DUCHENNE MUSCULAR DYSTROPHY WITH PUJ OBSTRUCTION: A CASE REPORT

Rupali P Shirke1, Shikha Handa1, Pramod Sharma1, Vipan Chander1
1Himalayan Institute of Medical Sciences, Swami Rama Himalayan University, Doiwala, Dehradun, Uttarakhand, India

Correspondence should be addressed to Rupali P Shirke

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ABSTRACT

Duchenne muscular dystrophy (DMD) is the most common hereditary condition with features of progressive weakness, intellectual impairment, pseudo-hypertrophy of calves with proliferation of connective tissue in muscle. Renal morbidities in the form of recurrent urinary tract infections are seen in these patients due to prolonged immobility however pelviureteral junction obstruction is very rarely seen.

KEY WORDS: Duchenne Muscular Dystrophy (DMD), PUJ Obstruction

INTRODUCTION

We present a case of 9 years old male, k/c/o of Duchenne muscular dystrophy, presenting to Department of Pediatrics, HIMS, Dehradun with foul smelling urine, burning micturition and itching and was subsequently diagnosed with PUJ Obstruction on ultrasound abdomen and was operated for the same. Patient’s recovery was uneventful.

CASE REPORT

A 9 years old male patient, known case of Duchenne muscular dystrophy, presented to pediatric opd with complaints of foul smelling urine, burning micturition and itching in genital region. On examination, gowers sign was positive with trendelenburg’s gait, distal muscle hypertrophy in bilateral lower limb with areflexia. Ultrasound abdomen was done which was s/o left sided PUJ obstruction. Pediatric surgery review was done and left sided pyeloplasty was done under general anesthesia. Patient recovered well with no post-operative complications.

DISCUSSION

Duchenne muscular dystrophy (DMD) is the hereditary condition with it’s a X-linked recessive trait, with abnormal gene at Xp21 locus. Incidence is 1 in 3600 live born infant boys. Poor head control in infancy may be the first sign of weakness. Common presentations seen are delayed walking, falling, toe walking, trouble running or walking upstairs developmental delay. Relentless progression of weakness continuing till 2nd decade, respiratory muscle involvement with frequent pulmonary infections and decreasing respiratory reserve, pharyngeal weakness leading to nasal regurgitation, nasal voice and aspiration, contractures of knees, hips and elbows are features associated with DMD. Pseudo-hypertrophy and wasting of thigh muscles are classical features. Cardiomyopathy with persistent tachycardia and myocardial failure is seen in 50-80% of patients. Smooth muscle dysfunction, particularly of gastrointestinal tract is a minor but often overlooked feature. Intellectual impairment occurs in all patients, but IQ <70 in 20-30% patients. There is no correlation of intellectual
impairment with severity of myopathy. Epilepsy is more common in pediatric age group. Death occurs about 18-20 years of age. Common cause being respiratory failure, intractable heart failure, pneumonia or occasionally airway obstruction or aspiration. Joyce Y Wu et al, reported a feature of autism in Duchene muscular dystrophy¹. Matsumura T et al, reported a case of Duchene muscular dystrophy with thrombotic thrombocytopenic purpura². Ghafoor T et al, reported a case of Duchene muscular dystrophy case with growth hormone deficiency [3]. Xiaozhou Du et al, reported an unusual case of left ventricular aneurysm in Duchene muscular dystrophy⁴. Byung Chun Chung et al, reported a case of acute gastroparesis with Duchene muscular dystrophy [5]. Velu Nair et al, reported a case of Diamond Blackfan anemia in a Duchene muscular dystrophy with successful bone marrow transplantation [6]. No case of duchenne muscular dystrophy with renal abnormalities have been reported till date. This is a very rare finding in a case of dmd which has not been reported so far.

REFERENCES