

## Clinical Image

# Dubowitz Syndrome

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Submitted: 21 April 2015

Accepted: 23 June 2015

Published: 25 June 2015

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## Keywords

- Syndrome
- Autosomal recessive Dismorphism
- Microcephaly

## Abstract

Dubowitz syndrome is an uncommon genetic disorder associated with typical facial dysmorphism and a variety of cutaneous, skeletal, systemic, immunological and hematological abnormalities. Thus far, only about 150 cases have been reported worldwide. We describe a nine year old boy with characteristic facies and a spectrum of other associated features which were consistent with the diagnosis of Dubowitz syndrome. Prompt diagnosis with regular follow-up and appropriate supportive care remains the key to the management of this disorder.

## INTRODUCTION

Dubowitz syndrome is a rare genetic disorder with peculiar facial features, that has been reported in the context of its unawareness among the clinicians, as most of the cases go under diagnosed and often missed. The pathogenesis of this syndrome is unknown and is presumed to represent the homozygous state of an autosomal recessive mutation [1]. However, Martinez et al have recently reported that mutations in NSUN2 may have a role in the pathogenesis of Dubowitz syndrome [2]. NSUN2 has been implicated in Myc-induced cell proliferation and mitotic spindle stability, which might help explain the varied clinical presentation in Dubowitz Syndrome [2]. Here, we report a case of a nine year old boy with facial dysmorphism and other associated features consistent with the diagnosis of Dubowitz syndrome.

## CASE

A nine year old male child born of a consanguineous marriage presented to the paediatric outpatient clinic with swelling of the left ileoinguinal region. A general physical examination revealed

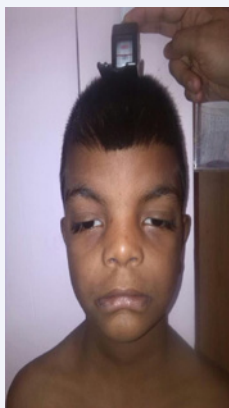


Figure 1 Distinctive facies.

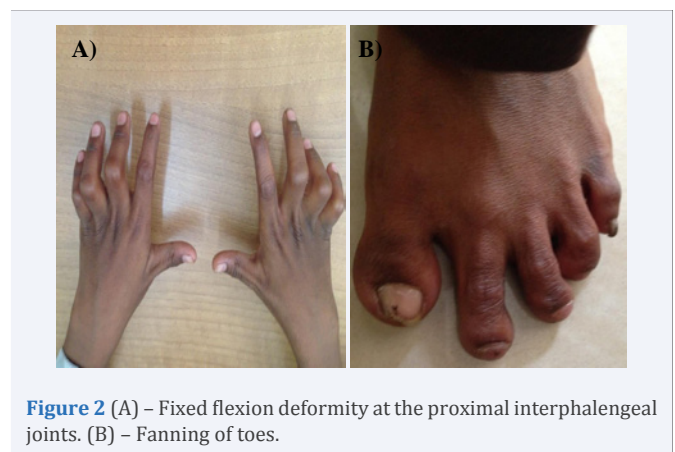


Figure 2 (A) – Fixed flexion deformity at the proximal interphalangeal joints. (B) – Fanning of toes.

that the child was short statured, had distinctive facial features with microcephaly, high arched and scanty lateral eyebrows, drooping eyelids, hypertelorism, low set ears, broad nasal bridge with rounded tip, high arched palate, broad mouth, long and smooth philtrum. Other features seen were fixed flexion deformity at the proximal interphalangeal joints of both the hands, cryptorchidism, left inguinal hernia and fanning of toes. Developmental assessment revealed a shy child with normal intelligence. Ophthalmological and Audiological assessment was normal. Systemic examination was unremarkable.

## DISCUSSION

The constellation of features in this child was consistent with the diagnosis of Dubowitz syndrome. This syndrome was first described by Victor Dubowitz in 1965 [3]. The phenotypic variability appears to be very broad suggesting actions of many modifying genetic and epigenetic factors [1,3]. Diagnosis of this rare syndrome is based primarily on the characteristic facial appearance, growth records and previous medical history [4]. There is no precise medical test that will conclusively consign the diagnosis of this syndrome since the genetic reason is unknown.

Till date, only 150 cases have been reported worldwide [5]. Apart from the characteristic facies, a variety of ocular, dental, cutaneous, skeletal, cardiovascular, gastrointestinal, neurological, immunological, and hematological medical difficulties have been described [5]. Further, patients with Dubowitz syndrome are at an increased risk of developing systemic malignancies, including Acute Lymphocytic leukemia as well as malignant lymphomas [6]. To the best of our knowledge, this is the first case to be reported from South India. Early identification with regular long term follow-up remains the cornerstone of management of such children.

## REFERENCES

1. Tsukahara M, Opitz JM. Dubowitz syndrome: review of 141 cases including 36 previously unreported patients. *Am J Med Genet.* 1996;63,1:277-289.
2. Martinez FJ, Lee JH, Lee JE, Blanco S et al. Whole exome sequencing identifies a splicing mutation in NSUN2 as a cause of a Dubowitz-like syndrome. *J Med Genet* 2012 Jun 49(6):380-5.
3. Dubowitz V. Familial Low Birthweight Dwarfism with an Unusual Facies and a Skin Eruption. *J Med Genet.*1965;42: 12-17.
4. Pascual JC, Betloch I, Banuls J, et al. What syndrome is this? Dubowitz syndrome. *Pediatr Dermatol.* 2005;22, 5:480-481.
5. Swartz KR, Resnick DK, Iskandar BJ, et al. Craniocervical anomalies in Dubowitz syndrome. Three cases and a literature review. *Pediatr Neurosurg.* 2003;38,5:238-243.
6. Kapoor S. Dubowitz syndrome and the increased risk of developing malignancies. *Pediatr Allergy Immunol.* 2015 May 22. doi: 10.1111/pai.12411.

### Cite this article

Nayantara Rao G, Mehta K, Sami MA, Rao NS (2015) Dubowitz Syndrome. *Ann Pediatr Child Health* 3(5): 1071.