreversible damage did not occur despite long duration of priapism.



Figure 1: Erect viable penis with edema of proximal shaft and adjacent scrotum

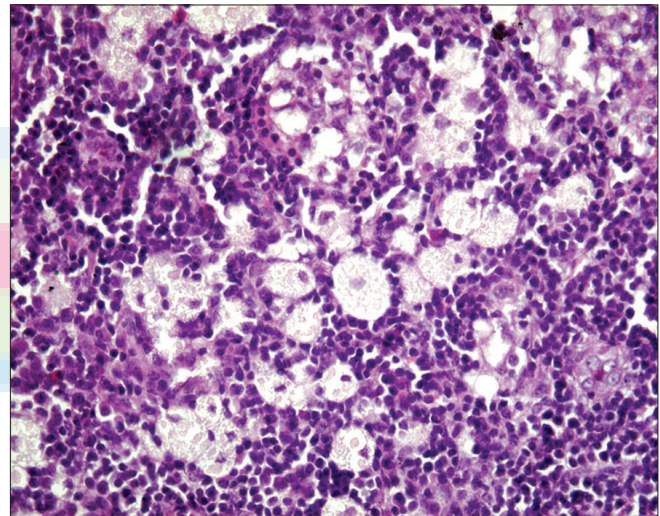


Figure 2: Lymph node biopsy showing infiltration of paracortical and cortical areas by foam cells (H and E, reduced from ×100)

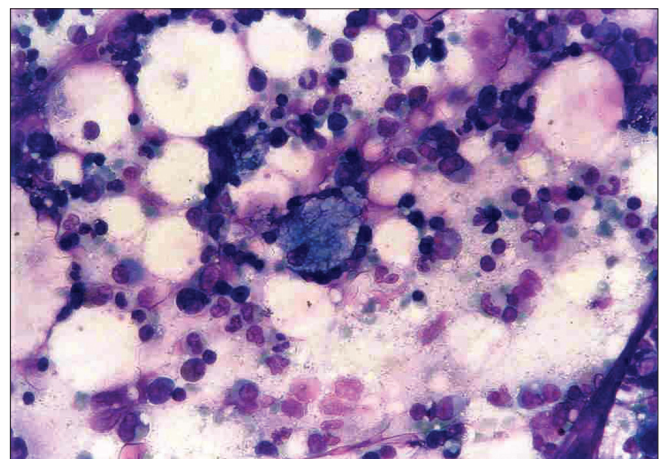


Figure 3: Bone marrow aspiration showing large histiocytes with abundant vacuolated cytoplasm (H and E, reduced from ×100)

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