Case Report

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LNS: Brick Red Sand Urine Disease

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Abstract

Lesch-Nyhan syndrome (LNS) is a rare, X-chromosome linked, genetically inherited disorder, present from the birth, but generally manifests itself in the first three to six months of life. The very first symptom, if observed carefully, is orange-coloured deposits of uric acid associated with blood in the diaper. Eight day old male child presented with respiratory distress and progressive loss of pain sensation. Radiological and hematological investigations were normal. Biochemically high urinary uric acid to creatinine ratio and uric acid crystals in urine helped diagnosis at an early day.

Keywords: LNS; Red Sand Urine; Uric Acid.

Introduction

Lesch-Nyhan syndrome (LNS) is a rare, X-chromosome linked, genetically inherited disorder. Deficiency of the enzyme Hypoxanthine Guanine Phosphoribosyl Transferase (HGPRTase) is responsible for the syndrome [1,2]. Gene mutation can either be a new or inherited from the mother, who is a carrier. Usually girls do not manifest this disease but remain silent carriers.

Though the disorder is present from the birth, generally it manifests itself in the first three to six months of life [1]. Moreover, less obvious features viz. developmental delay, hyperuricemia and hematuria, miss out the diagnosis at early days. Overproduction of uric acid at birth may not be recognized by clinical lab methods. Increased urine uric acid and uric acid stone may go undetected for months. Due to the deficiency or complete absence of enzyme HGPRTase uric acid builds up in the body fluid, excreting large amount of uric acid in urine[3,4].

The very first symptom of Lesch-Nyhan syndrome, if observed carefully, is orange-colored crystal-like deposits of uric acid in the diaper. Sometimes it is associated with blood due to Nephrolithiasis. Lack of muscle tone and writhing motions are some other symptoms [2] which need attention.

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Case Presentation

Apparently Normal, full term male baby of 3.25kg birth weight was delivered at SDM College of Medical Sciences and hospital at Dharwad (Karnataka). 1st minute APGAR score was 8/10 and after 5th minute 9/10.

On 2nd day baby developed respiratory distress, grunting and convulsions hence shifted to NICU. C/I reveal trouble feeding marked hypotonia. Routine Biochemical tests on serum and Complete Blood count reports were normal. Cerebro-spinal fluid (CSF) report indicated protein levels 1.7g/L with 11 lymphocytes/µl, gram stain and culture were negative. 2D echo and chest X-ray was normal. The USG cranium report indicated cerebral edema 2.2 & 3.2 cyst in the choroid plexus of the right lateral ventricle, NAC sepsis were suspected and treated.

On 6th day baby exhibited writhing, involuntary, repetitive limb movements. Baby had loose stool, and red colored urine. Reducing sugar in feces was absent. Urine routine examination revealed normal report except few calcium oxalate and uric acid crystals.

On 8th day the child was quiet and did not respond to any stimuli, no response to pinching pain, s/o spastic cerebral palsy and lack of muscle tone was noted. Suspecting encephalitis/meningitis CSF sample was sent for examinations which come out normal. Urine sample was sent to Biochemistry laboratory for screening and rule out any inborn error

of amino acid metabolism.

At the department of Biochemistry Screening tests for inborn error of amino acid and carbohydrates were performed by chemical methods followed by Thin layer chromatography [5]. All tests were negative. Looking at the colour of urinary sediments (Fig 1), blood was suspected. Benzidine test [6] for occult blood was performed which turned out mildly positive. When urine sediments were observed microscopically, large amount of uric acid crystals along with fewer red blood cells was observed under (40X) high power field (Fig 2 &3). With the suspect of LNS, urine uric acid [7] and urine creatinine[8] was estimated. Ratio of urine uric acid to urine creatinine was calculated which came out abnormally high [10.67 (mg / mg) [9]. Sample was repeated with the similar result.

Correlating symptoms, clinical examinations, Radiological and biochemical finding case were diagnosed as LNS.

Disscusion

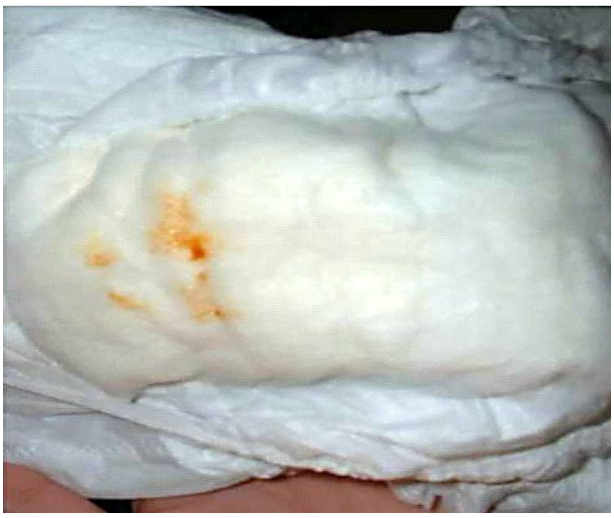

Fig. 1: Red colour granular sand seen on diaper of baby

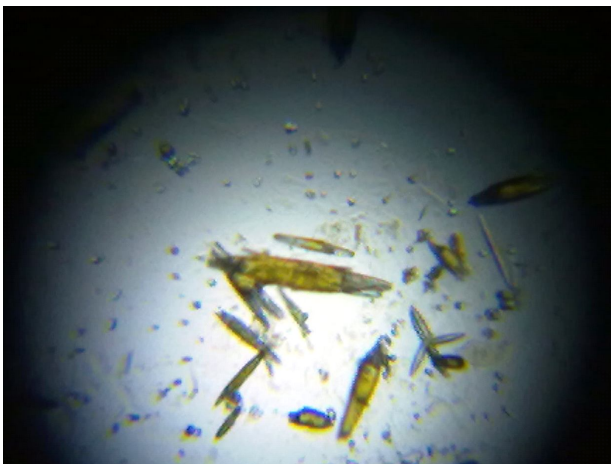

Fig. 2: Uric acid crystal under 10X

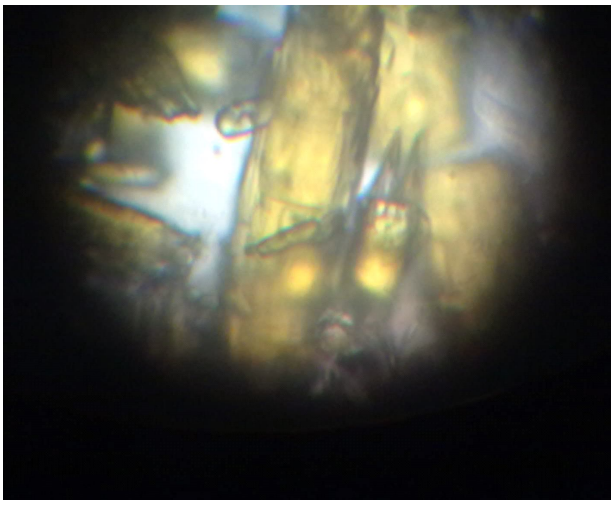

Fig. 3: Uric acid crystal under 40X

LNS is a genetic disease, common symptoms are irritability, lack of muscle control, cerebral palsy, type of anemia resulting from inability to use vitamin B_{12}. LNS diagnosis is based on characteristic triad (i) hyperuricemia, (ii) spectrum of neurological dysfunctions and (iii) cognitive and behavioral disturbances[1]. Though it takes time for symptoms to become obvious. One of the early clues of disease is the observation of orange crystals in the diapers of a newborn. HGPRT deficiency leads to hyperuricemia, precipitating crystals of uric acid in the renal system which may be passed in the urine [10]. Enzymatic determination of HGPRT activity in red blood cells or fibroblasts is confirmatory tests.

Life Expectancy

The prognosis of disease is poor. A significant proportion of patients die suddenly and unexpectedly from unknown causes. Death is usually due to nephrolithiasis or complications from hypotonia, respiratory abnormalities, aspiration or pneumonia.

Treatment

There are no guidelines to prevent but genetic counseling may help when deciding whether to have children.

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