Journal of Pediatric Diseases (2018) Volume 2
doi:10.24294/jpedd.v2i2.978

The first two years with hydranencephaly: an unusual medical course of a male child with a missing part of the brain

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ABSTRACT

Hydranencephaly is an extremely rare anomaly of cerebral structures that occurs in-utero. Cerebral hemispheres are destroyed with transformation into a membranous sac with cerebrospinal fluid and parts of cortex and white matter. The disorder has an incidence of 0.2% in children. Clinical symptoms in neonates are seizures, respiratory failure, flaccidity or decerebrate posturing. Complete absence of the cerebral hemispheres and falx are present. We present a case of a male newborn with hydranencephaly. A possible correlation with in utero exposure to vasoactive drugs will be discussed.

Keywords: Hydranencephaly; child; drug abuse

1. Introduction

Hydranecephaly is an extremely rare condition in which the brain's cerebral hemispheres are completely absent and replaced by sacs filled with cerebrospinal liquid. A child with hydranencephaly may appear like a normal newborn at birth. The head size and spontaneous reflexes may all seem normal. After a few weeks of birth the child becomes irritable and has higher muscle tonus. Seizures and hydrocephalus may develop. Other symptoms like visual impairment, growth delay, amaurosis, quadriplegia and intellectual deficits may occur. Hydranencephaly is an extreme type of porencephaly and may be caused by vascular disorders after the 12th week of pregnancy. Some infants may have additional abnormalities at birth including seizures, myoclonus and respiratory problems. The prognosis for children with hydranencephaly is bad, and many children with this disorder die before age 12 months. However, in some cases, infants with hydranencephaly may survive for several years.

2. Case Report

We report of a male newborn with a birth weight of 3200g. Surprisingly, pregnancy was not mentioned by the mother. She went to the gynecologic department due to lower abdominal pain. APGAR-score was 10 in 5 minute. Spontaneous delivery was performed in the 40th week, birth length was 52 cm and head circumference 34,5 cm. Umbilical postnatal arterial pH was 7.22. Marihuana and cigarette abuse (10/day) during pregnancy by the mother was described. Unknown status of hepatitis b of the mother lead to simultaneous vaccination of the child. Postpartially adaption was good, Spontaneous reflexes were normal initially. 2/6 heart murmur was found at the third day of life. Examination revealed a small muscular ventricle septal defect. A marmorated skin colour and muscular rigidity was also found. MRI revealed a hydranencephaly with normal primary branches of arteria carotis interna bilaterally. More distal of these arterial branches no vessels were found, so a closure of the distal parts of the arteria carotis interna was discussed. A ventriculoperitoneal shunt with rickham reservoir without pressure valve was inserted. Postoperative course was uneventful. CMV-, rubella- and toxoplasmosis-antibodies were not found. Blood specimen, blood sugar and blood gas analysis was normal. To date of publication, aged 4 months, the child is in a good condition. The child shows surprisingly spontaneous muscular movements, does not fix with the eyes but gains weight. Intensive care at home is necessary.

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3. Discussion

Hydranencephaly is an extreme rare congenital defect. It can occur after meningitis, intracerebral infarction, ischemia and injury of the brain. Hydranencephaly is classified as porencephaly, where cerebrum and falx are completely absent. Differential diagnosis includes schizencephaly, severe hydrocephalus and holoprosencephaly. When destruction of the brain is complete, the cerebellum, midbrain, thalami, basal ganglia and choroid plexus typically remain preserved to varying degrees. In most cases the fetal head remains enlarged due to the continued production by the choroid plexus of cerebrospinal fluid[1]. The exact cause of hydranencephaly remains unclear in most cases, the most often found cause is vascular insult such as stroke or injury, intrauterine infections, or traumatic disorders after the first trimester of pregnancy. Intrauterine infections are also described as causing factors. Toxoplasmosis and viral infections such as enterovirus, adenovirus, parvovirus, cytomegalic, herpes simplex, Epstein-Barr, and syncytial viruses are possible infectious causes. Another cause factor is described to be monochorionic twin pregnancies, involving the death of one twin in the second trimester, which in turn causes vascular exchange to the living twin through placental circulation through twin-to-twin transfusion, causing hydranencephaly in the surviving fetus[1]. An accurate, confirmed diagnosis is generally impossible until after birth. Prenatal diagnosis by fetal ultrasonography can identify characteristic physical abnormalities. Clinical evaluation with detailed patient history, and advanced imaging techniques, such as angiogram, CT and MRI are the most accurate diagnostic techniques[1,2]. Medical text identifies that hydranencephalic children simply have only their brain stem function remaining[1] thus leaving formal treatment options as symptomatic and supportive. Severe hydrocephalus causing macrocephaly can easily be managed by placement of a shunt and oftentimes displays a misdiagnosis of another lesser variation of cephalic condition[1]. Plagiocephaly, the asymmetrical distortion of the skull, is another typical associated condition that is easily managed through positioning and strengthening exercises to prevent torticollis, a constant spasm or extreme tightening of the neck muscles[4]. Medical research exclusive to hydranencephaly is limited. There are research efforts in the realm of neural tube defects and prevention of congenital neurological conditions[5].

References

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