Rare disease

Hemihydranencephaly: rare disease with key to secrets of the brain

Lorraine S Dias,¹ K N Shivashankara,¹ G Vivek²

¹Department of Medicine, KMC, Manipal, Karnataka, India;

²Department of Cardiology, KMC, Manipal, Karnataka, India

Correspondence to G Vivek, vivekgraman@gmail.com

Summary

A 21-year-old male presented to us in status epilepticus. On investigation, he was found to have an extremely rare disorder called 'hemihydranencephaly'. Review of literature revealed that hemihydranencephaly was not always incompatible with life and that research into this disorder would help in understanding the development, functioning, untapped potential and reorganisation capacity of the brain.

BACKGROUND

Hemihydranencephaly is theorised to be caused by a rare vascular anomaly wherein there is occlusion of a unilateral carotid artery, prior to the third trimester of gestation.¹ The condition manifests with contralateral hemiparesis, seizures and various degrees of mental retardation.² We report on a 21-year-old male who presented with status epilepticus and was diagnosed with this disorder. We also review available literature.

CASE PRESENTATION

The 21-year-old male was born at full term of non-consanguineous healthy parents. The prenatal and perinatal periods

had been uneventful. His mother had noticed delayed developmental milestones and left hemiparesis at 3 months. Although he was evaluated at another health facility, no records or brain imaging studies were available. He had two episodes of generalised tonic-clonic seizures in the year preceding admission at our hospital. He had not been on any medication. At presentation, he was in status epilepticus. Left sided hemiparesis with disuse atrophy and left asymmetrical pupil were noted. He was dilantinised and seizures were controlled. He had severe mental retardation. Motor skills were comparable to that of an 8 month old and language skill of a 1.5-year-old child. Routine laboratory investigations were normal. MRI brain

Figure 1 T1W MRI of brain, coronal view showing an absent right hemisphere replaced by cerebrospinal fluid and an occluded right internal carotid artery.

Table 1 Features of cases reported with hemihydranencephaly

Author	Observation	Neuroradiological findings	Neurologic signs
Muir ¹¹	4 years	Left hemihydranencephaly	—
Warkany ⁹	3.5 years	Left hemihydranencephaly	—
Moser and Seljeskog ⁷	At birth	Left hemihydranencephaly with mass effect on left	Hydrocephalus, left hemiparesis, mild mental retardation
Suzuki <i>et al</i> ⁶	20 months	Left hemihydranencephaly	Right hemiparesis, left optic atrophy, mild mental retardation
Ohtsuka ¹⁰	11 years	Left hemihydranencephaly with underdeveloped brainstem	Right hemiparesis, anophthalmia, seizures, severe mental retardation
Van Doornik <i>et al</i> ⁶	5 years	Right hemihydranencephaly	Left hemiparesis strabismus, partial facial paralysis, mild mental retardation
Greco <i>et al</i> ²	4 months	Left hemihydranencephaly	Right hemiparesis, strabismus, hydrocephalus, mild mental retardation
Ulmer <i>et al</i> ⁴	36 years	Left hemihydranencephaly	Right hemiparesis, headache
Altschuler <i>et al</i> ¹²	3.5 months	Left hemihydranencephaly with mild hydrocephalus	Right hemiparesis, bilateral hypertonia, rightward head rotation
Balpande <i>et al</i> ¹³	13 years	Right hemihydranencephaly	Left hemiparesis
Present patient	21 years	Right hemihydranencephaly	Left hemiparesis

showed an absent right hemisphere replaced by cerebrospinal fluid (CSF) and an occluded right internal carotid artery (figure 1).

OUTCOME AND FOLLOW-UP

He was discharged on an oral maintenance dose of dilantin and is under follow-up.

DISCUSSION

Hydranencephaly represents a distinct disorder where both cerebral hemispheres are absent and replaced by CSF. While hydranencephaly is a rare disorder, it has been well documented in literature and is incompatible with life.

Hemihydranencephaly is an even rarer disorder characterised by unilateral absence of one cerebral hemisphere. Occlusion of an ipsilateral carotid artery takes place before the third trimester of gestation, resulting in an absent hemisphere which is replaced by CSF. However, the meninges, basal ganglia, pons, medulla, cerebellum and falx are preserved.^{3 4} The brain damage is assumed to occur after neural migration and before synaptogenesis.⁵ Predisposing factors include infections (toxoplasmosis, rubella, cytomegalovirus and herpes simplex), medications (warfarin, oestrogen, valproic acid), maternal irradiation, drug abuse (cocaine), von Willebrand's disease, immune thrombocytopenias, congenital factor X and V deficiencies twin-twin transfusions and intrauterine death of a monozygotic co-twin. In principle, the developing brain should be able to compensate for this defect via the circle of Willis and leptomeningeal collateral vessels.⁴ As this does not occur, the role of predisposing factors remains to be investigated.

To the best of our knowledge, this is the eleventh case reported to date^{2 4 6–13} and only the second case reported from India.¹³ In previous case reports, the majority of the patients had an absent left hemisphere (table 1).^{2 4 6–13}

Our patient is the third reported case with an absent right hemisphere. The other two cases with right hemihydranencephaly had only mild mental retardation whereas our patient has severe mental retardation.^{6 13} It is interesting to note that even with an absent left hemisphere a person can lead a near normal life, as illustrated by the case report of Ulmer *et al*.⁴ Also, Greco *et al* have noted that in spite of destruction of one hemisphere; language

development is still possible, thus implying that each hemisphere has adequate potential of language development.² Similarly, Carr *et al* studied 33 patients with hemiplegic cerebral palsy and demonstrated that central motor reorganisation occurred in 21 subjects.¹⁴ It would be rational to suppose that a similar motor reorganisation may be the reason why some patients with hemihydranencephaly have better motor skills than expected.

Learning points

- ▶ Hemihydranencephaly is not incompatible with a near normal life.
- ▶ Our patient has been diagnosed with hemihydranencephaly at the age of 21 years. However, in cases which are detected early (in postnatal period), perhaps a multidisciplinary intensive rehabilitation program can be initiated to realise the untapped potential of the brain.
- ▶ Given that in comparison with other destructive encephalopathies, hemihydranencephaly has a better prognosis; it would be worthwhile to employ available resources for further research into this anomaly.
- ▶ Further research into hemihydranencephaly would help in understanding the development, functioning, untapped potential and reorganisation capacity of the brain.

Competing interests None.

Patient consent Obtained.

REFERENCES

1. Myers RE. Cerebral ischemia in the developing primate fetus. *Biomed Biochim Acta* 1989;**48**:S137–42.
2. Greco F, Finocchiaro M, Pavone P, *et al*. Hemihydranencephaly: case report and literature review. *J Child Neurol* 2001;**16**:218–21.
3. Mori K. Anomalies of the central nervous system. In: Mori K ed. *Neuroradiology and Neurosurgery*. New York: Thieme-Stratton 1985:39–42.
4. Ulmer S, Moeller F, Brockmann MA, *et al*. Living a normal life with the nondominant hemisphere: magnetic resonance imaging findings and clinical outcome for a patient with left-hemispheric hydranencephaly. *Pediatrics* 2005;**116**:242–5.
5. Eyre JA, Miller S, Clowry GJ, *et al*. Functional corticospinal projections are established prenatally in the human foetus permitting involvement in the development of spinal motor centres. *Brain* 2000;**123**(Pt 1):51–64.

6. **van Doornik MC**, Hennekam RC. Hemi-hydranencephaly with favourable outcome. *Dev Med Child Neurol* 1992;**34**:454–8.
7. **Moser RP**, Seljeskog EL. Unilateral hydranencephaly: case report. *Neurosurgery* 1981;**9**:703–5.
8. **Suzuki M**, Seki H, Yoshimoto T. Unilateral hydrocephalus combined with occlusion of the ipsilateral internal carotid artery. *Surg Neurol* 1985;**24**:27–30.
9. **Warkany I**. *Congenital Malformations: Notes and Comments*. Chicago, IL: Year Book Medical 1971.
10. **Ohtsuka H**. A rare patient with a false median cleft lip associated with multiple congenital anomalies. *Ann Plast Surg* 1986;**17**:155–160.
11. **Muir CS**. Hydranencephaly and allied disorders: a study of cerebral defect in Chinese children. *Arch Dis Child* 1959;**34**:231–46.
12. **Altschuler EL**, Matsumura B, Capoor J, *et al*. Left-hemispheric hydranencephaly with less favorable findings. *Pediatrics* 2005;**116**:1603–4; author reply 1604.
13. **Balpande DN**, Pathak C, Agrawal A, *et al*. Hemihydranencephaly: a case report. *Iran J Pediatr* 2009;**19**:180–18.
14. **Carr LJ**, Harrison LM, Evans AL, *et al*. Patterns of central motor reorganization in hemiplegic cerebral palsy. *Brain* 1993;**116**(Pt 5):1223–47.

This pdf has been created automatically from the final edited text and images.

Copyright 2011 BMJ Publishing Group. All rights reserved. For permission to reuse any of this content visit <http://group.bmj.com/group/rights-licensing/permissions>.

BMJ Case Report Fellows may re-use this article for personal use and teaching without any further permission.

Please cite this article as follows (you will need to access the article online to obtain the date of publication).

Dias LS, Shivashankara KN, Vivek G. Hemihydranencephaly: rare disease with key to secrets of the brain. *BMJ Case Reports* 2011;10.1136/bcr.12.2010.3658, date of publication

Become a Fellow of BMJ Case Reports today and you can:

- Submit as many cases as you like
- Enjoy fast sympathetic peer review and rapid publication of accepted articles
- Access all the published articles
- Re-use any of the published material for personal use and teaching without further permission

For information on Institutional Fellowships contact consortiasales@bmjgroup.com

Visit casereports.bmj.com for more articles like this and to become a Fellow