ASSESSMENT OF THE ACCURACY IN PRENATAL DIAGNOSIS OF CONGENITAL MALFORMATIONS. ANALYSIS OF 101 QUESTIONNAIRES FILLED IN BY PARENTS OF NEONATES HOSPITALIZED IN THE DEPARTMENT OF CONGENITAL MALFORMATIONS POLISH MOTHER'S MEMORIAL RESEARCH INSTITUTE

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Abstract

Introduction:

Prenatal diagnosis is an integral part of modern perinatal care. In the article results of questionnaires pertaining to the prenatal process of diagnosis are presented. Parents whose children were afflicted with congenital malformations of all types responded to enquiry

Materials and methods:

Between March 2014 and March 2015 150 of 355 infants were hospitalized in the Department of Pediatric Intensive Care and Congenital Malformations in Łódź, and 150 had congenital malformations.

Results:

101 parents of 150 children (67,3%) have given the feedback. Anomalies were such as: of the digestive system (37%), CHD (25%), OUN (14 %), genito-urinary (13%), skeletal system (9%) and respiratory system (2%). In 65 children of 101 the defects were detected prenatally. The obstetric US exam was the most frequently pointed out as performed (more than 1200). The biochemical markers and genetic tests in were performed in 34 pregnancies. The high percentage of ability to detect malformation was reported in the group of fetal echo examinations.

Conclusions:

1. Prenatal ultrasound exams were the least effective method of making appropriate prenatal diagnosis of congenital malformation.

2. Fetal echocardiography had a high level of sensitivity and specificity in detecting congenital malformations.

3. Prenatal cardiologists proved to be the most effective in detecting congenital malformations 89,3 % of detected abnormalities.

4. Biochemical exams had a positive result in only one case of Down Syndrome.

Key words: *prenatal diagnosis, congenital malformation, accuracy of diagnosis*

INTRODUCTION

Prenatal diagnosis is one of the most important milestones in contemporary perinatal care[1,2]. In developed countries the birthrate is lowering and women wait more and more longer to get pregnant. These changes require the effective tools of prenatal management.

The meaning of prenatal diagnosis in today's medicine should be underlined because congenital anomalies constitute one of the most serious problems. They occur in 5% of live-born neonates and in 20% of still-born newborns[1,3,4,5,6,7].

Congenital heart defects are still difficult to detect during routinely performed US examinations in obstetric practice. Assessment of the heart's anatomy and function is one of the most complicated

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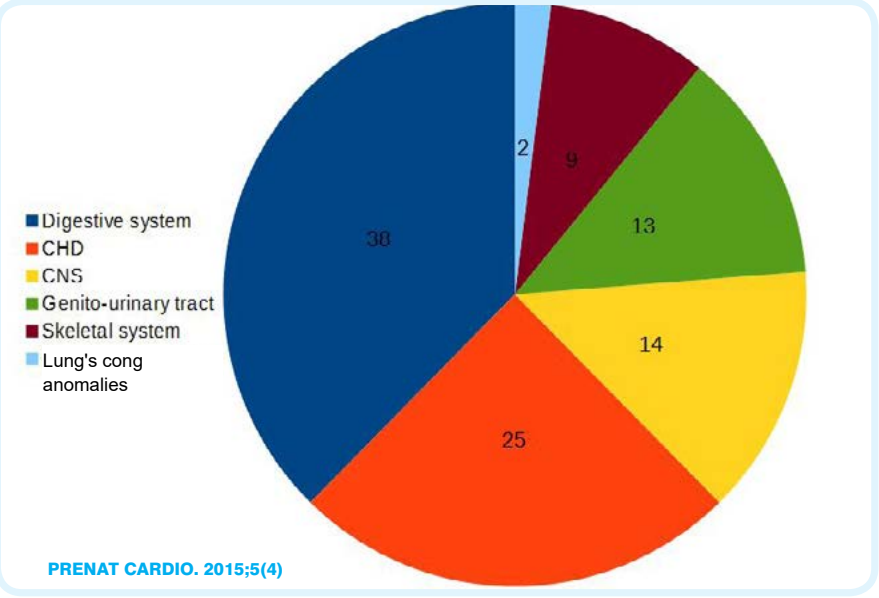

Fig. 1: Percentage of congenital malformations in questionnaires.

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Weeks of GA	Number of gravidas
12 hbd	1
13 hbd	2
16 hbd	2
18 hbd	5
20 hbd	7
22 hbd	10
24 hbd	14
27 hbd	2
28 hbd	7
30 hbd	11
33 hbd	4
Total	65

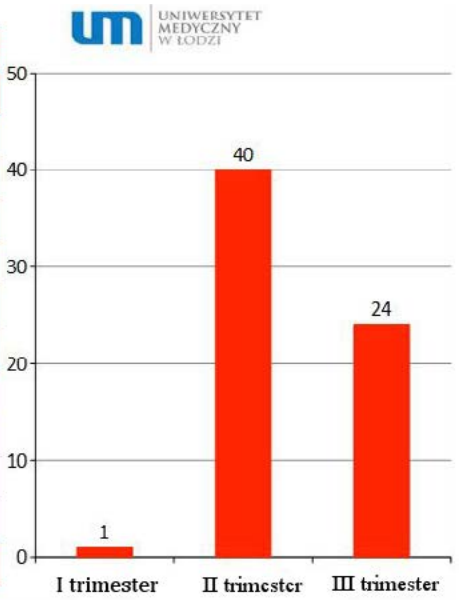
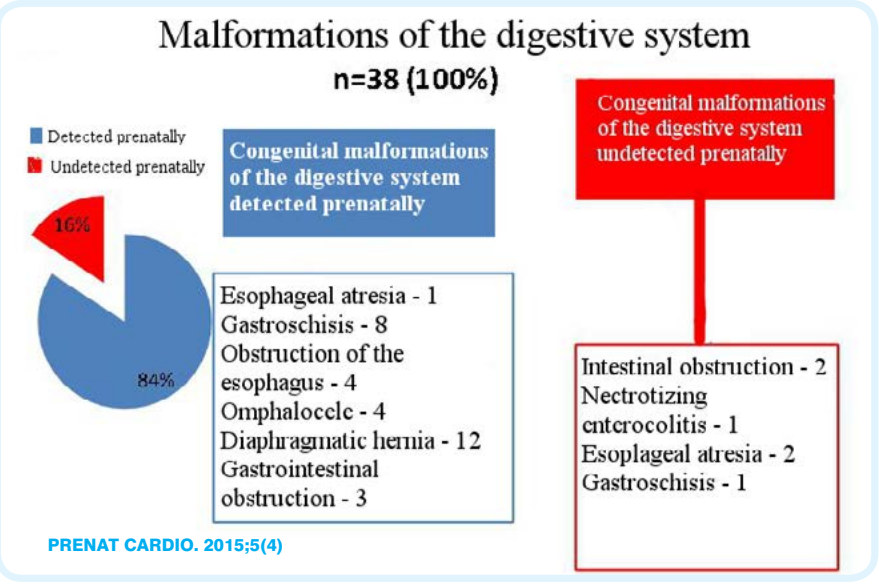


Fig.2: Time-of-prenatal-diagnosis-of-the-malformation



elements of this examination, but can contribute to the detection of CHD in prenatal life with a high level of sensitivity and specificity[8,9,10].

Prenatal diagnosis of XXI century is not just medical news or a subject to deliberate, but routine in the management of each pregnancy. It is necessary to perform prenatal tests accurately and professionally.

This article referring to effectiveness of prenatal diagnosis is written on a basis of 101 questionnaires filled in by the parent of a newborn hospitalized in the Department of Neonatal Intensive Care and Congenital Malformations, Polish Mother's Memorial Research Institute in Łódź.

MATERIALS AND METHODS

One hundred and one questionnaires were harvested among the parents between March 2014 and March 2015. Parents of all neonates afflicted with congenital anomaly were asked to fill in the form. During these 12 months 355 neonates were hospitalized in the Department, 150 of them suffered from congenital malformations.

RESULTS

101 parents of 150 newborns (67,3 %) gave their feedback. The most frequent defects were: anomalies of the digestive system 37 %, malformations of the cardiovascular system (25 %), central nervous system malformations (14 %), genito-urinary tract anomalies (13 %), skeletal anomalies (9 %) and lungs malformations (2 %) (Fig.1, Tab.1).

Moreover, there was one case of coexisting congenital heart disease and Down's Syndrome, one case of Treacher-Collins Syndrome and one case of Pierre-Robin Syndrome.

In 65 newborns out of 101 the defects were detected prenatally. In 40 of them the malformations were detected in the second trimester (at mean gestational age of 24 weeks according to last menstrual period). Twenty four out of 65 fetuses were diagnosed in the third trimester. In one case the diagnosis was made in the first trimester (Fig. 2).

Fig. 3: Digestive-system-defects

In 16% (6 cases) of neonates with digestive system anomalies (38 % of all malformations) the defect was missed prenatally (Fig.3). The percentage of undetected CHD was 36% (Fig.4). Four cases (29%) were missed prenatally in referrence to central nervous system malformations (Fig.5). Lung malformations were detected prenatally in both cases (n=2) (Fig.6). The highest percentage of overlooked defects concerned the skeletal system (Fig.7) and genito-urinary tract abnormalities (Fig.8).

The obsteric US exam was the most frequently pointed out as performed in all gravidas. More than 1200 examinations were reported in questionnaires.

Fetal echocardiographic examination was performed in 66 of the respondents (13 fetuses had a CHD among this group).

Biochemical markers and genetic tests were reported by 34 respondents. PAPP-A test was the most frequently performed in pregnant women. Amniocentesis was done in 5 gravidas and cordocentesis (PUBS) was performed in one gravida (Fig. 9a). There was one positive result of biochemical and genetic test (which detection of DS) among 34 performed (Fig. 9b).

The high percentage of ability to detect malformation was reported in the group of fetal echo examinations. These exams helped to detect 71,2 % of anomalies in the group of 66 respondents who had prenatal fetal echo exam.

The routine US scan turned out to be the least helpful in detection of congenital malformations. The percentage of observed anomalies was only 57,1 % (Fig. 9c).

Prenatal cardiologists and obstetricians certified by the Polish Ultrasound Society detected 32 and 10 anomalies respectively. These 42 defects constitute 89,3 % of all anomalies described postnatally in newborns (Fig. 10).

The analysis of age of mothers who had undergone biochemical /genetic tests uncovered 25 anomalies detected

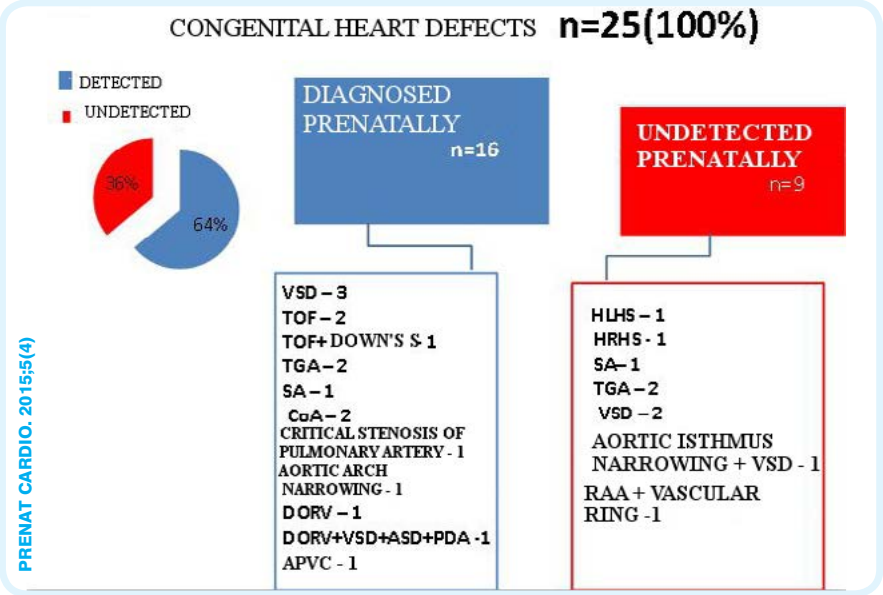


Fig. 4: CHD

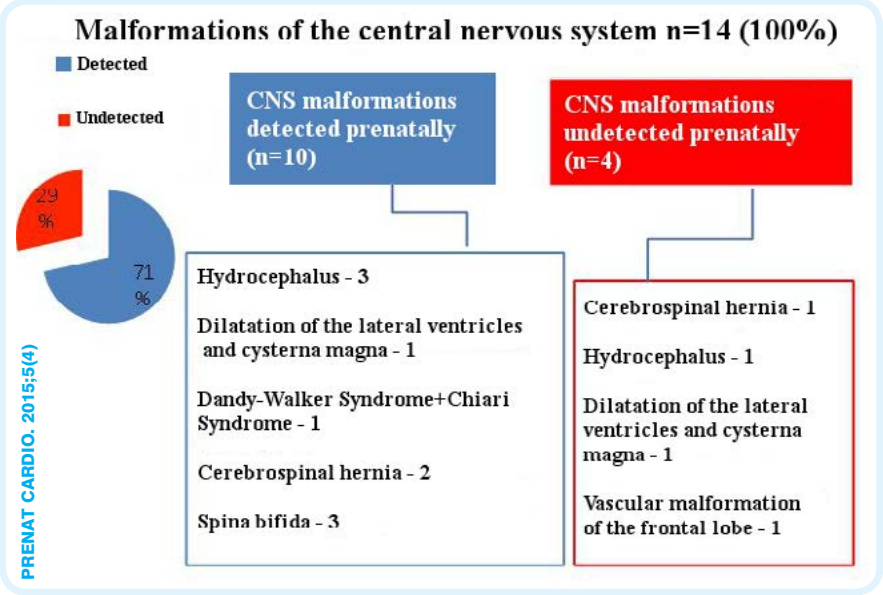


Fig. 5: Central nervous system malformations

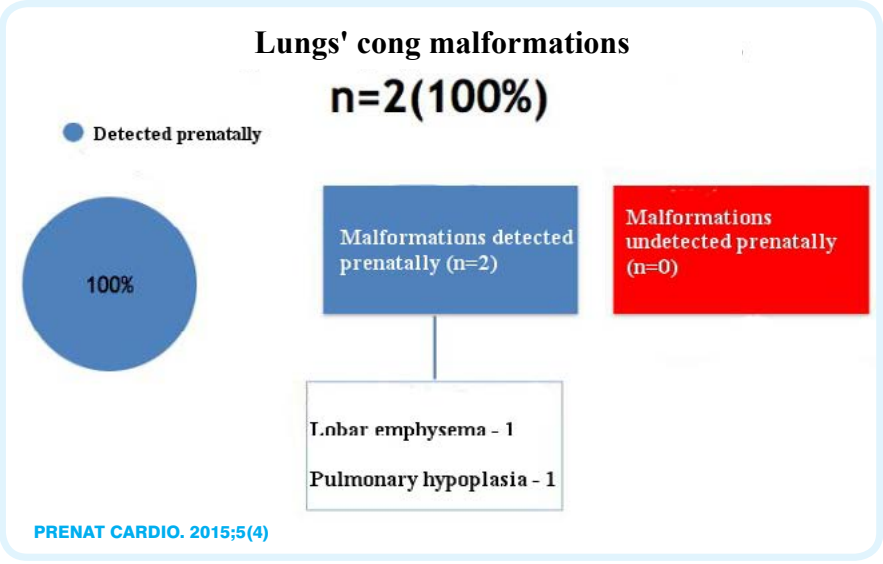


Fig. 6: Respiratory system malformations

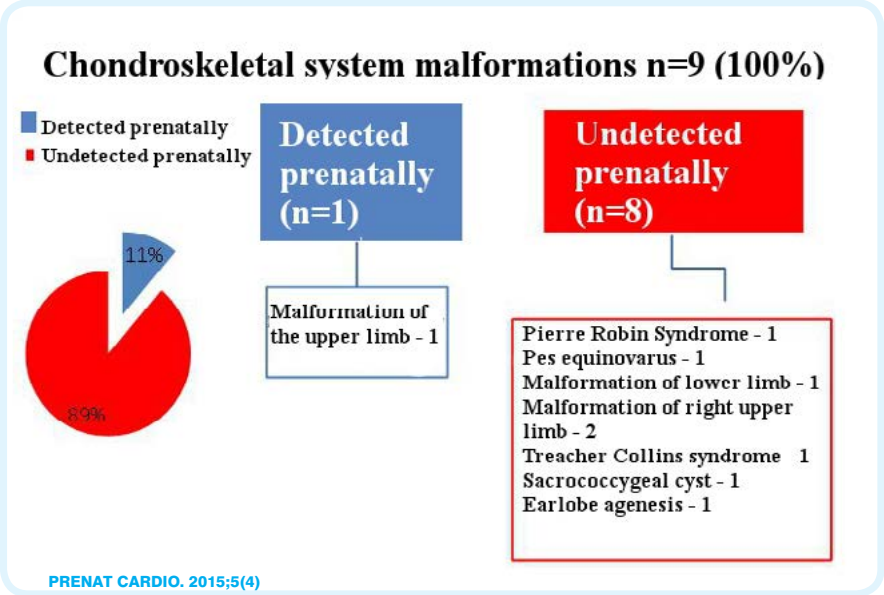


Fig. 7: Skeletal system malformations

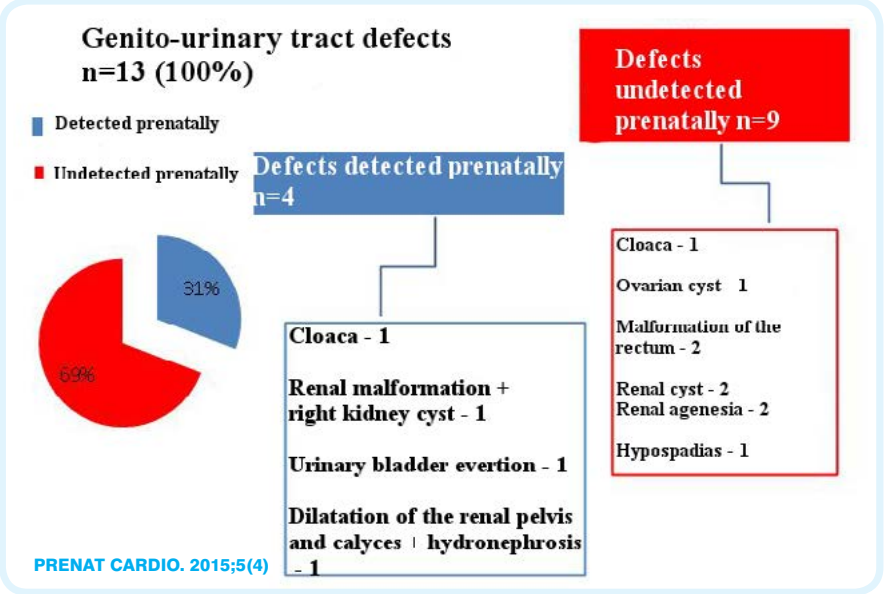


Fig. 8: Genito urinary tract defects

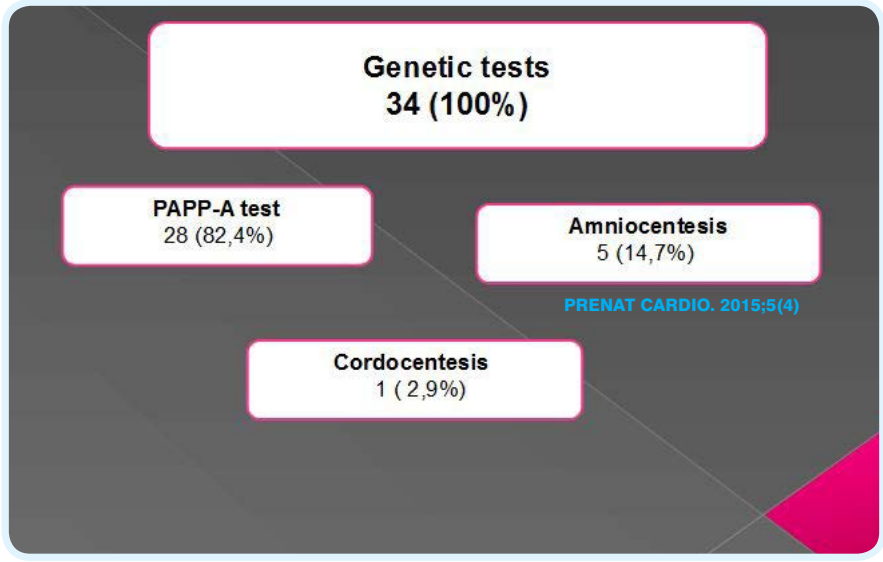


Fig. 9a: Biochemical and genetic tests performed in mothers who responded to questionnaire

and overlooked (73,5 %) occurring in fetuses of gravidas under 35 (Fig. 11).

DISCUSSION

The constant and significant progress in methods of prenatal screening and diagnosis is seen. These tests are becoming a standard in care of pregnant women[8,9,10].

However, we should ask the question: has everything been done? Can we without any doubt admire the present status quo of prenatal diagnosis?

In this report 150 parents were involved in the questionnaire and 101 (67,3%) responded. This percentage of feedback is believed to be high and satisfactory bearing in mind that parents are suffering from psychological trauma, they try to deny the illness of their child and sometimes acuse the neonatologist or obstetrician caring for their baby of the overlooked malformation.

Regarding to the most accessible exam - the basic US scan in this cohort, 20 examinations were needed to detect one anomaly. It is worth noting that in 518 examinations out of 1207 the anomaly became undetected (one patient underwent 50 US exams suggesting "normal" growing fetus).

In the group of respondents in which the malformation was undetected, one patient underwent on average 12,3 exams, in the group of detected anomalies - 11,7. This result is rather unoptimistic and indicates the necessity of constant education and training in US scanning and further searching for the perfect method in prenatal malformation screening.

The percentage of detection of congenital malformations in this report was 57,1 % pertaining to basic US exam and 71,2 % regarding fetal echocardiographic exam. The small effectiveness of the routine US exam (50,7 %) and its poor significance as a prenatal CHD screening method was noted by R. Sharony et al. and scientist from the USA who evaluated the problem of common access to prenatal screening[11,12,13], and also authors from India recalling the insufficient level of awareness in gravidas[14].

The analysis of questionnaires points out that biochemical tests performed in 34 patients allowed detection of one case of DS. In the group of prenatally detected malformations 81,5% of results were normal. Moreover, in all malformations undetected during pregnancy and checked by biochemical markers their results were normal, not even suspicious.

The aim of performing biochemical and genetic testing should be reminded. These tests are indicated to show increased risk of genetic abnormality. Isolated CHD are not detected by these methods. Encouraging results are provided by fetal echocardiography. Fourty seven anomalies were detected in 66 gravidas (it takes 1,05 exams to detect one anomaly). These ratios pertaining to biochemical tests and basic US exams were 1,2:1 and 20:1 respectively.

What is more, fetal echo exam thanks to its high specificity and sensitivity was the most effective method in prenatal CHD screening both in our study as well as in the literature[15,16,17,18].

The economic aspect should be also taken into consideration. The mean cost of the basic US in private practice is about 100 PLN, fetal echo costs 350 PLN. The cost of PAPP-A test ranges from 250 PLN. Genetic screening from mother's blood starts from 1500 PLN up to 2500 PLN. Therefore one anomaly detected by means of routine US costs 2000 PLN, by genetic methods from 250 up to 2500 PLN, and by fetal echocardiography - 350 PLN. The cost of transport of the neonate whose life is threatened should be added. Transport in utero is significantly less dangerous and much less expensive comparing with postnatal detection and ambulance transfer to tertiary center[19].

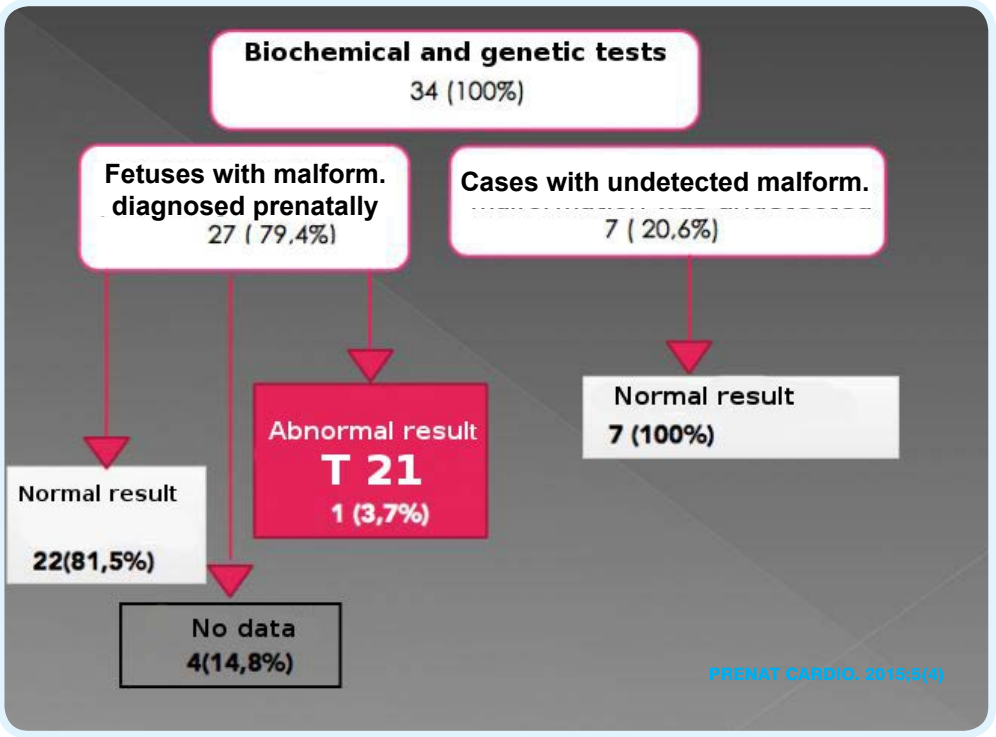


Fig. 9b: Prenatal sceening in presensted material to questionnaire in 34 fetuses/neonates with malformations

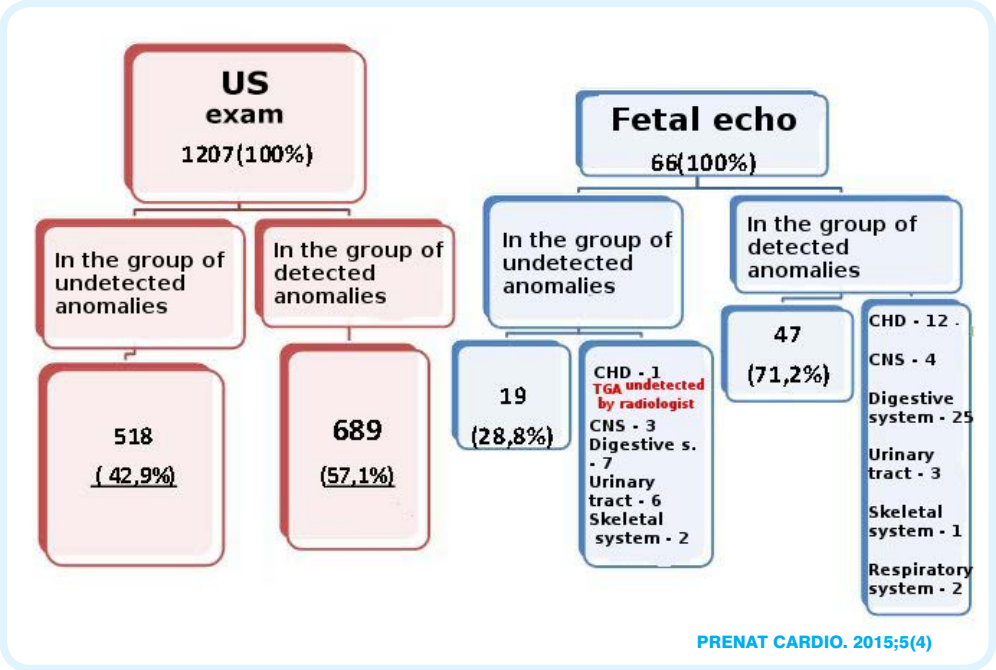


Fig. 9c: Prenatal us and echocardiographic examinations

Similar encouraging results regarding congenital malformation detection rates (including heart defects) were reported by R. Chaoui - 84,2% of true positive results in 2008[20,21].

It is necessary to underline that some of the authors recommend two fetal echo examinations performed at 14 and 18-22 weeks of gestational age[22,23,24,25,26,27]. This management may contribute in increasing the diagnosis rate of malformations[28].

What needs to be done in order to reach a high detection rate of fetal malformations? Focusing on the job of prenatal cardiologists, exceptional at detecting the most common in humans anomalies such as heart defects (89,3% of detected defects) it is believed that level of traineeship and experience plays a key role in proper prenatal diagnosis. Obstetricians certified by the Polish Ultrasound Society were also very successful in detecting the malformations.

These results confirm findings of Chaoui who noted that obstetricians after the basic course of US detect about 30% of anomalies, after special traineeship - up to 50-60% and in tertiary care centers, where specialists are employed, this ratio reaches 90%[29].

What is more, the majority of anomalies (73,5%) detected and undetected in fetal life were found in children of mothers under 35. This fact should be stressed because the National Health Fund in Poland refunds these exams only in women after 35. Apparently, the clerks at the NHF based their calculations on statistics and knowledge about congenital malformations dated back to the eighties and nineties of last century. The analysis of questionnaires clearly shows that the age of the gravida has less significance in the development of congenital malformations than we expected in the past decades.

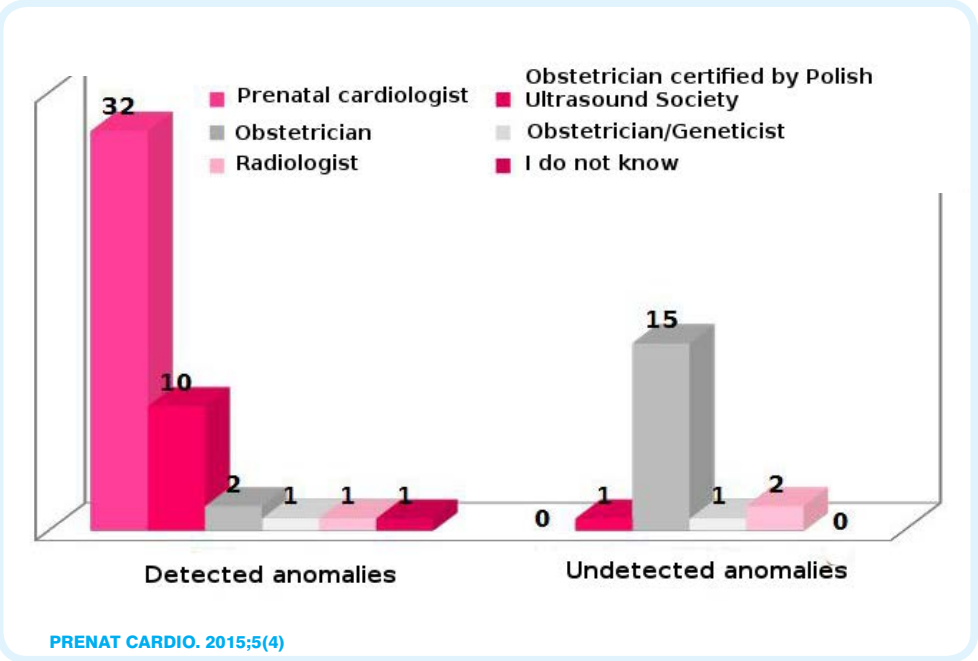


Fig..10: Fetalechocardiographic examination performed by specialist

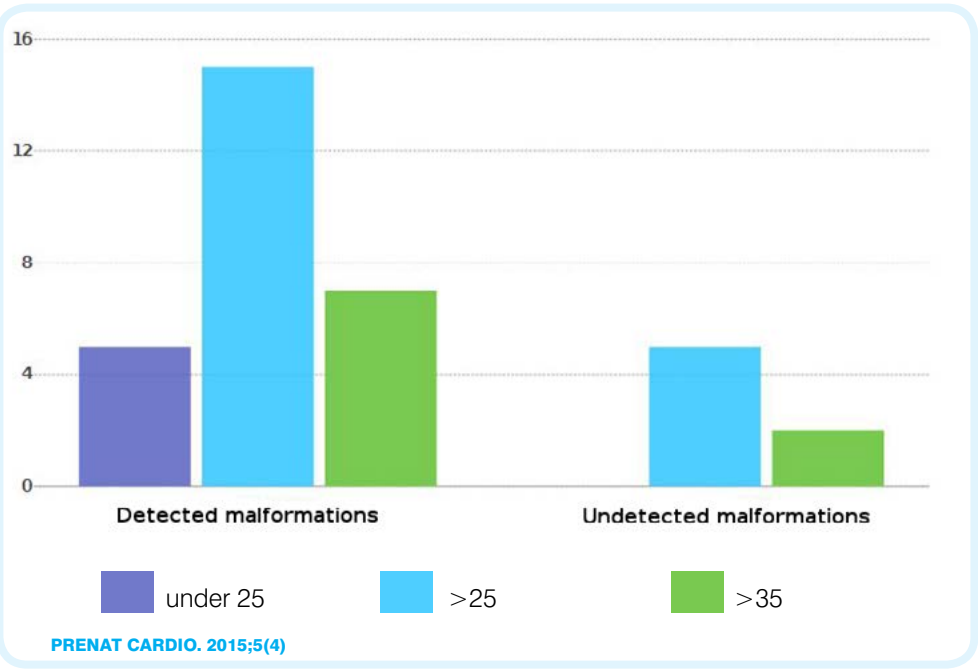


Fig.11: Detection of congenital malformations by biochemical and genetic-tests in different age groups

The next aspect worth drawing attention to is the quality of US examinations, which is disappointing. It needs improvement by special traineeships for obstericians in order to become a more reliable tool of prenatal diagnosis.

CONCLUSIONS

1. Although the access to basic US exam is easy and common, it was the least effective method of making appropriate prenatal diagnosis of congenital malformations in our cohort.

2. Fetal echocardiography had a high level of sensitivity and specificity in detecting congenital malformations.

3. Prenatal cardiologists proved to be the most effective in detecting congenital malformations with 89,3% of detected abnormalities.

4. Biochemical exams had a positive result in only one case (with the postnatal outcome of confirmed DS).

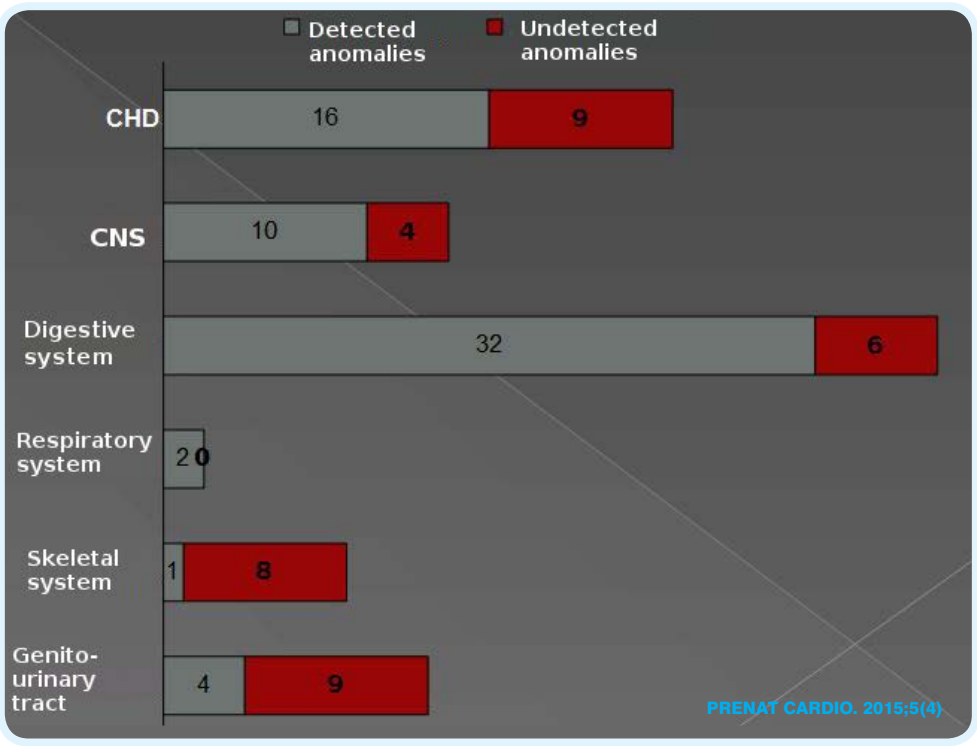


Table 1: Percentage of groups of prenatally detected malformations hospitalized in Neonatal Intesive Care Unit

References:

1. Respondek-Liberska M.: Prenatal cardiology, Czelej, Lublin 2006
2. Perenc M.: Prenatal diagnoses of congenital malformations - invasive and noninvasive methods. Przew. Lek. 2002, 5, 5, 94-99
3. Dangel J.:Prenatal diagnoses: myths and reality. Nauka 3/2007, 31-47
4. Benoit RM, Copel JA. Antenatal screening with fetal echocardiography: when and how. Contemporary OB/GYN, Vol. 48, No 9, September 2003, p.59
5. Eik-Nes S, Lee W, Carvalho JS, Chaoui R, Copel J, Hecher K, Paladini D. Cardiac creening examination of the fetus: guidelines for performing the 'basic' and 'extended basic' cardiac scan. ISUOG Guidelines Ultrasound Obstet Gynecol 2006; 27: 107-113
6. Respondek-Liberska M.: Prenatal echocardiography and prenatal cardiology, Mak-Med, Gdańsk 1998
7. Gembruch U. Prenatal diagnosis of congenital heart disease. Prenat Diagn. 1997 Dec;17:1283-98
8. Respondek-Liberska M. Prenatal echocardiography in 1st,2nd and 3rd trimester in singelton pregnancy. Prenatal Cardiology 2012,2,6-11
9. Respondek -Liberska M. Atlas wad serca u płodu. ADI 2011, 32.
10. Wójtowicz A. Detection of fetal heart defects based on screening protocol of prenatal heart examination . Prenat Cardio 2012, 2, 11-16
11. Sharony R, Fejgin MD, Biron-Shental T, Hershko-Klement A, Amiel A, Levi A.: Who should be offered fetal echocardiography? One center's experience with 3965 cases. Isr Med Assoc J. 2009, 11, 542-5
12. Sharland G.: Fetal cardiac screening: why bother?. Arch Dis Child Fetal Neonatal Ed. 2010, 95:F64-8
13. Wigton TR, Sabbagha RE, Tamura RK, Cohen L, Minogue JP, Strasburger JF.: Sonographic diagnosis of congenital heart disease: comparison between the four-chamber view and multiple cardiac views. Obstet Gynecol. 1993, 82,219-24
14. Warrier D, Saraf R, Maheshwari S, Suresh P, Shah S.: Awareness of fetal echo in Indian scenario. Ann Pediatr Cardiol. 2012, 5, 156-9
15. Gottliebson WM, Border WL, Franklin CM, Meyer RA, Michelfelder EC.: Accuracy of fetal echocardiography: a cardiac segment-specific analysis. Ultrasound Obstet Gynecol. 2006, 28, 15-21
16. Abu-Rustum RS, Daou L, Abu-Rustum SE.: Role of ultrasonography in early gestation in the diagnosis of congenital heart defects. J Ultrasound Med. 2010, 29, 817-21
17. Plesinac S, Terzic M, Stimec B, Plecas D.: Value of fetal echocardiography in diagnosis of congenital heart disease in a Serbian university hospital. Int J Fertil Womens Med. 2007, 52, 89-92
18. Słodki M, Rychik J, Moszura T, Janiak K, Respondek-Liberska M.: Measurement of the great vessels in the mediastinum could help distinguish true from false-positive coarctation of the aorta in the third trimester. J Ultrasound Med. 2009, 28,1313-7
19. Respondek-Liberska M, Sysa A, Gadzinowski J: Financial comparison of neonatal transportation: in utero, by ambulance and by air transportation. Ginekol. Pol. 2004, 75, 326- 331
20. Chaoi R, Rake A, Heling KS. Aortic arch with four vessels: Aberrant right subclavian artery.Ultrasound Obstet Ginecol. 2008,31,115-117.
21. Allan LD, Crawford DC, Chita SK. Prenatal screening for congenital heart disease. British Medical Journal.1986, 292, 1717-1719.
22. Carvalho JS, Allan LD, Chaoui R, et al; International Society of Ultrasound in Obstetrics and Gynecology. ISUOG Practice Guidelines (updated): sonographic screening examination of the fetal heart. Ultrasound Obstet Gynecol. 2013;41:348–359
23. Kemper AR, Mahle WT, Martin GR, et al. Strategies for implementing screening for critical congenital heart disease. Pediatrics. 2011;128(5). Available at: www.pediatrics.org/cgi/content/full/128/5/e1259
24. Rebecca F. Liberman, MPH, Kelly D. Getz, PhD,Angela E. Lin, Cathleen A. Higgins,Sepehr Sekhavat, Glenn R. Markenson and Marlene Anderka. Delayed Diagnosis of Critical Congenital Heart Defects: Trends and Associated Factors Pediatrics 2014;134;e373
25. Dawson AL, Cassell CH, Riehle-Colarusso T,et al. Factors associated with late detection of critical congenital heart disease in newborns. Pediatrics. 2013;132(3). Available at: www.pediatrics.org/cgi/content/full/132/3/e604
26. Oster ME, Lee KA, Honein MA, RiehleColarusso T, Shin M, Correa A. Temporal trends in survival among infants with critical congenital heart defects. Pediatrics.2013;131(5). Available at: www.pediatrics.org/cgi/content/full/131/5/e1502
27. L. R. Freud, A. Moon-Grady, M. C. Escobar-Diaz, N. L. Gotteiner, L. T. Young, D. B. McElhinney and W. Tworetzky: Low rate of prenatal diagnosis among neonates with critical aortic stenosis: insight into the natural history in utero
28. Respondek-Liberska M, Sklansky M, Wood D, Słodki M, Weiner S, Cuneo B, Huhta J, Gembruch U, Rizzo G, Sharland G, Achiron R, Pruetz J. Recommendations for fetal echocardiography in singleton pregnancy in 2015. Prenat Cardio 2015, 2, 28-34
29. Chaoui R. Fetal echocardiography: state of the art of the state of the heart. Ultrasound Obstet Gynecol. 2001;17:277-84

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Divison of work:

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M. Słodki: work with the manuscript

A. Zieliński: work with the manuscript

Iwona Maroszyńska: work with the manuscript,

Maria Respondek-Liberska: work with manuscript, final version