CASE REPORT

Alive with almost no Brain: Severe Post-Hemorrhagic Internal Hydrocephalus

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SUMMARY

Objectives: Severe post-hemorrhaghic internal hydrocephalus with almost complete atrophy of the cerebral parenchyma, as in the following case, is rare.

Case report: A 19yo Caucasian female with a history of premature birth, perinatal intraventricular bleeding, developmental delay, mental retardation, and epilepsy, was admitted for recurrent generalized tonic-clonic seizures. She was able to produce some noises but was unable to communicate with understandable speech. There was severe mental retardation, motor deficits, and tetraspasticity. She was able to sit and eat but was otherwise dependent on the parents' support. Monotherapy with primidon since age 15y was increased to 500mg/d. A CT scan of the cerebrum showed a massive internal hydrocephalus with atrophy of the basal ganglia, the white matter, the cerebellum, but also the cortex. Neurosurgeons decided against a shunt.

Conclusions: Despite severe atrophy of the cerebral parenchyma, severe post-hemorrhagic internal hydrocephalus, manifesting as psychomotor retardation, epilepsy, and tetraspasticity, is compatible with life.

KEY WORDS:
cerebral atrophy, epilepsy, hydrocephalus, cerebral bleeding, spasticity

INTRODUCTION

Causes of internal hydrocephalus are multifactorial and include genetic, vascular, infectious, or traumatic conditions [1,2,3]. One of the most frequent causes of infantile internal hydrocephalus is intraventricular hemorrhage, a common complication of prematurity, manifesting as cerebral palsy and cognitive decline [4]. Severe internal hydrocephalus with almost complete rarefication of the cerebral parenchyma, as in the following case, is rare.

CASE REPORT

A 19yo Caucasian female with a history of premature birth after seven uneventful months of gestation, perinatal intraventricular bleeding, developmental delay, mental retardation, and epilepsy, was admitted for recurrent generalized tonic-clonic seizures. Epilepsy started already in early childhood requiring antiepileptic treatment. Since age 15 years she was on a monotherapy with primidon (192.5mg/d), which she tolerated without obvious side effects. Since initiation of primidon she was seizure-free. The family history was negative for acquired or genetic neurological disease. Her cytogenetic investigation s was normal. The patient was able to sit and partially to eat but was otherwise dependent on the support of her parents with whom she lived since birth. On clinical neurological investigation she was able to produce some noises but was unable to communicate with words. There was severe mental retardation, motor deficits, and tetraspasticity (Ashworth scale 3) but no facial or limb dysmorphism. She scored 1 on the Everyday Speech Production Assessment Measure (ESPAM). She scored 6 on the mini-mental state examination (MMSE). Blood chemical investigations were normal. EEG showed sharp slow waves over the right occipital projection, questionable periodic lateralized epileptiform discharges over the right frontal projections, and diffuse theta slowing over the left hemisphere and delta-slowing over the right hemisphere, why primidon was increased to 500mg/d (figure 1). A CT scan of the cerebrum showed a massive internal hydrocephalus with atrophy of the basal ganglia, the white matter, the cerebellum, but also the cortex (figure 2). Neurosurgeons decided against a shunt.

DISCUSSION

Though the exact cause of the severe cerebral alterations in the presented patient remains enigmatic it is most likely that they resulted from neonatal intracerebral bleeding with ventricular intrusion. Arguments for such a pathomechanism are that the history was positive for neonatal intracerebral bleeding, that internal hydrocephalus is a frequent complication of neonatal intraventricular hemorrhage [2,5], and that post-hemorrhagic hydrocephalus in neonates can be induced in an animal model [4]. The absence of a neurosurgical intervention, like a ventriculostomy or shunt, does not exclude this pathomechanism, since such interventions are not indicated in each case. Differential diagnoses such as traumatic subarachnoidal bleeding, toxoplasmosis, or hereditary disease like pontocerebellar hypoplasia (PCH) [3], amyotrophic cerebellar hypoplasia, porencephalia or schizencephalia [1], or Dandy-Walker syndrome were excluded upon the individual and family history, laboratory tests, and the imaging findings. Particularly, the history for subarachnoid bleeding was negative, antibodies against Toxoplasma were negative, cerebral imaging was not typical for PCH, cerebellar

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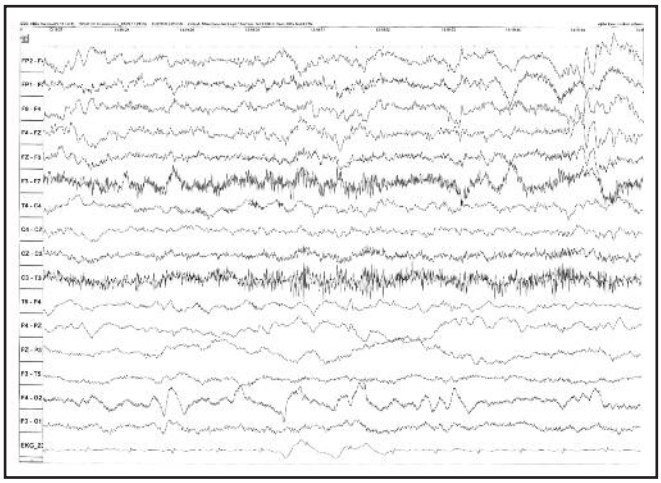

Fig. 1 : EEG showing sharp waves over the right frontal projections.

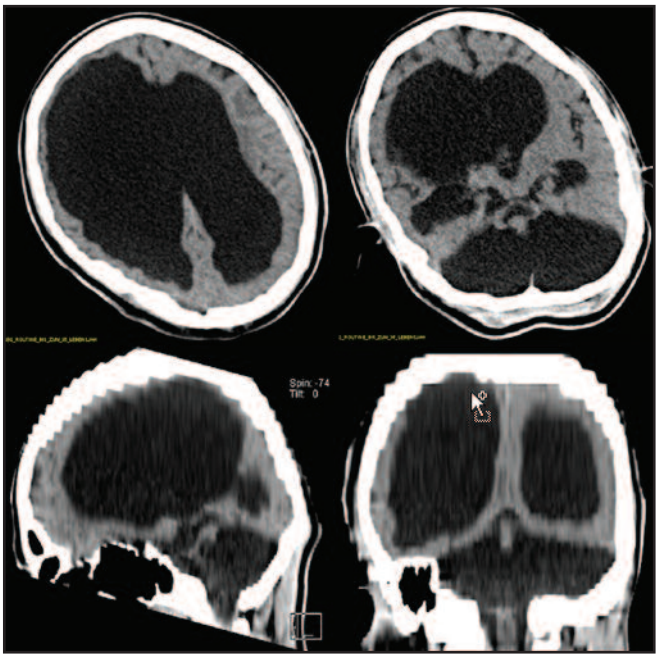

Fig. 2 : CT scan of the cerebrum showing severe internal hydrocephalus with massive atrophy of the subcortical white matter, basal ganglia, and thalamus but also of the cerebellum.

hypoplasia, porencephalia, or Dandy-Walker syndrome. Arguments for PCH, however, are that there was a small rest of the cerebellum, and that the patient presented with other features of PCH, such as developmental delay and spasticity[3]. Arguments against PCH however, are the absence of an increased respiratory rate, poor feeding, facial and hand edema, and of microcephaly.

This case shows that severe post-hemorrhagic internal hydrocephalus is compatible with life and manifests as psychomotor retardation, rare epileptic seizures, and tetraspasticity. Despite severe rarefication of the cerebral parenchyma and despite the resulting severe handicap, these patients can be integrated and can participate in family and social living.

REFERENCES

1 Bönnemann CG, Meinecke P (1996) Bilateral porencephaly, cerebellar hypoplasia, and internal malformations: two siblings representing a probably new autosomal recessive entity. Am J Med Genet 63: 428-33.

2 Koo H, Chi JG (1991) Congenital hydrocephalus - analysis of 49 cases. J Korean Med Sci 6: 287-98.

3 Rankin J, Brown R, Dobyns WB, *et al.* (2010) Pontocerebellar hypoplasia type 6: A British case with PEHO-like features. Am J Med Genet A 152A: 2079-84.

4 Chua CO, Chahboune H, Braun A, *et al.* (2009) Consequences of intraventricular hemorrhage in a rabbit pup model. Stroke 40: 3369-77.

5 Arnold D, Adis B, Rettwitz W, Lasch P, Kachel W (1988) Assessment and therapeutic possibilities in posthemorrhagic hydrocephalus of the newborn infant. Klin Padiatr 200: 299-306.