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ORIGINAL ARTICLE

FETAL MORPHOLOGICAL ULTRASOUND AND THE MAIN FINDINGS OF THE CENTRAL NERVOUS SYSTEM

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ABSTRACT

Introduction: Prenatal diagnosis of malformations is an important method of prevention and control of congenital anomalies. Central nervous system (CNS) malformations among these are the most common. Ultrasound during pregnancy plays a prominent and influential role in prenatal assessment of the central nervous system.

Objectives: to analyze the main findings of central nervous system alterations in fetal morphological ultrasound.

Methods: This is a cross-sectional, descriptive, retrospective and quantitative study, with secondary data collection. The sample consisted of 929 second-trimester morphological ultrasounds performed from January 2020 to January 2021, at Clínica Fértile.

Results: 929 reports were registered, of which 667 (71.8%) correspond to women under 35 years of age and 262 (28.2%) women over 35 years of age. Of these, 56 (6.0%) reports showed some type of morphological change and changes in the central nervous system were found in 15 reports with a prevalence of 1.61%. In women under 35 years, the prevalence of alterations in the central nervous system was 1.18%, the main alteration being hydrocephalus. in women over 35 years, the prevalence of alterations in the central nervous system was 0.43%, the main alteration being ventriculomegaly.

Conclusion: The prevalence of alterations in the central nervous system is 1.61%. In women under 35 years, the prevalence of alterations in the central nervous system was 1.18%, the main alteration being hydrocephalus. In women over 35 years, the prevalence of alterations in the central nervous system was 0.43%, the main alteration being ventriculomegaly.

KEYWORDS: CENTRAL NERVOUS SYSTEM, MALFORMATIONS, ULTRASONOGRAPHY

INTRODUCTION

Prenatal diagnosis of malformations is an important method of preventing and controlling congenital anomalies. Central nervous system (CNS) malformations among these are the most common. Neural tube defects are responsible for most congenital anomalies of the central nervous system (CNS) and result from the failure of the neural tube to close spontaneously between the 3rd and 4th weeks of embryonic development [1,2].

Congenital malformations of the central nervous system are related to alterations in the neural tube formation, including most neurosurgical management entities, dysraphism, and craniosynostosis; changes in neuronal proliferation; megalencephaly and microcephaly; neuronal migration disorders, lissencephaly, pachygyria, schizencephaly, agenesis of the corpus callosum, heterotopia and cortical dysplasia, spinal malformations and spinal dysraphism [3].

Globally, the incidence of congenital anomalies has been shown to vary from one geographic region to another, affecting about 3-7% of all newborns. Most causes of congenital anomalies are uncertain. However, evidence suggests that in about 25% of cases where causes are known, they appear to be multifactorial, involving a complex interaction between genetic and environmental factors. Likewise, morphogenesis errors that result in congenital malformations have been associated with some recognized genetic causes, including mutations in a single gene, chromosomal imbalances, and the action of teratogens. Folate deficiency has a recognized teratogenic effect, resulting in an increased risk of neural tube defects. Observational and interventional studies have also shown a protective effect of 50-70% in women who consume adequate amounts of folate in the neural tube[4].

The International Society of Ultrasound in Obstetrics and Gynecology (ISUOG) has published guidelines for the ultrasound study of the brain and spine in fetuses. The ISUOG guidelines are divided into two categories: basic CNS assessment; and neurosonographic assessment. The aim of this review was to describe, based on the ISUOG guidelines, how the ultrasound assessment of the fetal CNS should be performed. Ultrasound during pregnancy plays a prominent and influential role in prenatal assessment of the central nervous system. It provides an excellent window for viewing and evaluating the fetal central nervous system during the second trimester through the lateral ventricles and transthalamic view, effectively contributing to the diagnosis and treatment of congenital anomalies[5-7].

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Information on the prevalence and spectrum of malformations detected during prenatal care is crucial for genetic counseling and the design of population-based preventive programs[1].

The objective here is to analyze the main findings of central nervous system alterations on fetal morphological ultrasound.

METHODOLOGY

This is a cross-sectional, descriptive, retrospective and quantitative study, with secondary data collection. The research was carried out at the Fértile Clinic, which is private, located in Goiânia, Goiás.

The sample consisted of 929 second trimester morphological ultrasounds performed from January 2020 to January 2021, at Clínica Fértile. Ultrasounds with missing and/or inconclusive information were excluded.

Data were obtained from data contained in second trimester morphological ultrasound reports performed at Clínica Fértile, with the authorization of the institution.

The ultrasound report was considered a dependent variable. The independent variables were: maternal age (in years), gestational age (in weeks).

For the statistical analysis, an electronic spreadsheet was prepared in the program Microsoft Office Excel® 2010. The quantitative data were analyzed descriptively through the distribution of absolute and relative frequencies.

The present study was approved by the Research Ethics Committee (CEP) of Hospital e Maternidade Dona Iris, according to opinion number 5.002.479.

RESULTS

A total of 929 reports were recorded, of which 667 (71.8%) correspond to women under the age of 35 years and 262 (28.2%) to women aged 35 years or over. Of these, 56 (6.0%) reports showed some type of morphological alteration and anomalies in the central nervous system were found in 15 reports with a prevalence of 1.61%. In women under 35 years of age, the prevalence of central nervous system alterations was 1.18%, the main alteration being hydrocephalus. In women over 35 years old, the prevalence of central nervous system alterations was 0.43%, the main alteration being ventriculomegaly. With 80% of cases with multiple alterations.

Tables 1 and 2 and figures 1 and 2 illustrate the findings for patients aged < 35 and > 35 years, respectively.

Maternal Age	Gestational Age		Description of alterations
20	22	Multiple	Achondroplastic dwarfism / Dandy-Walker syndrome / Moderate hydrocephalus / Pyelectasis
20	23	Multiple	Cerebellar vermis agenesis / polyhydramnios / bilateral pleural effusion / amniocentesis indicated for karyotype
22	22	Multiple	Proboscis / nasal bone agenesis / choroid plexus cyst / omphalocele
24	23	Multiple	Dandy Walker disease / moderate hydrocephalus / 15mm posterior fossa cyst
25	23	Multiple	Neural tube defect / lumbosacral meningocele + mild hydrocephalus 15mm
27	33	Multiple	Cerebellar vermis agenesis / 14mm dilation of the cisterna magna and ventriculomegaly 14mm on the right and 11mm on the left
28	24	Multiple	Neural tube defect = lumbosacral spina bifida (5 vertebrae) + hydrocephalus + clubfoot
29	20	Multiple	Body stalk anomaly / second papyraceus conceptus
31	26	Multiple	Cerebral abnormality = posterior fossa cyst (28mm) with cerebellar vermis agenesis/mild hydrocephalus (Dandy Walker disease) - 17mm lateral ventricle
32	34	Single	13mm cerebral ventriculomegaly
33	27	Single	Neural tube defect/lumbosacral meningocele

Table 1 – Description of alterations in the central nervous system of morphological ultrasounds performed in the second trimester in women under 35 years old at Clínica Fértile, Goiânia, Goiás.

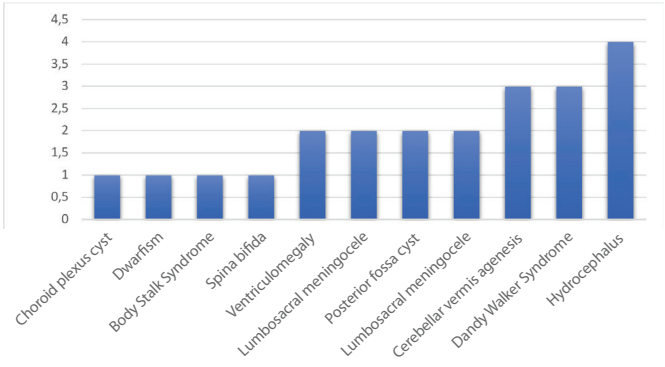

Figure 1 – Description of the main alterations in the central nervous system of morphological ultrasounds performed in the second trimester in women under 35 years old at Clínica Fértile, Goiânia, Goiás.

Maternal Age	Gestational Age		Description of alterations (Report)
35	23	Single	Choroid plexus cyst in regression (4mm)
39	24	Multiple	Neural tube defect (lumbosacral myelomeningocele 26mm / cranial deformity / ventriculomegaly 11mm / Chiari malformation type 2
41	23	Multiple	Holoprosencephaly / nasal bone absence / complex heart disease / pyelectasis / polyhydramnios / placental thickening
57	24	Multiple	15mm cisterna magna dilatation / cerebellar vermis agenesis / 11mm ventriculomegaly / mild hypoplasia of the left heart chambers / renal dysplasia / umbilical artery / fetal trisomy

Table 2 – Description of alterations in the central nervous system of morphological ultrasounds performed in the second trimester in women over 35 years old at Clínica Fértile, Goiânia, Goiás.

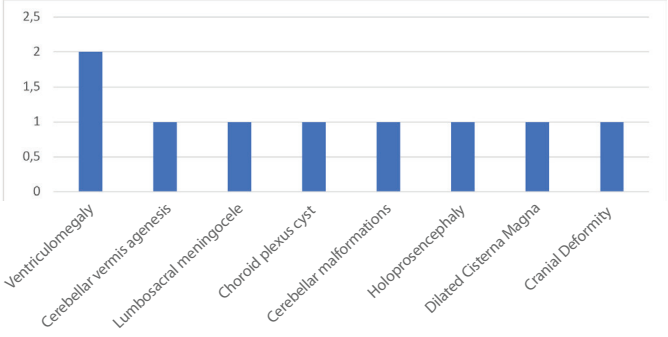

Figure 2 – Description of the main alterations of the central nervous system of morphological ultrasounds performed in the second semester in women over 35 years old at Clínica Fértile, Goiânia, Goiás.

DISCUSSION

Intracranial congenital malformations are anomalies of brain development caused by genetic and environmental influences. Advances in neuroimaging techniques and genetic research have led to a better understanding of the pathogenesis of many congenital malformations, adding insight into their clinical relevance and the intricate relationship between critical developmental periods, genetic predisposition, and environmental factors. When a malformation is discovered, there is a high probability of more malformations, which corroborates this study, which found 80% of cases of multiple alterations[8].

In this study, 56 (6.0%) reports showed some type of morphological alteration and anomalies in the central nervous system were found in 15 reports with a prevalence of 1.61%. In women under 35 years of age, the prevalence of central nervous system alterations was 1.18%, the main alteration being hydrocephalus. In women over 35 years old, the prevalence of central nervous system alterations was 0.43%, the main alteration being ventriculomegaly.

A retrospective study performed in a tertiary hospital in northern India from January 2007 to December 2013. Details of cases with prenatally detected CNS malformations were collected and related to fetal chromosome analysis and autopsy findings. Among 6,044 prenatal ultrasound exams performed; 768 (12.7%) had structural malformations and 243 (31.6%) had CNS malformations. Neural tube defects were responsible for 52.3% of CNS malformations and 16.5% of all malformations. The other main groups of CNS malformations detected prenatally were ventriculomegaly and midline anomalies[1] in agreement with the findings.

When evaluating 2,701 pregnant women in ultrasound exams, the sensitivity, specificity, positive predictive value and negative predictive value of the diagnosis of fetal CNS malformation were 85.7%, 100%, 100% and 99.9%, respectively[9].

In a retrospective cohort study, 47 patients were diagnosed with fetal CNS anomalies at a median gestational age of 31.1 weeks (range 24-38). The four most common anomalies found included intracranial cysts (19%), mild ventriculomegaly (15%), absence or dysgenesis of the corpus callosum (10%), and intracerebral hemorrhage (10%). Other CNS anomalies detected in this group of patients included hydrocephalus, Dandy walker malformation, dilated cisterna magna, microcephaly with lissencephaly, craniosynostosis, periventricular pseudocysts, global cerebral ischemia, cerebellar hypoplasia and subependymal nodule[10].

Knowledge of congenital malformations and their appearance in imaging sequences is essential to improve clinical outcomes and patients' quality of life[11].

Twins are about four times more likely to have congenital malformations than single children[12].

Each type of nervous system malformation is relatively uncommon, but collectively they constitute a large population. Treatment primarily consists of supportive therapies for developmental delays and epilepsy, but prenatal surgery for myelomeningocele offers a glimpse of future possibilities. The prognosis depends on several clinical factors, including examination findings, imaging features, and genetic findings. Treatment is best conducted in a multidisciplinary setting with neurology, neurosurgery, developmental pediatrics, and genetics working together as a comprehensive team[13].

Fetal structural abnormalities are found in up to 3% of all pregnancies and ultrasound screening has been an integral part of routine prenatal care for decades. Prenatal detection of fetal anomalies allows for optimal perinatal management, providing prospective parents with opportunities for additional imaging, genetic testing, and providing information about prognosis and management options[14].

It can be concluded that standardized structural ultrasound screening during pregnancy can early detect fetal

CNS malformations and has important clinical value in reducing the birth rate of malformed fetuses and in guiding obstetric treatment[15].

Congenital malformations are not uncommon and the central nervous system is the most commonly affected system. Health managers should emphasize primary prevention in the form of vaccination, nutrition and medication to reduce the avoidable participation of congenital malformations[15].

CONCLUSION

The prevalence of central nervous system alterations is 1.61%.

In women under 35 years of age, the prevalence of central nervous system alterations was 1.18%, the main alteration being hydrocephalus.

In women over 35 years old, the prevalence of central nervous system alterations was 0.43%, the main alteration being ventriculomegaly.

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