Annals of Clinical Case Reports

Clinical Image
Published: 24 May, 2016

# Isolated Hemi-hyperplasia

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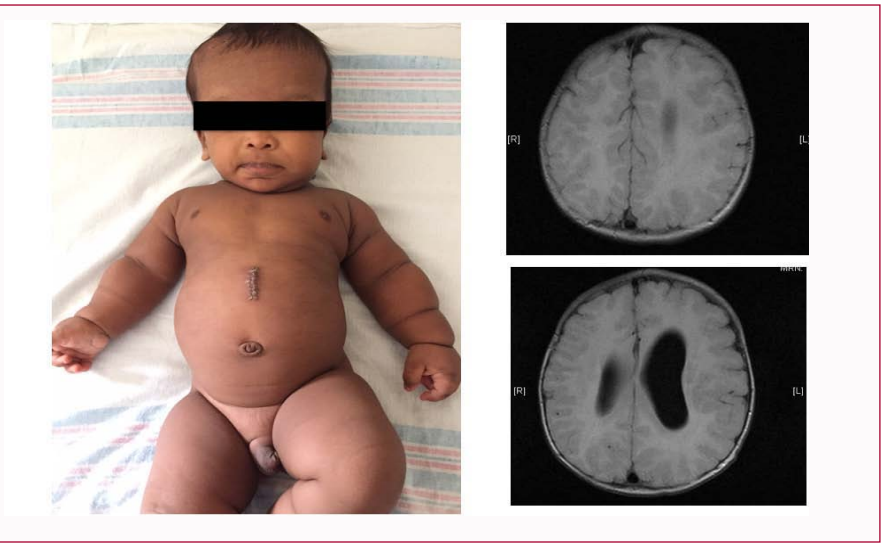
## Clinical Image

6-month old male presented with macrocephaly (progressive), macrosomia, macroglossia, splenomegaly, and left-sided hemi-hypertrophy. Magnetic Resonance Imaging showed large left cerebral hemisphere and hydrocephalus with normal fourth ventricle consistent with aqueductal stenosis. There was no evidence of any lipomas or hemangiomas. There was no family history of hemi-hypertrophy. Microarray based comparative genome hybridization and chromosome analyses were negative. Workup for Beckwith-Wiedemann syndrome (BWS), Neurofibromatosis, and PTEN hamartoma tumor syndromes (PHTS) was negative. A diagnosis of Isolated Hemi-hyperplasia was made which is characterized by asymmetric overgrowth of one or more regions of the body secondary to abnormal cell proliferation with an estimated incidence of 1 in 86,000. It is associated with seizures, undescended testes, inguinal hernia, communicating hydrocephalus, bicuspid aortic valve, cushing syndrome and increased risk of embryonal tumors particularly wilm's tumor. It is proposed to be part of the spectrum of phenotypes of BWS, which maps to 11p15.5.



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**Received Date**: *12 May 2016*
**Accepted Date:** *19 May 2016*
**Published Date:** *24 May 2016*

***Citation:***
*Singh D, Okeke O, Seeber LD, Akingbola O. Isolated Hemi-hyperplasia. Ann Clin Case Rep. 2016; 1: 1012.*