Case Report

Indian Journal of Maternal-Fetal and Neonatal Medicine
Volume 4 Number 1, January - June 2017
DOI: http://dx.doi.org/10.21088/ijmfnm.2347.999X.4117.16

Macrocephaly-Capillary Malformation: A Neonatal Case Report

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Received on 13 February 2017
Accepted on 22 February 2017

Abstract

Macrocephaly-capillary malformation (M-CM/ MCAP) was formerly known as Macrocephaly-cutis marmorata telangiectatica congenita. It is a multiple congenitial syndrome , which was first described in 1997 and it was renamed as " *Macrocephaly – capillary malformation"* in 2007. It is a multiple malformation syndrome which involves body and head overall abnormal growth, abnormalities of skin , vascular abnormalities and neurological problems (seizures, hypotonia), cortical brain malformations, most distinctively polymicrogyria. This disorder is attributed to PIK3CA gene mutation. Till now, There are only 130 reported cases of M-CM, there are likely many more affected individuals who have been misdiagnosed, are unrecognized or have not been published in the medical literature.

Keywords: Macrocephaly; Neonatal; Congenitial syndrome.

Case Report

A one and half month baby was admitted to our NICU with complaints of focal convulsions since 2 days and she had macrocephaly (42 cms), facial dysmorphism, several superficial vascular abnormalities on upper limb, axilla, chest and back, doughy skin, right hemi-hypertrophy of body. Ophthalmic findings were hypopigmented fundus, blunted foveal reflex s/o cones- rod dystrophy. Brain magentic resonance imaging s/o hemimegalencephaly with pachygyria (cortical malformation). Radiological examination of the skeleton showed asymmetry of the limbs. Lab findings were Hb: 6.7 % mg/dl, TLC: 3700 cumm, plt 2.11 lac/cumm, N37 L52 E1 M9 B1, Serum Bilirubin Total 1mg %, Direct: 0.2 mg%, Indirect 0.8 mg %, SGOT 180, SGPT 111, ALKPO4 217. CSF findings were colourless , clear in appearance, no blood, 32mg/ dl sugar and protein was 39 mg/ dl.Microscopy revealed no gram negative organism. CRP Was negative.

Fig. 1: Infant with facial dysmorphism and upper lip vascular anamoly

Fig. 2: Infant with severe capillary abnormalities & doughy skin

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Discussion

The infant reported herein presented macrocephaly-capillary malformation syndrome characterized by macrocephaly , superficial vascular anomaly, body asymmetry, doughy skin, hypotonia, focal seizures. Early diagnosis is helpful in prevention in future neurological, orthopedic, or oncologic complication. It is known as "Megalencephaly-capillary malformation-polymicrogyria syndrome (MCAP)"

Moore et al. (1997) [1] described 13 unrelated children with above abnormalities. Clayton-Smith et al. (1997) described 9 additional patients and recognized the macrocephaly-CMTC syndrome as a distinct entity [2]. Yano and Watanabe (2001) described 3 cases with features of macrocephaly-cutis marmorata telangiectatica congenita with poor clinical outcomes [3]. Gripp et al. (2009) noted that 2 of the patients had an initial diagnosis of megalencephaly, polymicrogyria-polydactyly hydrocephalus syndrome, a similar syndrome with overlapping features. Gripp et al. (2009) suggested that the 2 disorders may be related or on the same phenotypic spectrum; they proposed the term MPPH-CM to refer to this phenotypic spectrum [4]. Riviere et al. (2012) then sequenced the coding exons of PIK3CA in 29 individuals with megalencephaly with no mutations in the AKT3 or PIK3R2 genes and identified 14 additional PIK3CA mutations, with mutant allele frequencies ranging from 10 to 50%. Standard variant calling in exomes from 7 additional subjects with MCAP identified a mutation of the PIK3CA gene (C378Y) that was supported by 68 of 250 reads (27%) in another individual. Toriello and Mulliken (2007) suggested that the name MCMTC should be changed to MCM, for 'macrocephaly-capillary malformation.' The authors argued that the vascular lesions in this disorder are neither cutis marmorata nor cutis marmorata telangiectatica congenita, but are rather a type of capillary malformation in a patchy reticular pattern. Mirzaa et al. (2012) suggested use of the term MCAP rather than MCM to reflect the very large brain size, rather than simply large head size, that characterizes this syndrome, and the importance and high frequency of perisylvian polymicrogyria [5].

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