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GYNAECOLOGY
Case Report

The Value of the Ultrasound in the Antenatal Diagnosis of Dandy Walker Syndrome

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Abstract

The Dandy-Walker syndrome is a rare and severe cranial abnormality with complex disorders. It includes a posterior fossa cyst and a partial or complete absence of the cerebellar vermis. There are a lot of associated cranial and extra-cranial abnormalities and the prognosis is related to the severity of the associated anomalies. Here, we report two cases of Dandy-Walker syndrome diagnosed during 23rd and 24th weeks of gestation. Delivery was vaginal in each case. Causes, risk factors and diagnosis of Dandy-Walker syndrome are discussed referring to the presented literature.

Keywords: *Dandy-Walker; Brain; Congenital malformations*

Introduction

The Dandy-Walker syndrome is a rare and severe cranial abnormality with complex disorders. It includes a posterior fossa cyst and a partial or complete absence of the cerebellar vermis [1,2]. There are many associated cranial and extra-cranial abnormalities and the prognosis is related to the severity of the associated anomalies.

This syndrome is frequently associated with multiple neurological and extra-neurological deformations. The prenatal diagnosis of the syndrome of Dandy-Walker is made by the obstetric ultrasound. However, this diagnosis can be lacking in certain asymptomatic cases. Besides, the prognosis of this pathology is multi-factorial and the obstetric management is no consensual.

Case Report

Case 1

Mrs Z.F.,29 years old G3P2 with personal history of unexplained abortion in the first term of gestation, was sent to us for an important fetal hydrocephalus that was discovered during an obstetric ultrasound made in 23 weeks of amenorrhea.

The clinical and obstetric examination highlighted an uterine height increased, estimated at 26 cm, present fetal heart sounds and absent uterine contractions. The obstetric ultrasound showed a moderate hydrocephalus associated with an important cystic dilation of the fourth ventricle, an expulsion upward the tent of the cerebellum as well as a cerebellar agenesis. (Figure 1,2) An amniocentesis was made and showed a normal karyotype (46 XX). A biological and infectious status returned negative.

A therapeutic interruption of the pregnancy was refused by the patient. The vaginal delivery at 37 weeks of pregnancy is made and a female fetus was born .Her birth weight was 3100g. The APGAR score was 5-7-8 and she had dolichocephaly and polydactyly (Figure 3). The newborn child was hospitalized in Neonatology unit for obvious axial hypotonia. She died after 21 days of hospitalization in an infectious context.

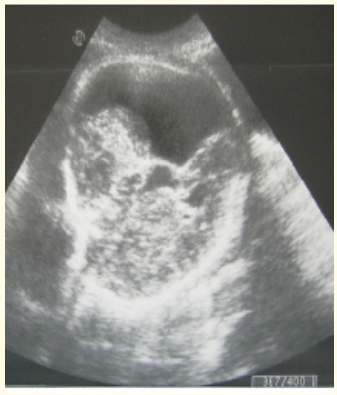

Figure 1: *Obstetrical ultrasound in axial section showing hydrocephalus, a major expansion of the posterior fossa with displacement of the supratentorial structures.*

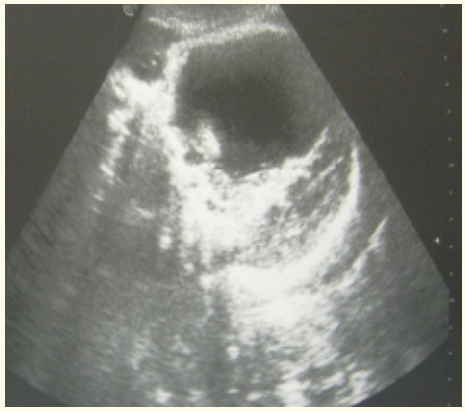

Figure 2: *Sagittal section of an obstetrical ultrasound showing a large cystic dilatation of the fourth ventricle associated with cerebellar agenesis.*

Case 2

Mrs A.H, 24-year-old, primipara, without pathological histories, having a link of consanguinity of 2nd degree with her husband, consulted us after 24 weeks of pregnancy. The clinical and obstetric examination highlighted a decreased uterine height , estimated at 19 cms, the foetal heart sounds were present whereas the uterine contractions were absent. The obstetric ultrasound highlighted a severe oligohydramnios associated with a cystic dilation of the posterior fossa repressing the cerebellum upward (Figure 4), hypoplastic lungs as well as two big multicystic loins.

An amniocentesis was made and revealed no chromosomal abnormality. A biological and infectious status containing a fasting and post-prandial glycemia as well as a serology of the toxoplasmosis, of the rubella, the syphilis and the cytomegalovirus returned negative. A therapeutic interruption of the pregnancy was made to the 25 weeks of pregnancy with the vaginal delivery of a male, his birth weight was 700g. He died in the immediate post-partum.

Figure 3: *Birth malformations with dolichocephaly, polydactyly and syndactyly.*

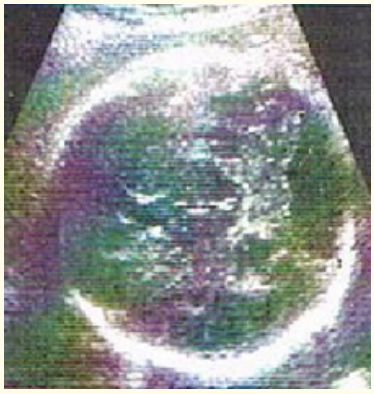

Figure 4: *Cystic dilatation in the posterior fossa.*

Discussion

The Dandy-Walker syndrome is a central nervous system anomaly. It was potentially severe associating a hydrocephalus, a complete or partial defect of the vermis and a cyst of the posterior fossa in communication with the 4th ventricle [1,2]. It is a relatively rare pathology with an incidence going of 1/25000 to 1/35000 births [3,4].

It is often admitted that the syndrome of Dandy-Walker corresponds to a malformative complex set in touch with a defect of development of the central nervous system arisen before the 6th or the 7th weeks of pregnancy [3,5]. According to the anatomo-pathology characteristics of cerebral defect, four forms of Syndrome of Dandy-Walker were described by Murray and al [6]

i. The Dandy-Walker deformation (DWM) which is characterized by a cystic dilation of the 4th ventricle in front of the choroid plexus , a total or partial vermis agenesis and an widening of posterior fossa with extra height of the tent of the cerebellum. These anomalies were noticed in the first observation.

ii. The Dandy-Walker variant (DWV) associates a cystic dilation of the 4th ventricle forward the choroid plexus and a hypoplasia of the posterior and inferior part of the vermis. It was the case of our second observation.

iii. Complex Dandy-Walker (DWC) or Dandy-Walker-Blake who is characterized by a choroid hernia in the outside of the 4th ventricle with vermis hypoplasia by compression.

iv. The mega cisterna magna (MCM) is an abnormally voluminous arachnoid pocket without association of vermis malformation.

The Dandy-Walker syndrome is frequently associated in variable degrees with intra and extra cranial deformations. Those deformations are caused by anomalies of the ectoderm and/or the mesoderm. The intra-cranial deformations consist essentially of corpus callosum agenesis. The extra-cranial deformations are dominated by a facial dysmorphia, skin defect of hands and cardiac and renal malformations [3,6].

Our first observation contained skin anomalies while the second observation contained multi-cystic loins. The syndrome of Dandy-Walker has heterogeneous etiology dominated by genetic factors (mailmen) and environmental agents effects [7]. The genetic etiopathogenesis is admitted without certainty and involves essentially the chromosomes 13 and 18. Chromosomes 9 and 17 are more rarely involved in the form of trisomies and of triploids. The environmental factors incriminated in the genesis of the syndrome of Dandy-Walker are dominated by infectious agents, in particular the virus of the rubella, the cytomegalovirus and the toxoplasmosis. Other environmental factors seem to play a role in the syndrome of Dandy-Walker among which alcohol consumption, maternal diabetes and the lack of riboflavin in food during pregnancy [7]. In our observations no risk factor was found except a consanguinity of 2nd degree reported for the second observation.

The prenatal diagnosis of the syndrome of Dandy-Walker is based on the conventional obstetric ultrasound completed by the foetal MRI. Of the 5th to the 7th weeks of pregnancy, the obstetric ultrasound looks for a characteristic sign consisting a big fluid space compared to the nuchal curvature . This space corresponds to the future 4th ventricle. The foetal hydrocephalus constitutes a no specific orientation sign of the syndrome of Dandy-Walker observed in 53 in 71% of the cases [3]. At our patients the prenatal diagnosis was late due to the lack of prenatal follow-up.

The clinical symptoms of the Dandy-Walker syndrome after the child birth are extremely variable according to the size of the cyst, the speed of appearance of the hydrocephalus and the type of the associated deformations. The clinical signs generally appear at the birth or from the first months of life. They associate signs of hydrocephalus, a motor slowdown, psychomotor delay and premature epilepsies. The infant mortality varies between 25 and 45% of the cases with an ascendancy for the children between one and 16 years old [4]. Later, signs of hydrocephalus settle down containing headaches, nausea, lethargy, oculomotor signs, ataxia, deafness of transmission, epileptic seizures, tonus disorders, a delay in walking and a decrease of the intelligence quotient observed in 50 in 70% [8]. The treatment of the Dandy-Walker syndrome is essentially surgical to decrease the intra-cerebral pressure exercised by the hydrocephalus on the intellectual structures. In utero, an endoscopic surgical operation turns out in certain cases useful between 24th and 32th weeks of gestation to create a ventriculo-amniotic shunt. This shunt will be replaced on birth by a ventriculo-peritoneal shunt.

The forecast of the Dandy-Walker syndrome is variable according to various parameters. The elements of bad forecast consist in the existence of associated deformations, an abnormal karyotype and a progressive and fast increase of the hydrocephalus [9,10]. The premature discovery in utero of the Dandy-Walker syndrome is very useful to assure an adequate follow-up of the pregnancy or envisage a possible medical interruption of the pregnancy. Such a decision must be multidisciplinary by taking into account associations of malformative anomaly and karyotype defect.

Conclusion

The Dandy Walker syndrome is a rare and grave deformation of the central nervous system. It is characterized by a cystic dilation of the posterior fossa and an agenesis of the cerebellar vermis. The clinical symptomatology is very polymorphic and it is correlated to the morphological anomalies. A prenatal diagnosis is made possible from the first quarter of the pregnancy due to the foetal ultrasound progress. The intra uterine surgical traitement is performed to decrease the intracerebral pressure. The forecast of this syndrome is variable according to several parameters; the most important is the coexistence with other deformations.

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