

Tecto-Cerebellar Dysraphia Manifesting as Occipital Meningocele Associated with Congenital Melanocytic Nevi and Pectus Excavatum

Amit Agrawal¹, MCh; Sudhakar Ratanlal Joharapurkar, MS² and Ata-Ullah Khan³, MD

1. Department of Surgery, Datta Meghe Institute of Medical Sciences, Sawangi (Meghe), Wardha, India
2. DMDPGME & R, Datta Meghe Institute of Medical Sciences, Sawangi (Meghe), Wardha, India
3. Department of Anesthesiology, Datta Meghe Institute of Medical Sciences, Sawangi (Meghe), Wardha, India

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Abstract

Background: Only few reported cases of tectocerebellar dysraphia with occipital encephalocele have been reported in the literature.

Case Presentation: Three month baby boy, the first child of healthy, consanguineous parents presented with a small swelling over the occipital region since birth. The child also used to have apneic spells without cyanosis and spontaneous recovery. CT scan showed absence of the cerebellar vermis, absence of tectum and the 4th ventricle communicating with the occipital meningocele sac and an occipital bone defect. The excision of the encephalocele sac was performed, however the child continued to have apneic spells and did not do well.

Conclusion: In our child irregular respiration probably was the manifestation of the tecto-cerebellar dysraphia syndrome complex and associated shunt malfunction followed by seizures decompensated the physiology of the child leading to fatal outcome.

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Key Words: Dandy-Walker Syndrome; Meningocele; Nevus, Melanocytic; Pectus Excavatum

Introduction

Dandy-Walker Syndrome (recently known as Dandy-Walker variant) is defined as a congenital malformation of structures in the posterior fossa and is characterized by complete or partial absence of cerebellar vermis, cystic dilatation of fourth ventricle and frequently hydrocephalus^[1].

Friede proposed a new clinico-pathological entity distinguished from Dandy Walker syndrome in the classification of aplasias of the cerebellar vermis that is known as "tectocerebellar dysraphia with occipital encephalocele"^[2]. Only few reported cases of this entity are there in literature^[2-5]. We report a rare case of tecto-cerebellar dysraphia manifesting as

* Corresponding Author;

Address: Division of Neurosurgery, Datta Meghe Institute of Medical Sciences, Sawangi, Wardha- 442005, Maharashtra, India

E-mail: dramitagrawal@gmail.com

occipital meningocele associated with congenital melanocytic nevi and pectus excavatum and also discuss the relevance of such findings.

Case Presentation

Three month baby boy, the first child of healthy, consanguineous parents (the parents are double third cousins) presented with a small swelling over the occipital region since birth. The child used to have apneic spells without cyanosis and spontaneous recovery.

There was no history of maternal exposure to environmental hazards during pregnancy. The family history was otherwise unremarkable. The pregnancy was uneventful with full-term normal delivery. Child was delivered at home and cried immediately after birth. Local examination revealed a small (1.5 cm x 1.5 cm), broad based, non-transilluminant, nontender, fluctuant and non-pulsatile swelling in the occipital region (Fig. 1). There was no bruit. In addition child had large hyperpigmented patch (?Haemangioma, ?congenital melanotic nevus) at the nape of the neck (Fig. 1) and pectus excavatum. Neurological



Fig. 1: Clinical photograph showing occipital swelling and large hyper-pigmented patch (?Hemangioma ?Congenital naevi) at the nape of the neck

developmental milestones were normal according to age. Neurological examination showed no gross motor or sensory deficit. There were no features of facial dysmorphism. The anterior fontanelle was lax. CT scan head showed absence of the cerebellar vermis, absence of tectum and the 4th ventricle communicating with the occipital meningocele

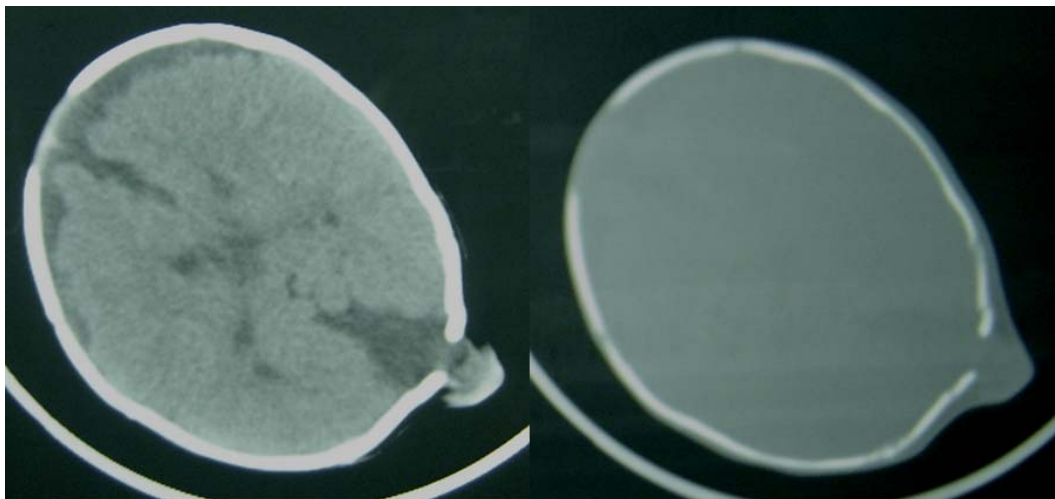


Fig. 2: CT scan with bone window showing absence of cerebellar vermis, distorted tectum and cystic lesion in the posterior fossa that was continuous with 4th ventricle and occipital lesion

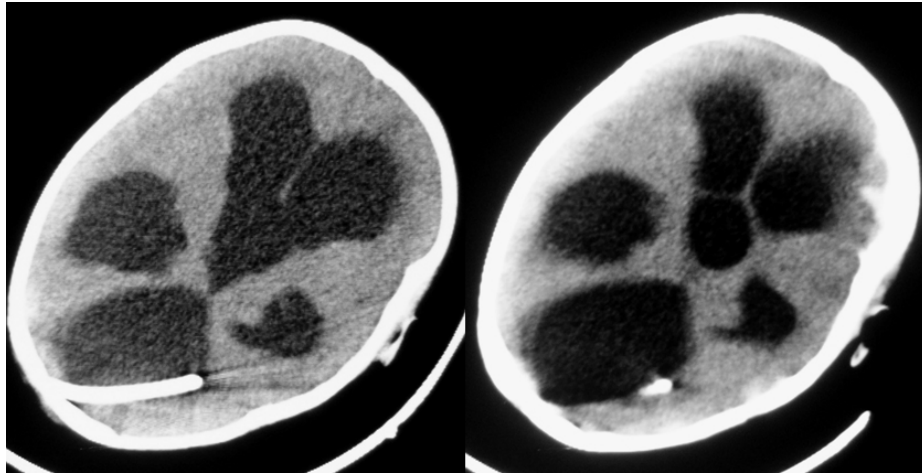


Fig. 3: The shunt tip probably embedded in cerebellar parenchyma leading to the shunt malfunction

sac and an occipital bone defect (Fig. 2). These features led to a diagnosis of Dandy-Walker variant in association with occipital meningocele. An elliptical skin incision was made on both sides encircling the whole circumference of meningocele. Scalp flap was separated all around the meningocele sac and redundant fat tissue was removed to reduce the size of swelling. After this a small sac could be defined, and the dura mater was thinned with one venous channel that could be seen at the lower end of the sac. The sac was not opened as it could be reduced and replaced through the defect. Pedicled pericranial graft was harvested and it was closed over the defect to keep the sac inside the cranium. The skin was closed in layers. Child was doing well at follow up till 10 months. However respiratory difficulty was persisting. Recently child presented with the history of altered sensorium and inability to accept feed and poor cry. On examination the child was dull, anterior fontanel was full. He was moving all four limbs. A repeat CT scan suggested shunt malfunction (Fig 3). The extensive dilatation of the posterior fossa cyst could be due to the reposition of the occipital sac contents into the cranial cavity. The shunt revision was performed and child showed improvement in his neurological status and started accepting feeds. After sixth day of shunt revision he had one episode of generalized tonic clonic seizures followed by an episode of apnea

and cardiac arrest. Although he could be revived from cardiac arrest but continued to deteriorate and expired.

Discussion

Tecto-cerebellar dysraphia with occipital encephalocele is thought to be a tandem malformation in which either an occipital dysraphia or an encephalocele induces aplasia of the vermis. The common findings in patients with tecto-cerebellar dysraphia with occipital encephalocele include occipital encephalocele, aplasia of the vermis and deformity of the tectum^[2]. The clinical features seen in tecto-cerebellar dysraphia with occipital encephalocele include episodic tachypnea and irregular breathing, opsoclonus, ataxia, marked hypotonia of the limbs, coloboma, and polydactyly^[3]. No special method has been proposed for treating occipital meningocele associated with Dandy-Walker syndrome^[5]. When cranium bifidum is present, the hydrocephalus may be concealed by the pressure buffering effect of the encephalocele^[6].

It has been reported that when it is associated with large cyst, the occipital meningocele will disappear after shunting even without surgical

repair^[7]. The occipital meningocele may be a part of huge Dandy-Walker cyst and need surgical repair^[5]. The neurological outcome in children with occipital meningocele is largely determined by the amount of neural tissue contained in the sac, which can vary in size from small insignificant masses with mature skin cover (as in present case) to massive masses with almost a complete prolapse of the brain and associated microcephaly^[5,7]. Congenital melanocytic nevi (CMN) are benign, neuroectodermal malformations composed of melanocytes and occasional neural elements that colonize the epidermis. These lesions may represent an anomaly in embryogenesis, and thus may be considered hamartomas^[8-10]. It is estimated that between 1% and 6% of the population are born with at least one CMN or congenital-nevus-like melanocytic nevi^[11]. Neurocutaneous melanosis and Dandy-Walker malformation are both forms of rare congenital neurodysplasia. Interestingly, 8 to 10% of patients with neurocutaneous melanosis also harbor an associated Dandy-Walker malformation, indicating that these developmental abnormalities share a common origin^[12]. There is possibility that excessive melanocytes hinder normal mesenchymal development^[12]. Since meningeal cells have been shown experimentally to play a critical role in cerebellar development, it has been hypothesized that the association of NCM with a Dandy-Walker malformation may be due to meningeal melanosis disrupting the normal development of the cerebellum and fourth ventricle^[13]. Since CMN have been shown to have potential for malignant transformation, there are guidelines to evaluate these lesions, assess their risk for transformation, and develop a management plan. Under the most widely used size classification for CMN, lesions are divided into small (<1.5 cm), medium (1.5–19.9 cm), and large (>19.9 cm) diameter CMN^[14]. The other risk factors include nevi located on the head, posterior neck, and/or paravertebral area seem to have the highest risk^[15]. In our patient the lesion was of medium size. Though, the risk of developing melanoma in small and medium CMN is very small but not zero^[16,17,18]. Appropriate investigations include a screening magnetic resonance imaging scan, neurological evaluation,

and serial clinical observations for the development of cutaneous melanoma^[19]. The treating surgeon must have a solid working knowledge of these challenging lesions, the disease's histology, its natural history and complications, and the options for treatment^[19].

Chest wall deformities is not an uncommon disease in infants and children, and pectus excavatum, or funnel chest, is the most common congenital chest wall deformity, accounting for more than 90% of such defects^[20,21]. The importance of this defect lies in the fact that it can be combined with organic problems as well as cosmetic problems^[21]. Diagnostic techniques for pectus excavatum include chest CT, cardiologic evaluation, and pulmonary evaluation as well as history taking and physical examination. Pulmonary evaluation is, however, limited to teenagers and adults who are able to cooperate^[22].

Conclusion

In our child irregular respiration probably was the manifestation of the tecto-cerebellar dysraphia syndrome complex and associated shunt malfunction followed by seizures decompensate the physiology of the child leading to fatal outcome.

References

1. Dandy WE, Blackfan KD. Internal hydrocephalus due to occlusions of the foramina of Magendie and Luschka. *Surg Gynecol Obstet*. 1921; 32: 113-24.
2. Friede RL. Uncommon syndromes of cerebellar vermis aplasia. II: Tecto-cerebellar dysraphia with occipital encephalocele. *Dev Med Child Neurol*. 1978;20(6):764-72.
3. Chowdhary UM, Ibrahim AW, Ammar AS, Dawodu AH. Tecto-cerebellar dysraphia with occipital encephalocele. *Surg Neurol*. 1989; 31(4):310-4.
4. Krishnamurthy S, Kapoor S, Sharma V, et al. Tectocerebellar dysraphia and occipital encephalocele: an unusual association with

- abdominal situs inversus and congenital heart disease. *Indian J Pediatr.* 2008;75(11):1178-80.
5. Cakmak A, Zeyrek D, Cekin A, Karazeybek H. Dandy-Walker syndrome together with occipital encephalocele. *Minerva Pediatr.* 2008;60(4): 465-8.
 6. Oi S. Radical reconstructive surgery for apert dysraphic state preserving neuro-function and structure: Operative technique for cranium bifidum apertum. (in Japanese) *No Shinkei Geka.* 1992;20(12):1232-40.
 7. Bindal AK, Storrs BB, McLone DG. Occipital meningoceles in patients with the Dandy-Walker syndrome. *Neurosurgery.* 1991;28(6): 844-7.
 8. Tromberg J, Bauer B, Benvenuto-Andrade C, et al. Congenital melanocytic nevi needing treatment. *Dermatol Ther.* 2005;18(2): 136-50.
 9. Zaal LH, Mooi WJ, Sillevs Smitt JH, et al. Classification of congenital melanocytic naevi and malignant transformation: a review of the literature. *Br J Plast Surg.* 2004;57(8):707-19.
 10. Chung C, Forte AJ, Narayan D, et al. Giant nevi: a review. *J Craniofac Surg.* 2006;17(6):1210-5.
 11. Marghoob AA, Borego JP, Halpern AC. Congenital melanocytic nevi: treatment modalities and management options. *Semin Cutan Med Surg.* 2003;22(12):21-32.
 12. Arai M, Nosaka K, Kashihara K, et al. Neurocutaneous melanosis associated with Dandy-Walker malformation and a meningoencephalocele. Case report. *J Neurosurg.* 2004;100(5 Suppl Pediatrics):501-5.
 13. Kadonaga JN, Barkovich AJ, Edwards MS, et al. Neurocutaneous melanosis in association with the Dandy-Walker complex. *Pediatr Dermatol.* 1992;9(1):37-43.
 14. Kopf AW, Bart RS, Hennessey P. Congenital nevocytic nevi and malignant melanomas. *J Am Acad Dermatol.* 1979;1(2):123-30.
 15. Bittencourt FV, Marghoob AA, Kopf AW, et al. Large congenital melanocytic nevi and the risk for development of malignant melanoma and neurocutaneous melanocytosis. *Pediatrics.* 2000;106(4):736-41.
 16. Illig L, Weidner F, Hundeiker M. Congenital nevi less than 10 cm as precursors to melanoma. *Arch Dermatol.* 1985;121(10):1274-81.
 17. Sahin S, Levin L, Kopf AW, et al. Risk of melanoma in medium-sized congenital melanocytic nevi: a follow-up study. *J Am Acad Dermatol.* 1998;39(3):428-33.
 18. Foster RD, Williams ML, Barkovich AJ, et al. Giant congenital melanocytic nevi: the significance of neurocutaneous melanosis in neurologically asymptomatic children. *Plast Reconstr Surg.* 2001;107(4):933-41.
 19. Arneja JS, Gosain AK. Giant congenital melanocytic nevi. *Plast Reconstr Surg.* 2007; 120(2):26e-40e.
 20. Sadler TW. Embryology of the sternum. *Chest Surg Clin N Am.* 2000;10(2):237-44.
 21. Shamberger RC. Current problems in surgery. 1996;6(XXXIII):471-542.
 22. Jo WM, Choi YH, Sohn YS, et al. Surgical treatment for pectus excavatum. *J Korean Med Sci.* 2003;18(3):360-4.