Delayed diagnosis of hydranencephaly in a nine-month-old child

Athanasios K. Petridis, Ulf R. Krause-Titz, Alexandros Doukas, Hubertus Maximilian Mehdorn

Department of Neurosurgery, University Hospitals Schleswig-Holstein, Campus Kiel, Germany

Abstract

We present a case of a child suffering of hydranencephaly. The interesting fact in the case is that there were no embryological examinations during pregnancy and therefore could the anomaly, which would have been easily avoided by regular visits to the obstetrician, not be diagnosed timely. Education of mothers is always necessary irrespective on how developed a country is and how good the medical treatment and diagnostic tools are. Every pregnant woman needs to be well educated in matters of pregnancy monitoring.

Introduction

Hydranencephaly is a severe and rare developmental defect during the embryogenesis occurring in less than 1 per 10,000 births worldwide and may be caused by vascular insults, injuries, infections, traumatic disorders after the 12th week of pregnancy or genetic failures.[1,2,3,4] Hydranencephaly is classically characterized as absence of supratentorial brain structures supplied by the anterior and middle cerebral arteries, whereby basal and infratentorial brain structures supplied by the posterior circulation might be preserved. The absent brain is replaced by cerebrospinal fluid (CSF) and the association to hydrocephalus is frequently given.[2,5,6] Diagnosis is usually made intrauterine by ultrasound (US) and/or magnetic resonance imaging (MRI) and leads normally to abortion.[7,8] We present a case of a 9 months old child with delayed diagnosis of hydranencephaly.

Case Report

A 9-month-old boy of a western European woman presented in our department with progressive gain of the head circumference after a medically non-observed pregnancy and a home delivery. The mother of the baby had a known psychiatric previous history but no other relevant medical conditions and did not want the pregnancy. As a result she did not seek regular medical supervision and gave up the baby directly after birth to a foster home. Even postpartum, the child was seen infrequently by pediatric physicians. The only response on speaking or acting of the kid was gripping with the left hand. He showed neither eye opening nor movement of the lower extremities. He presented several clinical signs for cerebrospinal hypertension, such as sunset phenomenon, venous marmoration and exposed fontanels. Furthermore he developed diabetes insipidus, an adrenal insufficiency, recurrent seizures and central temperature regulation disorders. Magnetic resonance imaging (MRI) of the brain (Figure 1) showed complete agenesis of the supratentorial brain structures replaced by CSF in terms of a hydranencephaly. Infratentorially the pons, the medulla oblongata and the left cerebellar hemisphere had only been developed.

After a long debate on ethics considering the incompatibility of this brain defect to a proper development we implanted a ventriculo-peritoneal shunt to prevent further gain of the head circumference. Few months later the boy could move all extremities and make unintelligible sounds. In consideration of the disease and its prognosis we desisted from further follow up examinations and therapy.

Discussion

Hydranencephaly is a severe and very infrequent developmental brain defect. The clinical presentation consists of a vegetative state with few reflexive functions like sucking, swallowing, crying, and moving the extremities, depending on the severity of the condition. The prognosis is poor with usually fatal outcome during the first year.[1,2,9,10]

Despite the feasibility to diagnose hydranencephaly intrauterine via US and / or MRI in the regular examinations by an obstetrician, delayed diagnosis is also possible, because the clinical status and facial configuration of the newborn can appear relatively normal at first.[1,3,11]. Therapy options are limited and the necessity of shunting in order to control the growth of head size has to be considered.[12]

A non-observed pregnancy, a birth at home and delayed diagnosis of this severe clinical condition in Europe is very remarkable. It is justified to reflect upon medical termination of the pregnancy to prevent maternal morbidity after an early diagnosis of hydranencephaly.

Conclusions

It is very important to keep in mind that even abnormalities like hydranencephaly can

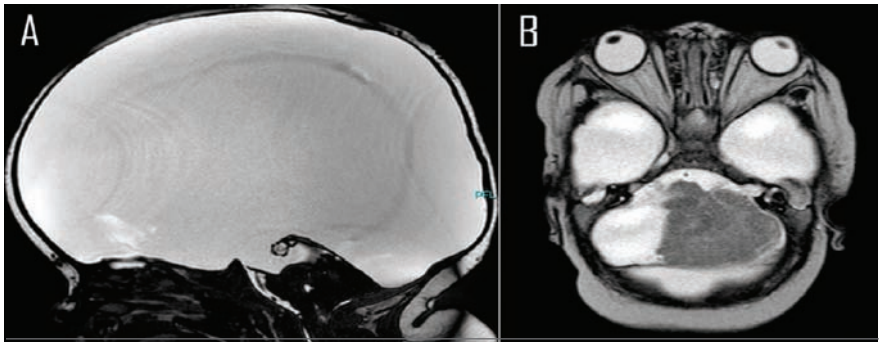

Figure 1. A) Sagittal magnetic resonance imaging (MRI) of the child's brain showing the lack of neocortex. Only the brain stem seems to exist maintaining the vital functions; B) In the transverse MRI there is a small part of brain which does not seem to have any structure.

Correspondence: Athanasios K. Petridis, Klinik für Neurochirurgie, Universitätsklinikum Schleswig-Holstein, Campus Kiel, Arnold-Heller-Straße 3, 24105 Kiel, Germany.
Tel. +49.431.597.8550.
E-mail: opticdisc@aol.com

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escape early diagnosis and lead to the delivery of a child which is highly handicapped. The social and psychological problems that can occur after a delivery of such a child can be devastating for the family. Despite a very efficient pregnancy monitoring system like the existing one in Europe, there are still families which do not visit the obstetrician and avoid monitoring during pregnancy. We would like to stress out the importance of educating mothers to participate in the child examination programs during pregnancy.

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