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Efficacy of Prenatal Ultrasound in Craniospinal Malformations According to Fetopathological and Postnatal Neonatological, Pathological Results

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ABSTRACT

Objective: Our objective is to examine the effectiveness of prenatal ultrasound diagnosis of craniospinal malformations compared to postnatal neonatological and pathological findings. *Methods:* Over a 7-year period, we preformed approximately 82.500 prenatal ultrasounds of 26.827 pregnancies. We detected 290 fetuses with 351 craniospinal malformations. *Results:* Craniospinal abnormalities were found as a part of multiplex malformations in 84/290 cases: in 47/84 cases (55.95%) there was complete concurrence between prenatal and postnatal results. In 15/290 fetuses the craniospinal malformation was associated with chromosomal abnormalities. In 9/15 (60%) of these fetuses, malformations were fully diagnosed with ultrasound. Isolated craniospinal malformations occurred in 191/290 cases, in 162/191 (84.82%) the results of prenatal ultrasonography and postnatal or post abortion examinations showed complete concurrence. In addition to the 290 fetuses with craniospinal malformations, there were an additional 17 who were thought by ultrasound to have a craniospinal malformation, which could not be documented after birth (false positives). *Conclusions:* Prenatal ultrasound accurately diagnosed 218/290 (75,17%) craniospinal abnormalities, and partially defined the abnormalities in 9.66%, failed to detect abnormalities in 15.17%, with an approximate 0.06% false detection rate.

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44/290 patients; craniospinal malformations; fetopathological and pathological examinations; prenatal ultrasound

Introduction

Craniospinal malformations are a relatively common but heterogeneous group among congenital anomalies. The most prevalent subgroup is neural tube defects with a prevalence of 0.1–0.8% (1–5). The anomalies of the brain and spine are usually of multifactorial origin but they can occur along with chromosomal aberrations and as part of multiplex malformations as well. In addition, intrauterine infections may also lead to the development of brain anomalies (6).

Abnormalities of the central nervous system (CNS) are usually severe with a poor prognosis. CNS anomalies are also the most frequent indication for termination of pregnancy (7). Therefore, early and precise detection of these anomalies is highly desired. Ultrasound is the main diagnostic tool that is used to effectively diagnose the malformations of the CNS (8,9).

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However, the efficacy of prenatal ultrasound tests vary greatly. Neural tube defects are usually detected with a high rate (25–94%), even at 12 weeks of gestation (3,10,11,12). In the second trimester, over 90% of the spina bifida cases are diagnosed with high certainty (3,13). On the other hand, the ultrasound is less effective in detecting anomalies such as microcephaly (6).

The aim of the current study was to examine the efficacy of prenatal ultrasound in craniospinal malformations according to fetopathological and postnatal neonatological/pathological result.

Materials and methods

Over a 7 year period, at our department there were 25.700 deliveries, 321 spontaneous midtrimester abortions, and 806 midtrimester terminations (TOP) because of fetal malformation. We performed approximately 82.500 prenatal ultrasounds.

We have processed the prenatal sonographic and postnatal clinical or fetopathologic details of 351 craniospinal abnormalities in 290 newborns and aborted fetuses at the 1st Department of Obstetrics and Gynaecology over a 7-year period. The patients were divided into three groups; Group 1: prenatal sonography and postnatal or fetopathological examinations yielded fully identical results. Group 2: the detected anomalies of the brain or spine had been partially discovered in prenatal investigations. Group 3: prenatal sonography had failed to detect the malformation identified in postnatal examinations or after termination.

Cases were considered partially recognized where the abnormality of the given organ was discovered during the ultrasound examination, but subsequent examinations (birth/abortion) showed that the final diagnosis was different from the presumed diagnosis. As an example among the brain anomalies, when ultrasound examinations showed lateral ventricle dilatation, but the postnatal studies demonstrated agenesis/dysgenesis of the corpus callosum.

The 1st Department of Obstetrics and Gynaecology operates as a prenatal diagnostic center, pregnant women are referred to our clinic from many parts of the country to be examined and consulted in progressive patient care. In the course of our work, besides other counseling situations, genetic counseling is often encountered with ultrasonic deviation detected in the intrauterine fetus. These deviations may include: 1 – Abnormalities detected in ultrasound examinations performed at other institutes, and confirmed by our genetic counseling's ultrasound examination, performed at our clinic. 2 – Abnormalities detected by ultrasonography during routine pregnancy care at our clinic, and these cases referred to the Genetic Counseling. 3 – Due to other reasons (eg. due to differences in biochemical parameters), cases referred to our Genetic Counseling, and the abnormalities were detected by our ultrasound examination. 4 – Differences detected during fetal echocardiography for other maternal and fetal reasons.

The malformations of the CNS that were associated with either chromosomal aberration or certain multiple disorders were investigated separately. According to EUROCAT guidelines (European Surveillance of Congenital Anomalies) we included major malformations and excluded minor anomalies (14). Newborns with two or more major anomalies were classified as multiple malformations. We also compared to what extent the various disorders were associated with polyhydramnios/oligohydramnios or small for gestational age. Gender distribution of the various malformations was also checked.

Sonographic investigations were performed in the Ultrasound Laboratory of the 1st Department of Obstetrics and Gynaecology. The investigations were conducted according to the professional protocol elaborated by the Hungarian Society of Obstetric and Gynaecological Ultrasonography.

Our work complies with the principles laid down in the Declaration of Helsinki. The work has been approved by the ethical committee of the institution.

In statistical procession calculating significance, we used the Chi-square ($\chi 2$) test. A disorder was regarded significant where $p < 0.05$ was established.

Results

In the seven-year period of investigation, a total of 290 fetuses or newborns were affected by a congenital malformation of the brain or spine, corresponding to a birth prevalence rate of 1.13%. At the time of delivery or abortion, the mean maternal age was 29.62 ± 5.85 years.

There were 114 deliveries (≥24 weeks of gestation) and 176 abortions; 7 spontaneous and 169 cases of termination (TOP). Table 1 contains the details of newborns and fetuses diagnosed with anomalies of the CNS. On average, deliveries took place around 33.68 ± 4.60 weeks with a mean birth weight of 2183.78 ± 892.92 g. More than half of the deliveries resulted in premature birth <37 weeks (72/114, 63.16%).

The mean gestational age at time of the abortions was 19.69 ± 2.19 weeks. Fetopathologic examination showed a mean fetal weight of 299.83 ± 143.06g (the data was missing in 18 cases).

In 191 fetuses, only the CNS was affected. Another 15 were associated with chromosome aberrations whereas other multiple malformations were detected in 84 fetuses. In 7 cases, both chromosome abnormality and multiple malformations were present.

On average, 2.1 ± 1.92 ultrasound examinations were performed during pregnancy; 2.5 ± 2.13 and 2.4 ± 1.89 prenatal tests had been done on fetuses affected by chromosome aberration or multiple malformation, respectively.

Prenatal ultrasonographic diagnosis and postnatal results completely coincided in 75.17%, i.e. 218/290 cases in fetuses with craniospinal disorders. In 28/290 cases (9.66%) discovery was partial, while in 44/290 patients (15.17%) the defects detected at autopsy or at postabortion/postnatal radiologic exam, but not found at prenatal ultrasound (false negative cases). (Table 2).

In 28/290 cases (9.66%) diagnosis was partial. In these (all with multiple lesions), there were 4 diagnoses that were not confirmed after delivery (false positive). Of the approximately 26.827 total patients with prenatal ultrasounds, 17 (0.06%) were diagnosed by ultrasound (12 ventriculomegaly, 3 agenesis/dysgenesis of corpus callosum, 2 Dandy Walker malformation) that were not confirmed post delivery.

In the 191 fetuses who only presented with malformations of the CNS, the results of prenatal ultrasound tests and postnatal or fetopathological examinations completely coincided in 162 cases (84.82%), showed partial agreement in 15 fetuses (7.85%) whereas prenatal investigation failed to reveal any disorders in 14 patients (7.33%).

Among the 84 patients with multiple malformation, the malformations were diagnosed completely in 47 cases (55.95%), partially in 11 fetuses (13.1%). However, no craniospinal anomaly was discovered in 26 fetuses (30.95%). Two systems were affected in 48 cases of multiple malformations and in 36 cases, the number of affected systems was ≥3. The associated malformations were as follows: malformations of the facial cranium (32 cases), cardiovascular malformations (27 cases) disorders of the extremities (24 cases), urogenital malformations (24 cases), disorders of the abdomen and abdominal wall (21 cases), other thoracic disorders (3 cases). In two patients, the associated disorder was diagnosed as fetal hydrops.

In 15 fetuses, malformations of the CNS were associated with a chromosomal abnormality. In 9/15 (60%) of these fetuses, malformations were fully diagnosed with ultrasound. In

Table 1. Data of cases of craniospinal malformations ($N = 351$).

	Total					Spontaneous Ab or TOP			Deliveries				
		Maternal age (years)					gestational weeks			gestational weeks		birth weight (grams)	
Type of anomalies	*cases*	average	SD	*min*	*max*	*cases*	Average	SD	*cases*	average	SD	average	SD
Ventriculomegaly/hydrocephaly	115	30,13	5,82	*16*	*43*	76	20,17	1,90	39	32,64	5,01	2083,77	913,43
Agenesis/dysgenesis of corpus callosum	26	27,81	5,09	*18*	*41*	5	20,80	1,79	21	35,24	4,04	2193,33	821,80
Spina bifida*	72	30,69	5,23	*20*	*44*	56	19,63	2,18	16	31,25	5,25	1705,00	866,54
Holoprosencephaly	26	30,23	5,81	*19*	*43*	12	19,25	2,45	14	31,93	4,60	1675,71	832,18
Dandy-Walker malformation/vermis hypoplasia	14	30,07	6,08	*20*	*38*	3	20,00	1,00	11	35,00	3,26	2331,82	750,56
Microcephaly	8	28,00	6,85	*18*	*35*	0			8	34,38	4,93	2335,00	862,02
Hydranencephaly	8	27,00	9,17	*18*	*43*	1	22,00		7	32,14	3,80	1894,29	719,97
Sacrococcygeal teratoma	8	29,50	6,99	23	43	2	19,00		6	34,33	5,32	2633,33	1022,38
Anencephaly/exencephaly	20	26,25	6,88	*16*	*39*	19	17,84	2,65	1	29,00		1170,00	
Encephalocele, meningocele	9	32,44	4,67	*27*	*41*	5	18,40	2,61	4	37,25	1,71	2967,50	1269,37
Arnold-Chiari malformation	2	22,00	11,31	*14*	*30*	1	18,00		1	35,00	0,00	1640,00	0,00
Other craniospinalis malformations	43	29,14	4,95	*20*	*42*	17	20,12	1,90	26	35,46	3,60	2701,67	696,57
Total	**351**	**29,62**	**5,85**	***14***	***44***	**197**	**19,69**	**2,19**	**154**	**33,68**	**4,60**	**2183,78**	**892,92**

Table 2. Accuracy of prenatal detection of craniospinal malformations ($N = 290$).

		I. totally discovered		II. partially discovered		III. not detected	
	cases	*N*	%	*n*	%	*n*	%
Isolated craniospinal abnormalities	191	162	84,82%	15	7,85%	14	7,33%
Associated with chromosome abnormalities	15	9	60,00%	2	13,33%	4	26,67%
Part of multiple malformation	84	47	55,95%	11	13,10%	26	30,95%
Total	**290**	**218**	**75,17%**	**28**	**9,66%**	**44**	**15,17%**

2 cases (13.3%), the diagnosis was only partial, while in the remaining 4 fetuses (26.7%), malformations were not diagnosed before birth or abortion. In seven cases, craniospinal anomalies were associated with Trisomy 21 (Down syndrome). Trisomy 13 (Patau syndrome) affected 5 fetuses, whereas Trisomy 18 (Edwards syndrome) appeared in 1 patient. Also, there was one case with the ring formation of the X chromosome.

Altogether, the 290 fetuses or newborns exhibited 351 malformations. The 351 malformations were divided into 12 groups (Table 3). The most common malformations were hydrocephalus and spina bifida with 115 and 72 cases. Spina bifida appeared without hydrocephalus in 53 fetuses, whereas in 19 cases the two abnormalities were associated. (Table 4). Agenesis/dysgenesis of the corpus callosum and holoprosencephaly presented in 26 cases each. Anencephaly/exencephaly appeared in 20, Dandy-Walker malformation in 14, while meningocele/encephalocele in 9 cases. Microcephaly, hydranencephaly and sacrococcyggeal teratoma were diagnosed in 8 fetuses each, whereas Arnold-Chiari malformation affected 2 patients. Other malformation of the CNS appeared in 43 cases (Table 5).

Table 3 shows the sensitivity of detection of different craniospinal malformations. 257/351 malformations (73.22%) were diagnosed with prenatal ultrasound examination. 37/351 were diagnosed partially (10.54%), while 57/351 were not recognized before birth or abortion (16.24%). We found high sensitivity in the anencephaly/exencephaly (95%), spina bifida (88.89%), hydranencephaly (87.5%) and hydrocephaly (80%) groups wile ultrasound sensitivity was lower in the microcephaly (25%), agenesis/dysgenesis of the corpus callosum (50%) and the other malformations of the CNS (46.51%) groups.

A combination of CNS malformations and small for gestational age was also often found (14.83%) (Table 6). Within this group, unusually high rates of small for gestational age was observed if microcephaly was the underlying malformation (37.5%).

The association of polyhydramnios and oligohydramnios with the craniospinal included polyhydramnios in 30% of the fetuses, while oligohydramnios was less prevalent, only presenting in 7.59% of the cases (Table 6). When compared to an average amount of amniotic fluid, we found a significant difference in the amount of amniotic fluid in most of the anomaly groups.

The boy/girl ratio was 1.13 in our cohort (Table 7). Dandy-Walker malformation, anencephaly/exencephaly and ventriculomegaly was significantly more common in boys ($p < 0,02$). On the other hand, significantly higher rates of microcephaly, saccrococcyggeal teratome and holoprosencephaly were observed in females ($p < 0,02$).

Comment

Prevalence

Based on our findings, the birth prevalence of craniospinal malformations was 11.28/1000. Our data far exceeded those in the EUROCAT study (2.41/1000), and those by Levi et al

Table 3. A. Accuracy of prenatal detection of craniospinal malformations ($N = 351$).

		I. totally discovered		II. partially discovered		III. not detected	
Type of anomalies	*cases*	*N*	%	*n*	%	*n*	%
Ventriculomegaly/ hydrocephaly	115	91	79,13%	6	5,22%	18	15,65%
Agenesis/dysgenesis of corpus callosum	26	13	50,00%	2	7,69%	11	42,31%
Spina bifida*	72	64	88,89%	5	6,94%	3	4,17%
Holoprosencephaly	26	19	73,08%	5	19,23%	2	7,69%
Dandy-Walker malformation/vermis hypoplasia	14	9	64,29%	2	14,29%	3	21,43%
Microcephaly	8	2	25,00%	1	12,50%	5	62,50%
Hydranencephaly	8	7	87,50%	0	0,00%	1	12,50%
Sacrococcygeal teratoma	8	6	75,00%	2	25,00%	0	0,00%
Anencephaly/ exencephaly	20	19	95,00%	0	0,00%	1	5,00%
Encephalocele, meningocele	9	6	66,67%	1	11,11%	2	22,22%
Arnold-Chiari malformation	2	0	0,00%	1	50,00%	1	50,00%
Other craniospinalis malformations	43	19	44,19%	12	27,91%	12	27,91%
Total	**351**	**255**	**72,65%**	**37**	**10,54%**	**59**	**16,81%**

*Associated and not associated with hydrocephaly cases in other Table

B. **Postnatal/post abortion diagnostics in cases of craniospinal malformations (N = 351)**

	Total	**Spontaneous Ab or TOP**			**Deliveries**		
		Diagnosis			Diagnosis		
Type of anomalies	*cases*	*cases*	Fetopsies	Radiology (X-ray US / MRI)	*cases*	Autopsies	Radiology (X-ray / US / MRI)
Ventriculomegaly/ hydrocephaly	115	76	76	4	39	12	31*
Agenesis/dysgenesis of corpus callosum	26	5	5		21	1	20
Spina bifida	72	56	56	2	16	8	8
Holoprosencephaly	26	12	12		14	8	8**
Dandy-Walker malformation/vermis hypoplasia	14	3	3		11	2	9
Microcephaly	8	0			8	1	7
Hydranencephaly	8	1	1		7	6	1
Sacrococcygeal teratoma	8	2	2		6	3	3
Anencephaly/ exencephaly	20	19	19		1	1	0
Encephalocele, meningocele	9	5	5	1	4	3	1
Arnold-Chiari malformation	2	1	1		1	0	1
Other craniospinalis malformations	43	17	17	2	26	7	19
Total	**351**	**197**			**154**		

*In 4 cases autopsy was performed, but the main diagnosis was radiological.
**In 2 cases autopsy was performed, but the main diagnosis was radiological.

Table 4. Spina bifida and ventriculomegaly/hydrocephaly.

		I. totally discovered		II. partially discovered		III. not detected	
Type of anomalies	*cases*	N	%	n	%	n	%
Hydrocephaly without spina bifida	96	73	76%	6	6%	17	18%
Spina bifida without hydrocephaly	53	46	87%	5	9%	2	4%
Spina bifida with hydrocephaly	19	18	95%	0	0%	1	5%

Table 5. Other craniospinal malformations.

Brain cysts	12
arachnoideal cyst	*3*
subependimal cyst	*2*
fossa posterior cyst	*2*
cerebellar cyst	*1*
intrehemispherial cyst	*1*
nucleus caudatus cyst	*1*
other brain cyst	*2*
Megacysterna magna	4
Dolicocephaly	2
Schizencephaly	2
Porenchephaly	1
Tumor cerebri	2
Hamartoma of galea aponeurotica	1
Hemocephaly	1
Aneurysm of vena Galeni	2
Craniorachischisis	1
Atrophy of optical nerve	1
Frontotemporal dilatation	1
Frontoparietal polymicrogyria	1
Agenesia of olfactoric lobe	1
Agenesia of olfactoric nerve	1
Temporal lobes are not divided	1
Agenesia occipital lobe	1
Cerebellar agenesia/hypoplasia	2
Absent frontal part of falx cerebri	1
Vertebral malformation	2
Extreme lordosis/scholiosis	3
Total	**43**

Table 6. Small for gestational age, oligo- and polyhydramnios were observed in fetuses with craniospinal malformations.

		Small for gestational age		Polyhydramnios			Oligohydramnios		
Type of anomalies	*cases*	n	%	n	%	p*	n	%	p*
Ventriculomegaly/ hydrocephaly	115	7	6,09%	37	32,17%	$p < 0.02$	10	8,70%	$p < 0.02$
Agenesis/dysgenesis of corpus callosum	26	6	23,08%	5	19,23%	$p < 0.02$	4	15,38%	$p < 0.02$
Spina bifida	72	11	15,28%	24	33,33%	$p < 0.02$	2	2,78%	$p < 0.02$
Holoprosencephaly	26	7	26,92%	6	23,08%	$p < 0.02$	3	11,54%	$p < 0.02$
Dandy-Walker malformation/vermis hypoplasia	14	3	21,43%	5	35,71%	NS	2	14,29%	NS
Microcephaly	8	3	37,50%	1	12,50%	NS	3	37,50%	NS
Hydranencephaly	8	1	12,50%	1	12,50%	$p < 0.05$	0	0,00%	$p < 0.02$
Sacrococcygeal teratoma	8	0	0,00%	2	25,00%	NS	0	0,00%	$p < 0.02$
Anencephaly/ exencephaly	20	1	5,00%	8	40,00%	NS	0	0,00%	$p < 0.02$
Encephalocele, meningocele	9	2	22,22%	2	22,22%	NS	0	0,00%	$p < 0.02$
Arnold-Chiari malformation	2	0	0,00%	0	0,00%	$p < 0.02$	0	0,00%	NS
Other craniospinalis malformations	43	11	25,58%	9	20,93%	$p < 0.02$	5	11,63%	$p < 0.02$
Total	**351**	**52**	**14,81%**	**100**	**28,49%**	$p < 0.02$	**29**	**8,26%**	$p < 0.02$

p* – in relation to the occurrence of average amount of amniotic.

Table 7. Gender distribution of craniospinal malformations.

Type of anomalies	*cases*	male	female	male/female	p
Ventriculomegaly/hydrocephaly	115	65	48	1,35	$p < 0.02$
Agenesis/dysgenesis of corpus callosum	26	10	16	0,63	NS
Spina bifida*	72	40	32	1,25	NS
Holoprosencephaly	26	8	17	0,47	$p < 0.02$
Dandy-Walker malformation/vermis hypoplasia	14	11	3	3,67	$p < 0.02$
Microcephaly	8	2	6	0,33	$p < 0.02$
Hydranencephaly	8	3	5	0,60	NS
Sacrococcygeal teratoma	8	2	6	0,33	$p < 0.02$
Anencephaly/exencephaly	20	12	6	2,00	$p < 0.02$
Encephalocele, meningocele	9	5	4	1,25	NS
Arnold-Chiari malformation	2	2	0		
Other craniospinalis malformations	43	23	19	1,21	NS
Total	**351**	**183**	**162**	**1,13**	NS

*In 6 cases the sex was unknown.

(3.5/1000) and Fadda et al (2.24/1000) (4,5,13). We found the birth prevalence of neural tube defect to be 0.039%.

The higher birth prevalence at our Clinic may be attributable to our clinic being a referral hospital.

Sensitivity

In our study, out of 290 newborns and fetuses with CNS system malformations, 218 were identified with prenatal ultrasound examination (75.17%), 28 were partially diagnosed (9.66%), while 44 were not detected before birth or abortion (15.17%). Our findings are comparable with the data of Levi et al (78.95%) (5). In a study by Fadda et al, they only counted those cases as prenatally diagnosed, where the malformation was identified before 24 weeks of gestation and found the sensitivity of the ultrasound to be 81.05% in their cohort (4). In our study, less, only 58.48% of the brain and spine anomalies were detected before 24 weeks. However, our data is in close correlation with those of Saltvendt (61.36%) (15).

According to our findings, 43.61% of the malformations were identified before 23 weeks of gestation. Our data shows a lower detection rate before 23 weeks than those of Levi et al (52%) and a much lower sensitivity than those of VanDorsten et al (88.24%) (4,16).

Comparing the individual disorders, our results showed partial agreement with the findings in the literature. We found a high ultrasound sensitivity in the anencephaly/exencephaly (95%), spina bifida (88.89%), hydranencephaly (87.5%), ventriculomegaly (80%), and sacrococcygealis teratome (75%) groups. In the EUROCAT study, the sensitivity of ultrasound in the detection of anencephaly was 96%, comparable with our findings (95%). In the same study, 68% of spina bifida cases were identified prenatally, which was lower than the 88.89% sensitivity in our data (13). The findings of Garne et al were in close correlation with ours: they were also able to detect hydrocephaly in 80% of the cases (17).

We found the prenatal ultrasound sensitivity to be relatively low in the agenesis of the corpus callosum (50%), other CNS malformations (46.51%), microcephaly (25%) and Arnold-Chiari malformation groups.

Chromosome abnormalities

Down syndrome was present in 7 out of 15 cases when CNS malformation appeared in association with a chromosomal abnormality. In all 7 of those cases, the craniospinal anomaly was

ventriculomegaly/hydrocephaly. Out of 115 ventriculomegaly cases, 10 fetuses had an abnormal karyotype (8.7%). This data showed correlation with those of Garne et al (9.2%) (17). On the other hand, the findings of D'Addario et al exceeded ours: they found chromosomal anomaly in 15% of all hydrocephaly cases (18).

Multiple malformation

In our study, 31.37% of the fetuses with brain or spine defects had multiple malformation (91/290 fetuses; 84 euploid, 7 aneuploid). In those cases, when the baby was born after 24 weeks of gestation, we found a higher rate of multiple malformations, 37.72%, while in case of abortion, the rate was lower, 23.3%.

Barros et al examined newborns with craniospinal malformations and found a higher, 57.5% association rate with other malformations. They also found that the most frequently associated malformations were those of the facial cranium (73.9%) (19). We also found the malformations of the facial cranium to be to most frequently associated. Garne et al found that in fetuses with hydrocephaly, other organs were affected in 44% of the cases, a higher rate than in our investigation (31.3%) (17).

Based on our findings, 84 euploid fetuses had multiple malformations (28.97%). Isaksen et al found a much lower association rate of 14.29% (20).

In our investigation, the detection rate was much lower in euploid fetuses with multiple malformations (47/84; 55.95%) than in fetuses with only craniospinal abnormalities (162/191; 84.82%).

We believe that major malformations in other organ systems interfered with the lower detectability of mild CNS malformations (ventriculomegaly in 9, other CNS malformation in 7 cases), because because the ultrasonographer's attention was focused on the other abnormalities (21).

Conclusions

Despite its limitations, the ultrasound examination is still the most common and most important diagnostic tool in the prenatal period, which can be used with great efficiency in the prenatal screening tests, as a non-invasive, mobile and inexpensive method of analysis. However, the sensitivity of the test is basically determined by the experience level of the examiner, so it is important that the tests are carried out at all times in accordance with the rules of the professional college.

Prenatal ultrasound was able to detect three quarter of craniospinal malformations before birth or abortion. Our results have confirmed that ultrasound tests play an important role in diagnosing CNS malformations but they do not always allow the detection of all brain and spine abnormalities. It has been concluded that anencephaly/exencephaly, spina bifida, hydranencephaly, ventriculomegaly, and sacrococcygealis teratome can be detected with great certainty in the fetus prenatally. In contrast, other CNS malformations, microcephaly and Arnold-Chiari malformation are found at a low rate. Being aware of the above is important for experts performing ultrasound tests, health professionals in providing genetic counselling and prenatal care and, also, neonatologists and paediatricians seeing newborns. During prenatal care, the expectant mother should be given adequate information about the efficacy of the examinations. If a malformation is detected postnatally, the couple should also be informed how reliably the specific malformation is detectable by prenatal sonography. It is

recommended to use alternative test method (eg MRI) for certain disorders (eg agenesis / dysgenesis of corpus callosum). In postnatal and post abortion diagnostics the use of combined modalities (US and/or MRI and/or X-ray and/or pathology) is best utilized for confirmation.

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Disclosure of potential conflicts of interest

No potential conflicts of interest were disclosed.

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