EVOLUTION OF A VARIANT DANDY-WALKER SYNDROME

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Abstract

The paper presents the evolution of a twenty-one-month-old male child, who was diagnosed, since he was an infant (5 months), with an incomplete Dandy-Walker syndrome.

Key words: incomplete Dandy-Walker syndrome, child, evolution.

Introduction

Classically, the incomplete Dandy-Walker syndrome is characterized by the vermis agenesis, the cyst dilatation of the fourth ventricle due to the rostral movement and the absence or atresia of the Magendie and Luschka foramens.

Hydrocephaly is not usually present from the congenital point of view, but it develops in the first months of life. 90% of the patients who develop a hydrocephaly are registered before the age of 1 year.

There are also incomplete variants, where the cerebellar hypogenesis is present without the dilatation of the forth ventricle and hydrocephaly. An ataxic syndrome occurs in less than 20% of the patients and it is usually late. The diagnosis of the Dandy-Walker malformation is confirmed by CT or skull RMN.

Case presentation

We present the case of the patient J.F.A., a male infant aged 1 year and 9 months, diagnosed ever since he was 5 months with variant Dandy-Walker syndrome, and who was admitted in 2nd Pediatric Clinic of the County Emergency Hospital, in February 2006 (O.F. 9442/2006).

The occurrence of balance and clinical-biological assessment dysfunctions represented the reasons of the admission.

The anamnesis revealed that he is the only child of a young couple, apparently healthy, with a higher education level. At the antenatal ultrasonography (third semester) he was diagnosed with megacysterna magna, cerebellar asymmetry. The child was born at 32 weeks (premature level I) with 2,400g in weight, Apgar =7/8, (perinatal ischemic hypoxia without requiring advanced resuscitation procedures at birth).

L.P. was artificially fed, diversified at 6 months, he took his vaccines according to the WHO scheme, and he followed the rachitism prophylaxis.

Belated psychomotor development: he held his head at 8 months, he could sit at 11 months, he could walk by himself at 1 year and 7 months, he uttered his first word at 1 year.

APP: bilateral inguinal-scrotal hernia (since he was 3 months old), variant Dandy-Walker syndrome starting with the age of five.

The anamnesis also mentions the fact that at the age of five he was diagnosed with incomplete Dandy-Walker syndrome using a skull CT which indicates the cerebellar vermis and hemispheres hypoplasia, especially on the left side, replaced by an arachnoidian cyst that communicates with the fourth ventricle [fig.1]. A ventricular system with occipital remote horns because of an interpontin cyst [fig.2]. Cerebellar spaces that are slightly symmetrically enlarged [fig.3].

The transfontanel ultrasonography, which was performed at the age of 5 months, shows the presence of a callous body, cerebellar hemispheres and vermis hypoplasia [fig.4]. The clinical picture at admission showed: relatively good general state, 11Kg in weight, without fever, cardio-circulatorily, respiratorily and digestively stabilized, inguinal-scrotal hernia; he also presented a normally conformed skull, closed FA, normal active and passive movements, muscular discreet hypotony, unsupported walk with balance dysfunctions, watching things, and poor vocabulary.

Biological assessments: HLG: Hb=12,20g%, Ht=37%, T=190000/mm^3, L=8000/mm^3, Ns=28%, E=4%, Ly=62%, M=6%.

F.O. (AO): normally outlined papilla, blood vessels and retina with a normal aspect.

Skull X-ray: no modifications of the bony structures at the neurocranium level, closed fontanels, visible sutures.

A skull CT at the age of one year and nine months was refused by the parents.

Neurological consult: discreet axial hypotony, psychomotor retard.

The treatment involved a neuro-motor recovery within the specialized service of the hospital (medical exercises).

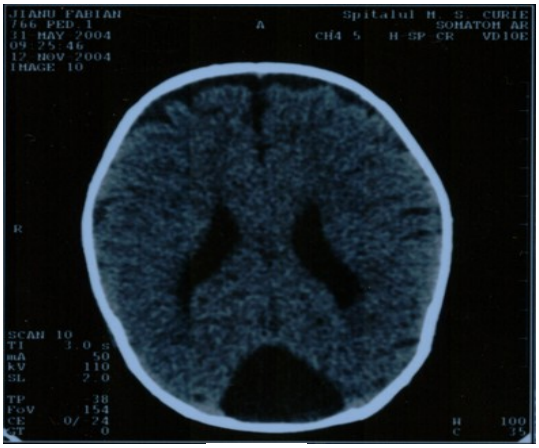

Fig. 1

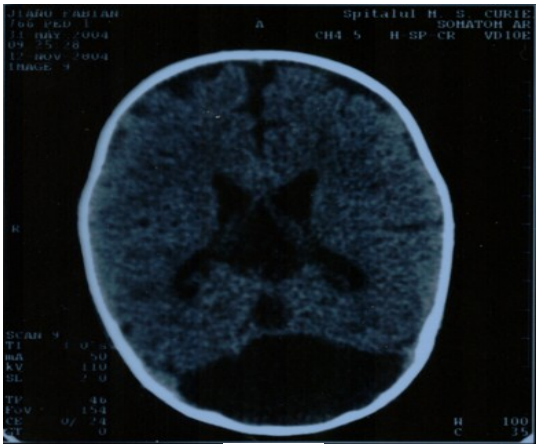

Fig. 2

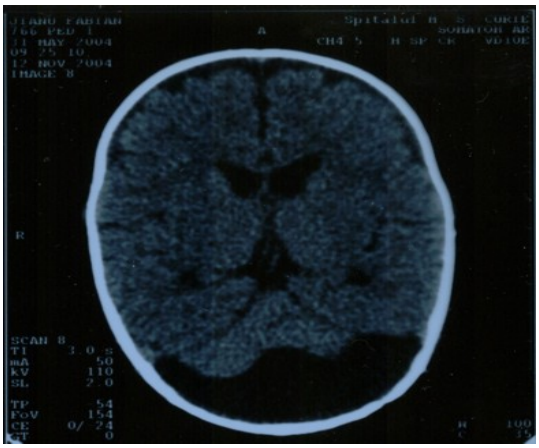

Fig. 3

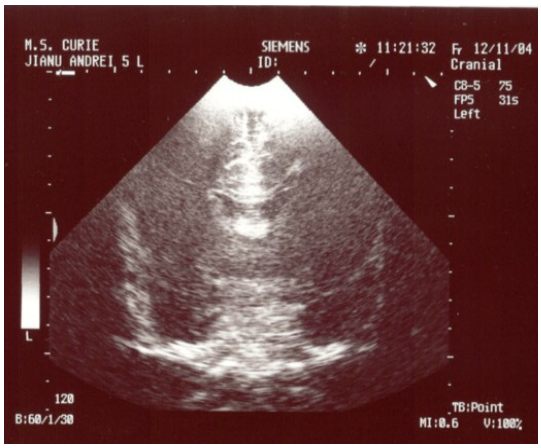

Fig. 4

Discussions

Classically, the Dandy-Walker malformation can be defined by: the cyst dilatation of the fourth ventricle, vermis agenesis, the absence of the foramens Magendie and Luschka.

However, there are variants of the Dandy-Walker malformation, with moderate dilatation of the fourth ventricle, the permeability of the Magendie foramen, and the partial vermis agenesis.

In the presented case, the antenatal ultrasonography (third semester) on the pregnant uterus showed megacysterna magna and cerebellar asymmetry, without performing amniocentesis or placental villus biopsy for the genetic study of this anomaly.

After birth, in the first year of life, the child did not develop an important hydrocephaly with its consequences: macrocephaly (PC at 5 months = 43cm, at 1 year and 9 months, PC=48cm, FA closed at 1 year and 2 months) or intracranial hypertension (ICH).

However, the evolution of the child presented a psycho-motor retard (he held his head at 8 months, he could sit at 11 months, he could walk by himself at 1 year and 7 months, he uttered his first word at 1 year). Two months after he could walk by himself, there appeared some balance dysfunctions and we took into consideration an ataxic syndrome, which can be noticed in less than 20% of the patient with Dandy-Walker syndrome

The presented case responds to the conditions of a variant Dandy-Walker syndrome where the cranial-cerebral exam described: hypogenesis (partial agenesis) of the cerebellar vermis and of the cerebellar hemispheres, especially the left ones, which were replaced by the arachnoidian cyst that communicates with the fourth ventricle, a ventricular system with occipital remote horns because of an interpontin cyst, cerebellar spaces that are slightly symmetrically enlarged.

Conclusions

1. The case was presented because it can be included in a variant Dandy-Walker syndrome, with low incidence of the affection in practice.
2. It shows the importance of the antenatal ultrasonography on the pregnant uterus as well as of the study of the chromosomal anomalies through amniocentesis or placental villus biopsy for the genetic advice (the transmission is autosomal recessive in the case of the Dandy-Walker syndrome).

3. The psychomotor retard in the case of a variant Dandy-Walker syndrome, even without developing hydrocephaly, remains important.
4. The possibility of developing an ataxic syndrome described in less than 20% of the patients within the Dandy-Walker syndrome.

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