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Case report

DANDY WALKER AND EXTREME MACROCEPHALY CAUSED BY ENORMOUS OCCIPITAL ENCEPHALOCELE

СИНДРОМ НА DANDY WALKER И МАКРОЦЕФАЛИЈА ПРИЧИНЕТА ОД НЕЛЕКУВАНА ОКЦИПИТАЛНА ЕНЕФАЛОЦЕЛА

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Abstract

Introduction. Dandy-Walker syndrome is a congenital brain malformation involving cerebellum with partial and complete vermian agenesis, enlargement of the fourth ventricle and surrounding fluid spaces, cyst formation in posterior cranial fosse pushing tentorium upward [1,2]. Hydrocephalus or an increase in the pressure of the fluid spaces may also be present or other malformation as corpus calosum hypoplasia or agenesia, occipital encephalocele, malformation of the heart, face, limbs fingers and toes [3-5].

The symptoms often occur in early infancy and include slow motor development and progressive enlargement of the skull. The diagnostic is done by ultrasound, CT and MRI [6-11].

The treatment of this syndrome may be complex and sometimes includes various experts such as pediatrician, pediatric neurosurgeon, physiatrist, psychologist, sociologist or others. The treatment consists of treating the associated problems such as hydrocephaly [12-15]. Prognosis of Dandy-Walker syndrome is variable and the morbidity and mortality depends on severity of the syndrome and associated malformations [16].

Aim. The aim of this paper was to demonstrate how severe spontaneous evolution of Dandy-Walker syndrome may be expressed and the problems and dilemmas which may appear related to its treatment.

Case report. A six-year-old boy was referred to the neurosurgeon because of the excessive growth of the skull in anteroposterior axis caused by a wide base occipital encephalocele. Although the psychological development was near the low limit of the IQ, the enormous head had not allowed verticalization of the child and further progress of his psychomotor development. The head was so heavy that could not be supported by the child's neck.

Surgical procedure. We performed a cranial skull reduction with primary cranioplasty assisted by a plastic surgeon and Pudentz shunt procedure.

Result. The follow-up period lasted two years. The child started to walk, hypotonia and Babinski signs disappeared, communication and his IQ improved. The esthetic results are quite acceptable allowing him better development.

Conclusion. The early recognition of anomalies such as Dandy-Walker syndrome with occipital encephalocele using ultrasound may suggest interruption of the pregnancy on time [6-9]. However, the right diagnostic procedure for detecting deformities of the newborn and infant's head at birth is MRI, and the adequate surgical treatment can prevent abnormal and excessive growth of the skull and disorders in the psychomotor development during child's growth. A multidisciplinary approach may prevent new disabled individuals in the society.

Keywords: Dandy-Walker syndrome, excessive macrocephaly, occipital encephalocele, surgical treatment

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Апстракт

Вовед. Синдромот на Dandy-Walker подразбира конгенитална малформација на мозокот, која го зафаќа малиот мозок со делумна и со комплетна агенезија на вермис, зголемување на четвртата мозочна комора и околните ликворни простори, формирање циста во задната черепна јама, туркајќи го нагоре тенториумот [1,2]. Хидроцефалија или покачување на притисокот во ликворните простори, а можни се и други малформации како агенезија или хипоплазија на корпус калозум, окципитална енцефалоцела, малформација на срце, лицето, екстремитетите и прстите [3,4,5].

С симптомите, често настануваат рано кај новороденчето, и вклучуваат бавен моторен развој и прогресивно зголемување на черепот. Дијагнозата се

поставува со ултразвучен преглед, компјутеризирана томографија и магнетна резонанца [6-11]. Третманот на овој синдром е комплексен, и некогаш вклучува разни специјалности, како што се педијатар, детски неврохирург, физијатар, социолог и др. Третманот се состои од третман на пропратните проблем како хидроцефалијата [12-15]. Прогнозта на синдромот на Dandy-Walker е варијабилна, морбидитетот и морталитетот зависат од тежината на синдромот и придружните малформации [17].

Приказ на случај. Шестгодишно машко дете беше упатено кај неврохирург, поради прекумерен раст на черепот во антеро-постериорен правец, предизвикан од нелекувана окципитална енцефалоцела на широка база. И поред психички развој, со коефициент на интелегенција блиску до долната граница на нормалата, големата глава не дозволуваше вертикализација на детето и натамошен психомоторен развој. Главата тежеше премногу за да биде држена од детскиот врат.

Хируршки третман. Ние реализиравме редукција на черепот со примарна краниопластика, со помош на хирург специјалист за пластична и реконструктивна хирургија, и вградивме Pudentz цистоперитонеален шант.

Резултати. Периодот на следење изнесува две години. Детето почна да оди, исчезнаа хипотонијата и знаците на Бабински, комуникацијата се подобри како и коефициентот на интелигенција. Естетските резултати се прилично прифатливи овозможувајќи му подобар развој.

Заклучок. Раното препознавање на овие аномалии, како синдромот на Dandy-Walker, со окципитална енцефалоцела, употребувајќи ултразвучен преглед може да оди во прилог на навремен прекин на бременоста [6-9]. Вистинската дијагностичка процедура за дијагностицирање деформитети на главата на новороденото е магнетната резонанца, а адекватниот оперативен третман може да спречи ненормален и прекумерен раст на черепот и пореметување во психомоторниот развој за време на раст на детето. Мултидисциплинарниот приод може да спречи појава на нови хендикепирани лица во општеството.

Клучни зборови: синдромот на Dandy-Walker, макроцефалија, окципиталма енцефалоцела, оперативно лекување

Introduction

Dandy-Walker syndrome is a congenital brain malformation involving cerebellum with partial and complete agenesia of vermis, enlargement of the fourth ventricle and surrounding fluid spaces, cyst formation in posterior cranial fosse, enlargement of posterior fosse pushing tentorium upward [1,2]. Hydrocephalus or an increase in the pressure of the fluid spaces may also be present. Corpus calosum hypoplasia or agenesia, occipital encephalocele and malformation of the heart, face, limbs fingers and toes may also be seen [3-5]. Dandy-Walker malformation is estimated to affect 1 in 20,000 to 30,000 newborns [1,18,19]. Research suggests that Dandy-Walker malformation could be caused by environmental factors that affect early development before birth. For example, exposure of the fetus to teratogens may be involved in the development of this syndrome [1,19]. In some cases a gene mutation may be found [1,19,20]. The symptoms often occur in early infancy and include slow motor development and progressive enlargement of the skull. In older children, symptoms of increased intracranial pressure and signs of cerebellar dysfunction predominate. Breathing problems, occulomotor nerve disorders, or other lower cranial nerves may be present [12-16]. Antenatal and postnatal diagnostic is done by ultrasound and MRI [6-11].

The treatment of this malformation may be complex and sometimes include various experts such as pediatrician, pediatric neurosurgeon, physiatrist, psychologist, sociologist or others. The treatment consists of treating the associated problems such as hydrocephaly (neuroendoscopy, implanting shunt), and may include other forms of therapy such as physical, occupational, and specialized education [12-15].

The prognosis of Dandy-Walker syndrome is variable with some children having normal cognition and others never achieving normal intellectual development even when hydrocephalus is treated [17]. The morbidity and mortality depends on severity of the syndrome and associated malformations. The existence of multiple malformations may shorten the life span.

The **aim** of this paper is to present this very rare case of Dandy-Walker malformation with enormous posterior fosse cyst and macrocephaly and to demonstrate how severe spontaneous evolution may be expressed and the problems and dilemmas which may appear related to the treatment.

Case report

A six-year-old boy was referred to the neurosurgeon because of the excessive growth of the skull in anterior and posterior axis, or dolychocephaly. Although the psychological development was near the low limit of the IQ, the enormous head did not allow verticalization of the child and sustained further progress of his psychomotor development.

His cranial perimeter was 1046 mm and he presented with bilateral hypotonia and bilateral Babinski sign. The computerized tomography showed partial ossify-

cation of the parietal and upper and lower occipital part of the cyst while middle occipital part of the cyst was not covered with bone. The distance of anteroposterior axis of the head was 332 mm; there was no cerebellar vermis with voluminous posterior fosse cyst and there was moderate dilatation of supra-tentorial ventricles. The diagnosis of Dandy-Walker syndrome was confirmed by MRI. MR veinography was performed in order to see the disposition of lateral sinuses.
The lateral sinuses were pushed upward to the parietal region together with the tentorium. We had no other data because the child was coming from an orphanage and was abandoned by his parents.

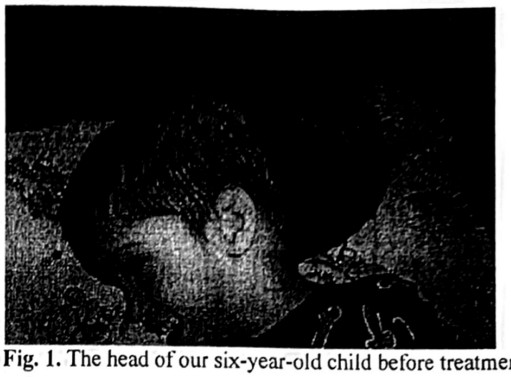

Fig. 1. The head of our six-year-old child before treatment

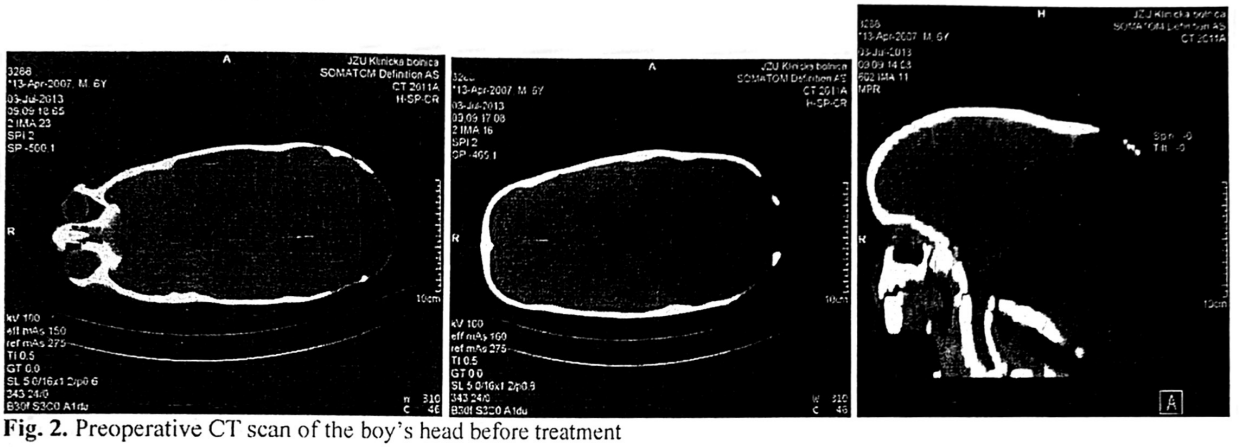

Fig. 2. Preoperative CT scan of the boy's head before treatment

Surgical procedure

The procedure was done under general endotracheal anesthesia with the child placed in a prone position with the head in Mayfield holder. The procedure was started with vertical median skin incision from the parietal to the lower occipital region. Supraperiosteal skin dissection was realized creating two skin flaps to the retroauricular region.The periostium was dissected from the dural tissue and the edge of bone defect followed by dissection of the periostium and dura from both sides of the parietal and lower part of the occipital bone to the lateral sinuses. Reducing the bone to 1 cm below the lateral sinuses we exposed the major part of the cyst and we opened it. Two large cores of ossification were left between periostium and dura in posterior parietal region right after lateral sinuses, one for each side for further cranial reconstruction. We observed a giant cyst covered with arachnoid. Excision of the free arachnoid was made and the cyst was opened. The forth ventricle and Silvius aqueduct were wide opened. Excision of the thick arachnoid free wall of the cyst was performed followed by diminishing the dura. Two large cores of ossification left between periostium and dura in posterior parietal region were rotated and used for reconstruction of the posterior wall of the vault; "Water-tide" closure of the dura was performed by 4-0 polypropylene suture. We proceeded with reduction of the excess skin and cranioplastic closure of the skin with interrupted Blair-Donatti 4-0 polypropylene sutures, without using epicranial drainage. Later a cysto-peritoneal Pudentz middle pressure shunt was inserted.

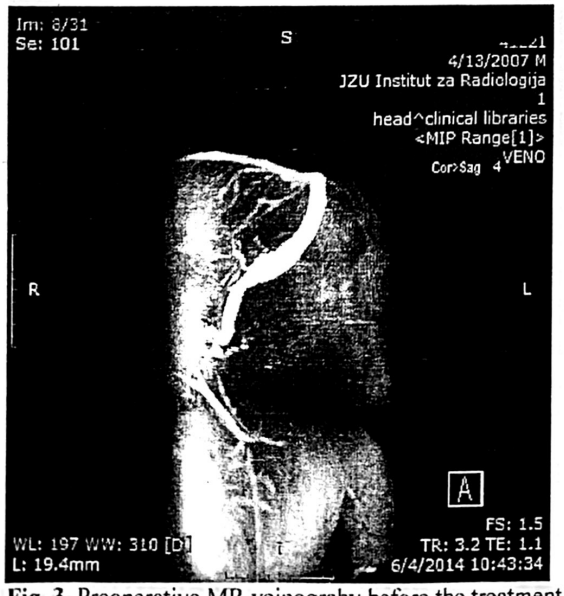

Fig. 3. Preoperative MR veinograhy before the treatment

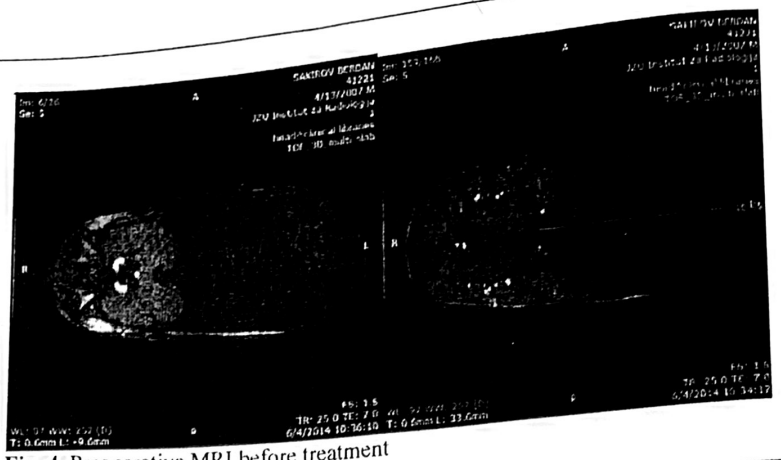

Fig. 4. Preoperative MRI before treatment

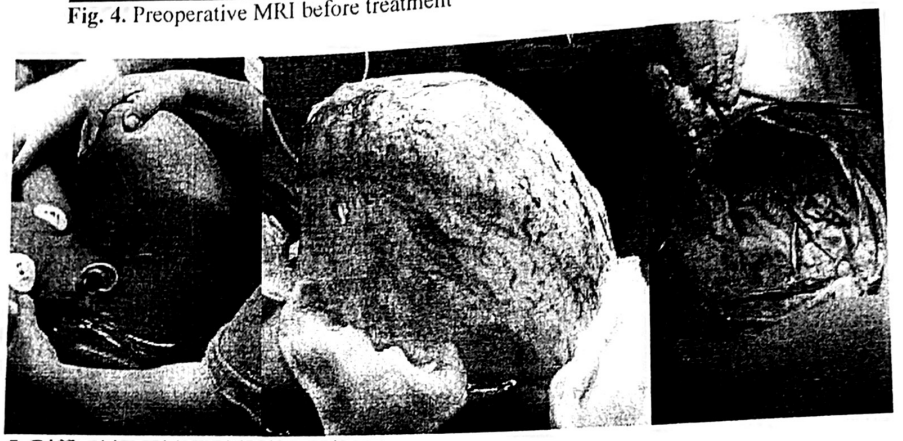

Fig. 5. Difficulties with positioning and surgery

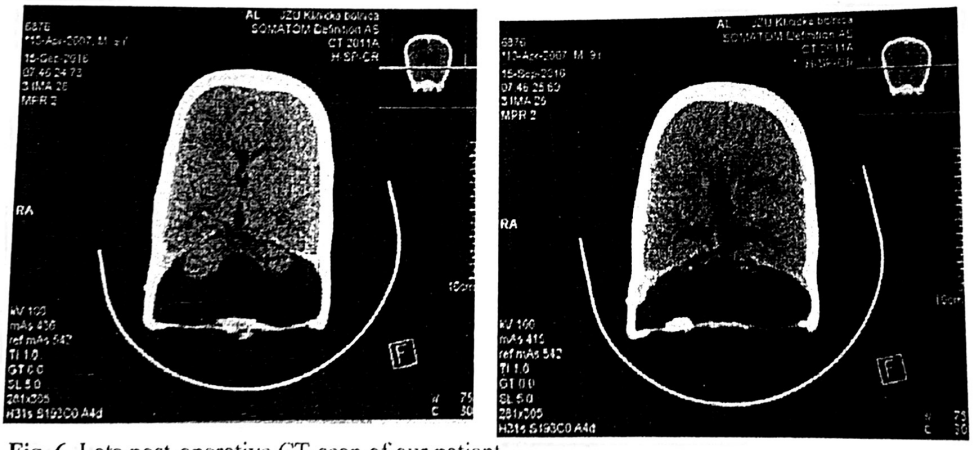

Fig. 6. Late post-operative CT-scan of our patient

Results

In the early postoperative period the child had nausea and vomited for 4 days. We achieved to verticalize the child on the 18th day after surgery helping him from the side. Three weeks after surgery the child was released from the hospital. Physiotherapy was applied in the parent institution. The follow-up period lasted two years. The child started to walk, hypotonia and Babinski signs disappeared, communication and his IQ improved. The esthetic results are quite acceptable allowing the child better development.

Fig. 7. Late postoperative photo of our patient

Discussion

Early prenatal diagnostic of Dandy-Walker syndrome is very important in order to interrupt the pregnancy on time. If the early diagnostic fails we are facing a baby with Dandy-Walker syndrome. Antenatal diagnostic has been improved in the Republic of Macedonia, but in this case it is obvious that failed.

The treatment of this malformation may be complex and sometimes includes various experts such as pediatriccian, pediatric neurosurgeon, physiatrist, psychologist, sociologist or others. The treatment consists of treating the associated problems such as hydrocephaly. A shunt procedure is to be inserted as soon as possible after establishing a diagnosis of Dandy-Walker syndrome to avoid excessive growth of the skull. Direct surgical approach may be realized if other local compressive problems exist including Silvius aqueduct narrowing. It is very rare to meet a case with Dandy-Walker syndrome with occipital encephalocele and excessive macrocephaly as ours. It is obvious that treatment at birth would have been the best option for this child.. But, this child was abandoned, with unsolved parental responsebility for a long period and hence he was referred to a neurosurgeon very late, at the age of 6.5 years. The major problem in this case was that the child had giant macrocephaly for more than 6 years. Facing the difficulties and the possible complications [21-23], the main question was whether to undertake any procedure and treatment or not. If we did not do anything in this case, then his further development would have been blocked because of a lack of ambulation, communication, etc... Thus, we took a risk and decided to diminish the head exciding the large part of the cyst, dura, bone and the skin, performing a primary cranioplasty in collaboration with a plastic surgeon and then inserting the Pudentz middle pressure shunt. Modulated shunt was to be taken into consideration, but we do not have such devices.

Conclusion

The early recognition of anomalies such as Dandy-Walker syndrome by using ultrasound may suggest interruption of the pregnancy on time. However, the right diagnostic procedure for detecting the deformities of the newborn and infant's head at birtht is MRI, and the adequate surgical treatment, which can prevent abnormal and excessive growth of the skull and disorders in the psychomotor development during child's growth. A multidisciplinary approach may prevent new disabled individuals in the society.

Conflict of interest statement. None declared.

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