Interdisciplinary Neurosurgery 16 (2019) 155–157

Contents lists available at ScienceDirect

Interdisciplinary Neurosurgery

journal homepage: www.elsevier.com/locate/inat

Letter to the Editor

Total cerebral dysgenesis - A very rare case

ARTICLE INFO

Keywords:
Cerebral
Dysgenesis
Agenesis
Cerebellum
Hydranencephaly

ABSTRACT

Introduction: Cerebral dysgenesis is the absence of formation or non-formation of a cerebellar hemisphere(s). Although many authors have described this disorder for over centuries now, no author has reported a case of total cerebral dysgenesis.

Case presentation: We present a case of a seven months old male infant who had difficulty feeding immediately after birth. He did not cry immediately after delivery. He could not move his limbs or the body. The mother did not have access to antenatal care. MRI of head revealed bilateral absence of cerebral hemispheres with normal skull vault filled with cerebrospinal fluid (CSF). The brain stem is visible, as well as part of the cerebellum. Electroencephalography (E.E.G) done revealed no electrical impulses over areas responsible for coordination of the limbs as well as the entire trunk. The kid was currently being managed conservatively. He was fed via Nasogastric tube (NG-Tube).

Conclusions: This presentation is puzzling as compare to hydranencephaly. If he miraculously advances with age, he will live with complex neurological syndromes.

1. Introduction

Esiner and Roback indicated that the use of cerebral agenesis is contradictory because agenesis means absence of formation or non-formation of an organ or of part of an organ [1] such as the report of agenesis of the corpus callosum [1,2]. They argue that malformation of the cortex or of other parts of the brain is not rare and is seen more often than non-formation. They therefore proposed the term cerebral dysgenesis to define variation in formation of the brain with resultant disorders of function of the brain or of part of the brain [1]. Looking at the imaging characteristic of our case, we think the cerebral hemispheres not form during the fetal development.

2. Case report

We report a case of seven months old male infant, who has difficulty feeding immediately after birth. He did not cry immediately after delivery. He could not move his limbs or the body. The mother did not take folic acid supplement during pregnancy. She also denied taking any teratogenic medication during pregnancy. She however did not have access to routine antennal care hence no ultrasound scan was done throughout the entire pregnancy. Her pregnancy history was uneventful and no past history of any chronic illness. Physical examination revealed an infant who does not perceive his environment. Vital signs were stable with a normal skin for his age. There were no obvious Skull defects. Head circumference slightly increased with slightly bulging anterior fontanelle. He did not cry to external stimuli and his general reaction was poor. The eyes did not follow faces or sound. Moro's, the startle, as well as grasp reflexes were present but weak. MRI of the head revealed bilateral absence of cerebral hemispheres with normal skull vault filled with cerebrospinal fluid which is seen on T1 as hypo-intense and as hyper-intense on T2. The brain stem is visible, as well as part of the cerebellum (Fig. 1A–F). These two brain structures are well developed which could be the possible reason why the kid is surviving. Electroencephalography (E.E.G) done revealed no electrical impulses over areas responsible for coordination of the limbs as well as the entire trunk. The kid was treated conservatively. He was fed through a Nasogastric tube (NG-Tube).

3. Discussion

Cerebral dysgenesis embodies a group of encephalopathies which are usually differentiated most cases from birth injury disorders. The disorders seen in instances of dysgenesis perhaps may be associated with malformation of the central nervous system in utero. Cerebral dysgenesis is frequently linked to disorders like congenital spastic diplegia, congenital atonic diplegia and diverse forms of amentia, congenital athetosis, choreoathetosis and paraplegia [1]. Pierre indicated that brain formation can chronologically be alienated into three [3] key stages which are the production of neural precursors, neural migration alone the radial glial guides and differentiation as well as survival [3]. Almost all the cases of cerebral dysgenesis report in literature have associated encephalopathies but our case is much more baffling because there are no obvious encephalopathies.

The advances in radiology make the detection of this kind of malformation much easier and early compare to later years where clinicians duel on autopsy finding. The radiology of the infant is much different from that of adults. Raybaud & Widjaja indicated that the structure of the immature brain tissue is different within the first months of life and the T1 relaxation time is much longer than in the mature brain, so modified sequences ought to be used. They observed that the T1-weighted (T1W) sequences require a longer repetition time (TR) while T2W sequences require longer TR and echo time. Furthermore, the cortex is finely outlined at birth on T1 and T2, the contrast becomes lost with increasing myelination until after 1 year on T1 and 2 years on T2 [4]. On FLAIR images the advancement is still

https://doi.org/10.1016/j.inat.2018.10.004
Received 20 June 2018; Received in revised form 30 August 2018; Accepted 8 October 2018

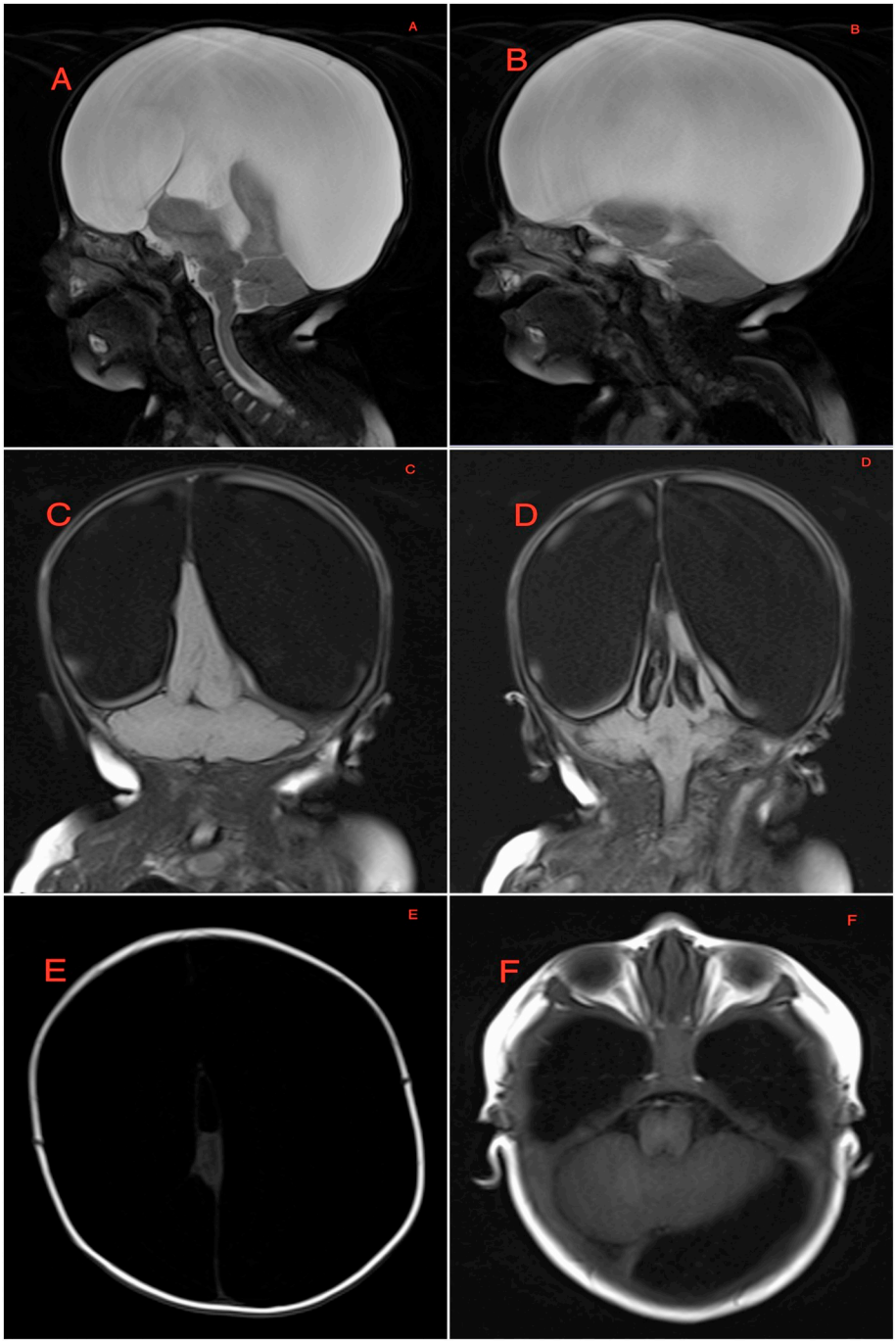

Fig. 1. A & B are sagittal T2 images showing the cranium vault filled CSF with only the brain stem and cerebellum visible. C & D are coronal T1 images showing the brain stem and cerebellum as well as the right and left ventricles with no cerebral hemispheres. E & F are axial T1 images. E; shows a small shadow of the corpus callosum and ventricles while F shows brain stem and cerebellum without temporal lobes and occipital of the cerebral hemispheres.

more multifaceted because at birth, the extremely excessive water content of the white matter is annulled by the saturation pulse and the sign is fairly like to T1 and in the subsequent weeks, the myelin precursors accrue and the signal intensifies to look more like an established T2. Nevertheless, it remains relatively heterogeneous for a much longer time than on ordinary T2 sequences, as the mature pattern is not reached until about the age of 3 to 4 years [4]. Looking at the MRI in our case, one will notice bilateral absence of cerebral hemispheres with normal skull vault filled with cerebrospinal fluid which is seen on T1 as hypo-intense and as hyper-intense on T2. The brain stem is visible, as well as part of the cerebellum. Our differential diagnosis is hydranencephaly [5]. Our patient has feed via NG-tube since birth. We are managing the patient conservatively without operation because his head circumference is not increasing in size.

4. Conclusion

Most of the cases described in literature are all about dysgenesis but not total dysgenesis or agenesis as seen in our case. We think that this presentation is not compatible with life and the patient will not live for long. If he miraculously advances with age, he will live with complex neurological syndromes.

Ethics approval and consent to participate

The ethical committee of West China hospital full approved our case study. The child's parents were informed about our intension to involve him in a case study and they agreed to partake in the study.

Consent for publication

The child's parents were dually informed about our intention to publish his case and they fully concerted to the use of these documents. A written concern for publication was signed. The hospital also concerted to the use of this information for publication.

Competing interests

All the authors have no competing interest to disclose.

Funding

Not applicable.

Authors' contributions

All the authors contributed equally to the manuscript design and writing.

Acknowledgments

Not applicable.

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