

Case Report

Dandy-Walker malformation and neurocutaneous melanosis in a three-month-old infant

Mohamed Osman Swar (1), Shaza Mohamed Mahgoub (2), Rehab Omer Yassin (1), Atika Mohamed Osman (1)

(1) Ahfad University for Women, Sudan

(2) Mohamed Alamin Hamid Hospital for Children, Omdurman, Sudan

ABSTRACT

Dandy-Walker Malformation (DWM) is a rare congenital malformation of the brain. It is characterized by cystic enlargement of the fourth ventricle which is communicating with an enlarged posterior fossa, cerebellar dysgenesis, high tentorial insertion and hydrocephalus. Neurocutaneous Melanosis (NCM) is a congenital neurocutaneous syndrome characterized by large or multiple melanocytic nevi and benign or malignant melanocytic tumors of the leptomeninges. We report three months old boy who presented with projectile vomiting associated with a noticeable increase in head size. Several congenital nevi were seen all over his body with evident signs of hydrocephalus. The association of DWM and NCM is a rare complex, and to our knowledge, this is the eleventh case to be reported in the literature. In this article, we discuss the proposed pathogenesis, classification and management of the condition.

Key words:

Dandy-Walker malformation, Neurocutaneous melanosis

INTRODUCTION

Neurocutaneous Melanosis (NCM) is a non-hereditary congenital neurocutaneous syndrome characterized by large or multiple melanocytic nevi and benign or malignant melanocytic tumors of the leptomeninges. It is believed that abnormal neural crest cell differentiation and migration, which is usually governed by specific genes (PAX3, ZIC2, WNT, SLUG, SOX10), lead to NCM [1]. Meningeal cells are known to have a strong role in cerebellar development, thus early meningeal melanosis may disrupt the normal development of the cerebellum and the fourth ventricle leading to DWM [2]. Cutaneous lesions are usually recognized at birth – though more may appear later- but neurological manifestations appear later as the pressure gradient between the cerebrospinal fluid and venous pressure decreases [3]. Malignant transformation of leptomeningeal tumors occur in 40 - 60% of patients [4]. Dandy-Walker syndrome affects 1 in 25000 to 30000 newborns; more commonly seen among patients with Trisomy 9, 13, 18 and 21 but multi factorial pathogenesis is suggested in sporadic cases. The condition presents

Correspondence to:

Swar M O,

Head Department of Pediatrics and Child Health,
Ahfad University for Women, Omdurman, Sudan

E-mail: moswar2@hotmail.com

How to cite this article:

Swar MO, Mahgoub SM, Yassin RO, Osman AM. Dandy-Walker malformation and neurocutaneous melanosis in a three-month-old infant. Sudan J Paediatr 2013; 13(2):61-65.

in the form of Dandy-Walker Malformation that includes enlargement of the posterior fossa, high-set tentorium, agenesis of the cerebellar vermis, cystic dilation of the fourth ventricle that is communicating with the posterior fossa and hydrocephalus. In Dandy-Walker complex, the condition is associated with other congenital malformations e.g. neurocutaneous melanosis, holoprosencephaly, agenesis of corpus callosum, encephalocele, spina bifida, congenital heart or kidney disease and polydactyly or syndactyly. Dandy-Walker Variant is a mild form of malformation with normal lateral and third ventricles, normal brain stem and there is no torcular-lambdoid inversion [5]. Though shunt placement, corticotherapy and chemotherapy may lead to transient relief, the prognosis of the association of NCM and DWM is still very poor and affected children usually die before age 4 years [6].

CASE REPORT

Three months old boy presented to our hospital with persistent projectile vomiting for 35 days prior to admission. His mother noted that there was a noticeable increase in his head size in addition to multiple pigmented skin patches that were seen since birth. He was a product of a non-consanguineous marriage and pregnancy and delivery were uneventful. There was no family history of a similar condition. On examination, the child was fully conscious and had no dysmorphic features. There were eleven well defined non-hairy, hyper pigmented nevi with regular borders seen all over his body. Three giant nevi were seen at the lower back and a fourth one showed elevation and plaque formation at the dorsum of the left foot and extending along the toes to the sole (Fig1,a,b,c).



Figure 1A - Giant nevi at Lumbosacral area



Figure 1B - Giant nevus at dorsum of left foot



Figure 1C - Giant nevus extending to the sole of left foot

Head circumference was 47cm (more than the 97th centile for his age) with widely open anterior and posterior fontanels. There were dilated veins on the temporal area and a divergent squint of the left eye (Fig 2).



Figure 2 - Prominent forehead, nevi and divergent squint

His weight, height, spine and other systems were normal. Laboratory investigations done showed normal values for his age. Brain CT Scan showed hypo plastic cerebellar hemispheres and vermis, mega cistern magna, grossly dilated ventricles with signs of tension hydrocephalus. There was no evidence of space occupying lesion and no calcifications or hemorrhages seen (Fig 3 a, b, c). The patient received supportive treatment and shunt placement after which the head circumference decreased slightly and no new neurological deficits seen.

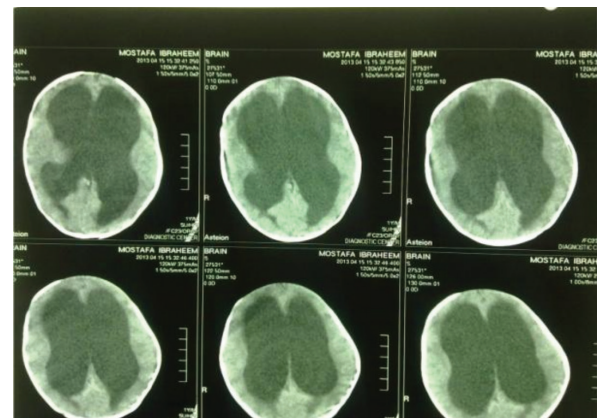
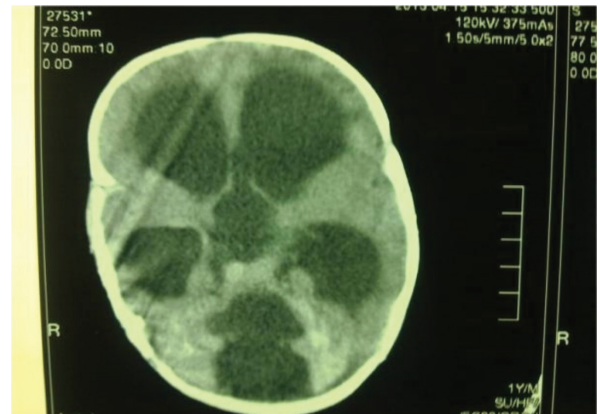
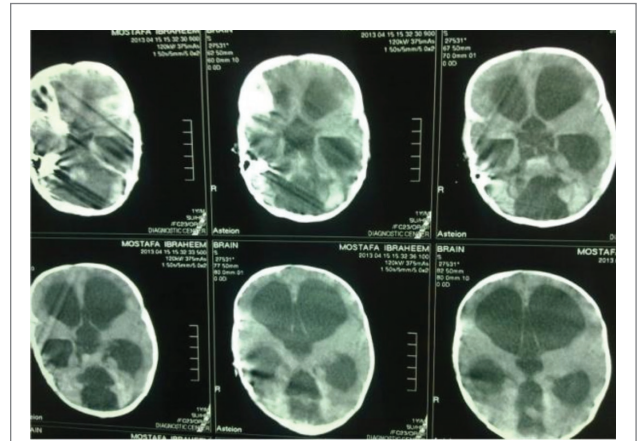


Figure 3 - Serial CT scans showing hypo plastic cerebellar hemispheres and vermis, mega cistern magna and grossly dilated ventricles

DISCUSSION

Neurocutaneous melanosis is a rare congenital disorder characterized by excessive proliferation of melanin-producing cells in both the skin and leptomeninges. It is non-heritable and to our knowledge, only 100 cases are reported in the literature and 10% of them were associated with Dandy -Walker Malformation (DWM) [7]. Although the exact pathogenesis is not fully understood, the condition is thought to result from an abnormality in the development of neural crest –derived melanocyte precursors, or melanoblasts, of the skin and pia mater [8]. The criteria set for diagnosis of NCM includes a) large or multiple congenital melanocytic nevi in association with meningeal melanosis or melanoma, b) no evidence of cutaneous melanoma, except in patients in whom the examined portions of the meningeal lesions are histologically benign, c) no evidence of meningeal melanoma, except in patients in whom the examined areas of the skin are histologically benign. Confirmation of the diagnosis of NCM is made by histological findings whether by endoscopy or at autopsy, however, the paramagnetic properties of melanin and the sensitivity of MRI allows for diagnosis of NCM as well as DWM. Giant congenital melanocytic nevus, usually at the Lumbosacral region, is present in two thirds of patients with NCM. In our patient, three giant nevi were seen in the lumbosacral region and a fourth one on the left foot. The risk of development of symptomatic NCM in patients with giant congenital nevus ranges from [2.59 - 11%]. Half of patients with symptomatic NCM manifest neurological symptoms

and signs related to hydrocephalus during the first year of life [10]. Hydrocephalus also results from the associated DWM probably secondary to induction failure of the opposing cerebral plates at or before the seventh week of gestation. Where both pathologies of NCM and DWM are seen as in our patient, signs and symptoms of hydrocephalus appear early. NCM may also be associated with Neurofibromatosis type 1 and Sturge Weber Syndrome. Posterior fossa malformation is sometimes associated with facial hemangioma, arterial anomalies, cardiac defects and eye abnormalities, collectively called PHACE Syndrome [11, 12, 13]. The association of NCM and DWM has poor prognosis and patients die before the age of 4 years with the neurological deterioration and malignancy transformation which is reported to occur in 515%- of these patients [14]. Because of this risk, some dermatologists advocate prophylactic resection of these nevi. Even after surgical treatment, corticotherapy and chemotherapy, more than half of all patients die within 3 years after presentation with neurological symptoms [15,16].

CONCLUSION

The association of NCM with DWM is increasingly recognized and it carries a very poor prognosis. Early shunt placement, surgical resection of nevi, chemotherapy and corticotherapy are helpful but not curative. More research on the pathogenesis during embryonic life is needed in order to advocate the possible methods of management.

REFERENCES

1. Samat HB, Flores- Samat L. Embryology of the neural crest: its inductive role in the neurocutaneous syndrome. *J Child Neurol* 2005 Aug; 20(8):637-43
2. Kadonaga JN, Barkovich AJ, Edwards MS, Frieden IJ. Neurocutaneous melanosis in association with the Dandy-Walker complex. *Pediatr Dermatol* 1992 Mar; 9(1):37-43
3. In Yong Cho, Sung-Kyun Hwang, Sung-Hak Kim. Dandy Walker Malformation Associated with

- Neurocutaneous Melanosis. *J Korean Neurosurg Soc* 2011 Nov;50(5):475-477
4. Kadonaga NJ, Frieden IJ. Neurocutaneous melanosis: definition and review of the literature. *J Am Acad Dermatol* 1991; 24: 747-755
 5. Osebach RK, Menezes AH. Diagnosis and management of the Dandy-Walker Malformation: 30 years experience. *Pediatr Neurosurg* 1992; 18(4):179-89
 6. Rhodes RE, Friedman HS, Hatten HP Jr, Hockenberger B, Oakes WJ, Tomita T. Contrast-enhanced MRI imaging of neurocutaneous melanosis. *AJNR Am J Neuroradiol* 1991; 12:380-382
 7. Marnet D, Vinchon M, Mostofi K, Catteau B, Kerdroan O, Dellemmes P. Neurocutaneous melanosis and the Dandy-Walker complex: an uncommon but not so insignificant association. *Child Nerv Syst* 2009 Dec; 25(12):1533-9
 8. Demirci A, Kawamura Y, Sze G, Duncan C. MR of parenchymal neurocutaneous melanosis. *AJNR Am J Neuroradiol* 1995; 16:603-606
 9. Rufo Campos M, Castro Mendez E, Ruiz del Portal L. Neurocutaneous melanosis and Dandy-Walker syndrome. *An Esp Pediatr* 1998; 48:318-20
 10. Bittencourt FV, Marghoob AA, Kopf AW, Koenig KL, Bart RS. Large congenital melanocytic nevi and the risk for development of malignant melanoma and neurocutaneous melanocytosis. *Pediatr* 2000; 106:736-741
 11. DeDavid M, Orlow SJ, Provost N, Margoob AA, Rao BK, Huang CL, et al. Study of large melanocytic nevi and associated malignant melanomas: review of cases in the New York University Registry and the world literature. *J Am Acad Dermatol* 1997; 36:409-416
 12. Purkait R, Samanta T, Thakur S, Dhar S. Neurocutaneous syndrome: a prospective study. *Indian J Dermatol* 2011 Jul; 56(4):375-9
 13. Coats DK, Paysse EA, Levy ML. PHACE: a neurocutaneous syndrome with important ophthalmologic implications: case report and literature review. *Ophthalmology* Sep 1999; 106(9): 1739-41
 14. Frieden IJ, Reese V, Cohen D. PHACE Syndrome. The association of posterior fossa brain malformations, hemangiomas, arterial anomalies, coarctation of the aorta and cardiac defects, and eye abnormalities. *Arch Dermatol* Mar 1996; 132(3):307-11
 15. Mena-Cedillos CA, Valencia-Herrera AM, Arroyo-Pineda AI, et al. Neurocutaneous melanosis in association with the Dandy-Walker complex, complicated by melanoma: report of a case and literature review. *Pediatr Dermatol* 2002; 19:237-242
 16. Kadonaga NJ, Barkovich AJ, Edwards MS, Frieden IJ. Neurocutaneous melanosis in association with Dandy-Walker complex. *Pediatr Dermatol* 1992; 9: 37-43