Anencephaly and rachischisis: about an observation

Authors: N . KACI MEDJAMIA , S .DJELTI , Y BENMOUMEN
Department of gynecology-obstetrics " NOUAR FADELA ", Oran, Algeria
Email:medjamia.kaci@yahoo.fr

Introduction

Neural tube defects (NTDs) are a group of congenital malformations caused by a neural tube closure defect during the 4th week of embryonic development. This defect can reach all levels of the neural plate from the cephalic end to the caudal end with a variability of extent, this defect can be responsible for abnormalities of the nervous tissue, meninges, and also bone and skin anomalies. These AFTNs can lead to two main forms: encephalic dysraphies (anencephaly, encephalocele, etc.) and spinal cord dysraphies (or spina bifida).
In continental Europe, the frequency is between 0.6 and 1.3/1000. From 1981 to 2000, the Paris Registry of Congenital Malformations reports a prevalence of anencephaly of 0.5/1000 births (live or not), identical to that of spina bifida. During this period, in this same registry, and retaining only live births, the prevalence of anencephalias increases from 0.6 to 0/10000, and from 4.4 to 0.8/10000 for spina bifida and this is due to antenatal diagnosis and medical terminations of pregnancy. These malformations are influenced by multi-factorial factors, both genetic and environmental (nutritional deficiency), although the prevalence decreases since the development of prevention by folic acid. Finally, there is a sex-ratio imbalance, especially in the case of anencephaly, which is three times more frequent in female fetuses.
The prognosis of AFTN is generally poor with a severe handicap, even lethal (in case of anencephaly), hence the interest in diagnosing them early and if possible from embryonic life. It is therefore important to diagnose them early and if possible from embryonic life. And above all to look for forms of good prognosis (meningocele without spinal cord involvement) as well as those of moderate severity such as for example the sacral myelomeningocele of little extent, and this management must be multidisciplinary including imaging, histoembryology and surgery (orthopedics, neurosurgery, pediatric surgery) .

goal

The aim of this work is to detect these malformations at an early stage and this is done thanks to antenatal screening which is essentially based on morphological ultrasound.

Materiel and methods

In our observation we report the case of a 34 year old woman, third gesture, two normal deliveries who presented for management of a delivery on a pregnancy of 32 SA has complete dilation on transverse presentation (neglected shoulder), to note that this pregnancy was not followed and no ultrasound was made during this pregnancy, a Caesarean section was performed urgently allowing the extraction of a nonliving child anencephalic female with an extended rachischésis.

Conclusion

Neural tube defects remain frequent anomalies in our context. They are responsible for significant mortality and morbidity. The prevention strategy for neural tube defects should be strengthened by insisting on pregnancy follow-up, Folic acid supplementation in pregnancies at high risk of recurrence of neural tube defects.

Figures

Figure 1

Figure 2

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