



## Modern Aspects of Accurate Diagnosis of Neural Tube Defects

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(Received 10 Dec 2021; accepted 24 Dec 2021)

### Dear Editor-in-Chief

Neural tube defects (NTDs) are one of the most common fetal malformations of the central nervous system (CNS) and are object of study in every pregnancy because they occur in the early stages of fetal development. Although some of them are compatible with life, they represent a significant social problem for the family and society. Malformations are due to improper closure of the neural tube and can occur sporadically or as a syndrome. Their etiology is multifactorial, related to demographic, exogenous and endogenous maternal factors.

We aimed to provide information on modern aspects of accurate diagnosis of NTD and to help in the prevention for future pregnancies.

In malformations of the CNS, significance is given to the age of the mother, as the frequency is in the form of a U-shaped curve, with the highest frequencies at a very young age - under 18 yr and over 38 yr (1). In addition to the age of the mother, interfamilial marriage, the season of conception, variations in amniotic fluid and the sex of the fetus are etiopathogenetic factors associated with malformations from the group of NTDs (2, 3).

Based on known genetic mechanisms or empirical studies, many of malformations of CNS, including NTDs, have an increased risk of recurrence in future pregnancies. Women who have

risk factors for NTDs and a history of fetus or live birth with a malformation undergo detailed sonographic examinations during a new pregnancy, which should reassure parents that the current fetus is not affected.

Detection of neural tube defects in the fetus is possible through prenatal biochemical screening, early amniocentesis, and genetic testing. For example, elevated maternal serum alpha-fetoprotein (MSAFP) levels are known to be an important indicator of fetal malformations, such as NTDs. Compared to biochemical testing of MSAFP performed on its own for screening, routine ultrasound (US) examination after the second trimester is more likely to detect NTD. US examination easily presents disorders in morphology, as the method allows the study of the internal and external anatomy of the fetus. In the general population, the two methods complement each other, but in the presence of factors that put the pregnant woman at risk group, the fetal scan should be performed by a specialist with more experience in sonography. Despite the proven effectiveness of ultrasound, it can create some dilemmas in counseling, as detected malformations may be questioned as normal variants or transient findings (4).

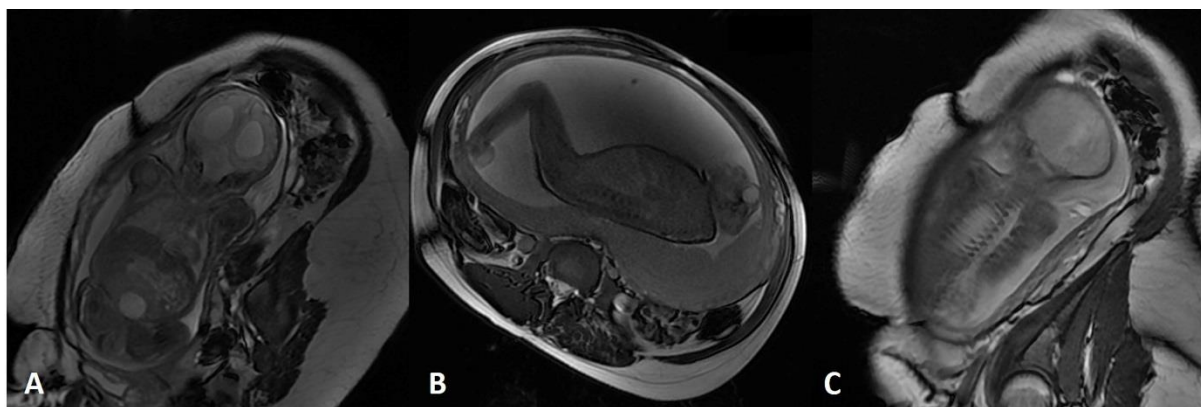
Many malformations of the CNS present a diagnostic challenge in ultrasound examinations dur-



ing the second trimester of pregnancy, so the use of new methods such as magnetic resonance imaging (MRI) for prenatal detection of structural changes is a justified goal (5). In cases where prenatal ultrasound findings are not absolutely certain, MRI examination is an important addition to the final diagnosis and assessment of the type of NTD. It provides more informative and definite data on both the presence of NTD and the presence of associated malformations, thus giving doctors and parents the opportunity to make an informed decision about the outcome of the pregnancy. MRI in pregnant women is used to detect malformations of CNS after the 18<sup>th</sup> week of gestation. Despite of some difficulties as the period of the pregnancy and movements of the

fetus during the scanning, MRI has several advantages over sonographic examination, which are used in medical practice. An important advantage of MRI is its higher resolution, which allows a better image of the CNS, both the gray and the developing structures of the white matter (6).

The figure presents some of the results of fetal MRI examination by our pilot project for Bulgaria, conducted by a multidisciplinary team at the Research Complex for **Translational Neuroscience** at the Medical University of Plovdiv (Fig. 1). We were able to confirm the results of the MRI examination after a fetopathological autopsy.



**Fig. 1:** Fetal brain MRI with data findings of NTDs: (A), Spina bifida cystica, Hydrocephalus (B), Anencephaly: (C), Spina bifida cystica

We can conclude that in cases where the sonographic diagnosis is suspicious for NTDs, MRI is a recommended method that provides medical professionals and parents with the necessary information to determine the outcome of pregnancy.

### Conflict of interest

The authors declare that there is not conflict of interest.

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