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Original Research Article

Efficacy of the regional reference system of prenatal diagnosis based on the analysis of results of Maternal-Fetal Medicine Centre in Olsztyn

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ARTICLE INFO

Article history:
Received 29 July 2013
Received in revised form
2 November 2013
Accepted 17 March 2014
Available online 26 April 2014

Keywords:
Congenital malformations
Prenatal diagnosis
Ultrasound diagnosis
Epidemiology of birth defects
Congenital heart defects

ABSTRACT

Introduction: Congenital malformations are morphological abnormalities acquired in intrauterine life and identifiable at birth.
Aim: The aim of the analysis of outcomes of the Maternal-Fetal Medicine Centre (MFMC) practice in Olsztyn in the years 1998–2012 was to assess the efficacy of regional reference system of prenatal diagnosis.
Material and methods: MFMC in Olsztyn performs reference non-invasive prenatal diagnosis for Warmia-Masuria Province. Patients are referred to MFCM by general gynecologists in the case of a suspected malformation or difficulty in the assessment of fetal morphology, and compulsory in the case of women above 35 years of age (NHF program). The analysis included total number of identified malformations, types of abnormalities, maternal age and gestational age at the time of diagnosis, geographic distribution in the province, number of identified malformations that were reported in national records of Polish National Registry of Fetal Cardiac Pathology.
Results and discussion: In the material of MFMC urinary tract defects were predominant. There was a significant amount of defects in children of mothers below 35 years of age. Most defects were diagnosed in the first pregnancy. Mothers of infants with malformations more often lived in urban areas. Most defects were diagnosed in pregnant women living in Olsztyn and geographically closest counties, and the least defects were diagnosed in patients living on the eastern edge of the province.
Conclusions: Results of the analysis confirm the significance of the presence of a facility with a profile similar to MFMC in our region. Factors that improve detection of fetal abnormalities in our region may include: extension of the NHF program to the entire population of pregnant women, improvement of first trimester diagnosis, increase of the availability of reference fetal echocardiography in pregnant women over the age of 35. Infrastructure improvement in the province may also positively affect the detection of anomalies.

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http://dx.doi.org/10.1016/j.poamed.2014.03.001
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1. Introduction

Congenital malformations are morphological abnormalities acquired in intrauterine life and identifiable at birth.[1] It is estimated that congenital malformations and genetic disorders are the cause of 50% of miscarriages in the preimplantation period and approximately 30% in later stages of pregnancy.[2]

According to statistics, approximately 2.0%–3.0% of children are born with at least one congenital malformation and in 1.4% of newborns multiple congenital anomalies are found. In about 0.6%–1.0% of infants chromosomal anomalies are found.[3]

Congenital fetal anomalies are among prematurity and utero-placental insufficiency the most common cause of perinatal and neonatal mortality. The most predominant congenital abnormalities include central nervous system (CNS), cardiovascular and urinary tract malformations.[1,4,6]

Maternal-Fetal Medicine Centre (MFMC) in Olsztyn exists since 1996. It performs reference non-invasive prenatal diagnosis – up to 2006 in accordance with the program ''Prenatal Centre'' developed by a group of Centers and approved by NHF, and since 2006 on the basis of the nationwide ''Program of comprehensive intrauterine diagnosis and therapy in the prevention of consequences and complications of congenital malformations and fetal diseases'' created by the NHF.

The basis for the referral of pregnant woman to MFMC is suspected anatomical abnormality at a routine ultrasound examination performed by general gynecologist, or difficulty in the evaluation of fetal morphology. Pregnant women above 35 years of age are referred to obligatory examinations (NHF program).

The purpose of the MFMC is to improve the prenatal detection of malformations in Warmia-Masuria Province. Prenatal diagnosis of congenital defects results in the change of pregnancy care model, determination of type of prenatal and neonatal care, specifying indications for extended diagnostics, including genetic testing, and identifying the optimal place, time and mode of delivery for the diagnosed pathology. All of the above mentioned elements have a beneficial effect on mortality attributable to birth defects and the improvement of long-term prognosis for health and quality of development of children.

Given the specified objectives, a significant organizational concept of the Center includes cross-functional team of MFMC (obstetrician-gynecologist, neonatologist) and close cooperation with geneticist and reference centers.

2. Aim

Analysis of the results of MFMC practice in the years 1998–2012 presented in the article aims to assess the effectiveness of the implemented regional reference system of prenatal diagnosis with regard to the above aim. Raising this issue also resulted from the exceptional situation of MFMC, being the only site with such profile in this region, which is an unusual situation in the country.

3. Material and methods

The analysis included the total number of identified malformations, types of abnormalities, maternal age and gestational age at the time of diagnosis, geographic distribution in the province, number of identified malformations that were reported in national records of Polish National Registry of Fetal Cardiac Pathology.

4. Results

In the analyzed 14-year period more than 26 000 ultrasound examinations of the fetus were performed. Number of examinations per year has increased from 350 in 2000 to 3500 in 2011. During this period, 840 fetal defects were diagnosed.

In Table 1 number and type of diagnosed defects are presented. Unlike statistics, the material of MFMC shows that urinary tract defects are predominant. This is due to the fact that borderline dilations of pelvicalyceal system, in which postnatal diagnosis of the urinary tract was recommended, were also included in this group.

Table 2 shows maternal age at diagnosis of fetal defects. Predominant number of fetal defects in patients above 35 years of age is noticeable.

Data presented in Fig. 1 confirm the validity of echocardiographic evaluation of the fetal heart at 18–22 weeks gestation in patients above 35 years of age, which results from percentage distribution of identified cardiovascular defects in relation to age.

Table 1 – Type of birth defect according to ICD10.

Defects	Number of cases
Urinary tract	271
Central nervous system	199
Cardiovascular system	162
Musculoskeletal system (i.e. Omphalocele, Gastroschisis, congenital diaphragmatic hernia)	102
Other	36
Gastrointestinal system	31
Reproductive system	30
Respiratory system	6
Multiple	3
Total	840

Table 2 – Age of patients with confirmed birth defect.

Age of patients	Number of cases
<20	101
21–25	242
26–30	249
31–35	145
36–40	75
>40	23
No data	5
Total	840

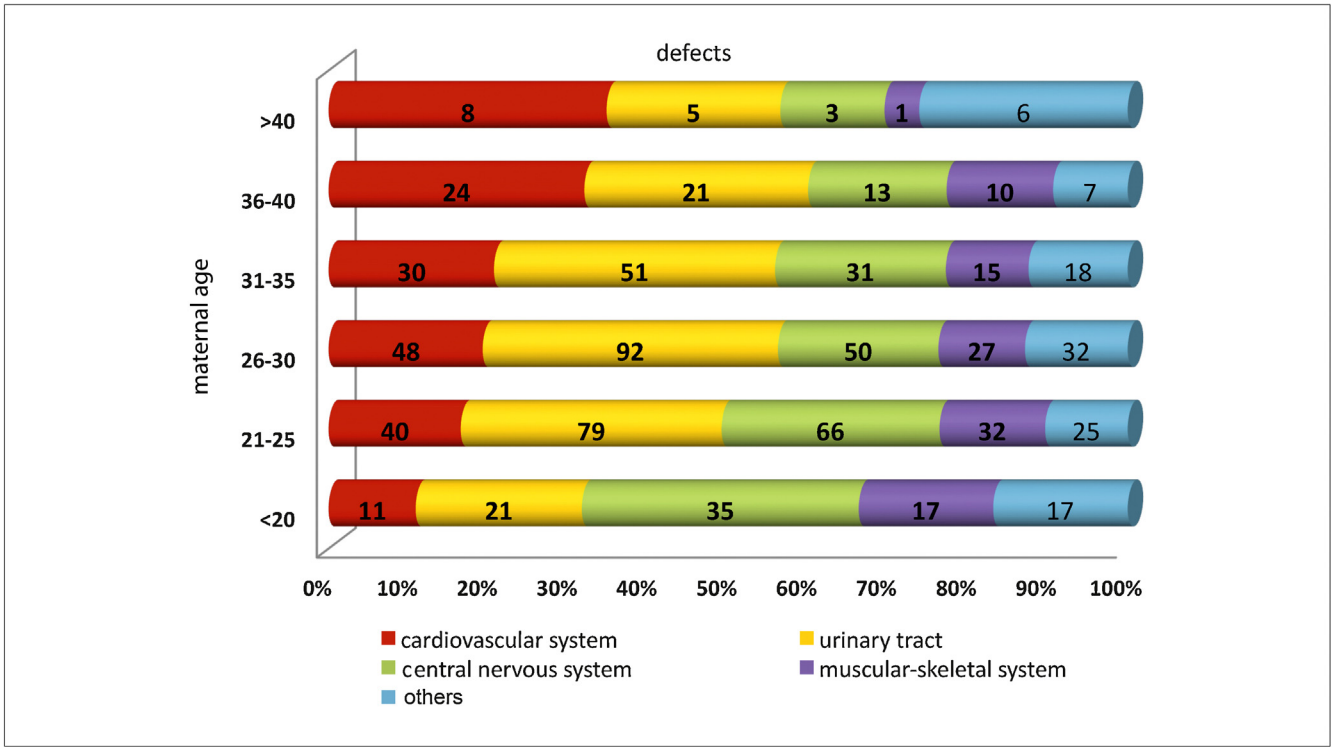

Fig. 1 – Defects and maternal age.

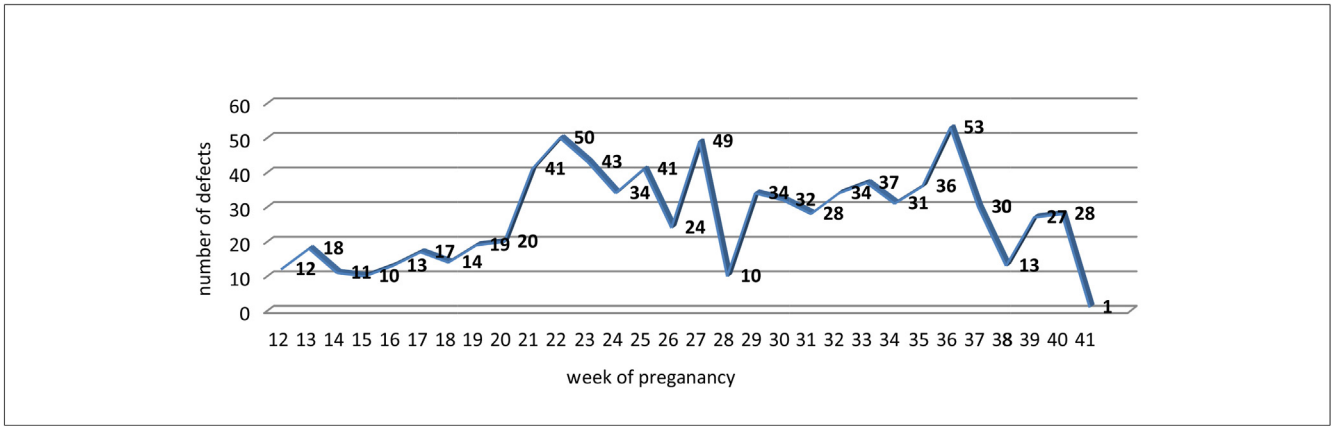

Fig. 2 – Identification of defect and week of pregnancy.

Summary of the number of identified defects in relation to maternal age at diagnosis (Fig. 2) demonstrates the improvement of screening recommended by Polish Gynecological Society[5] (12, 20, and 30 weeks gestation) in our province. At the same time, it proves the need to improve quality of first trimester diagnosis.

In case of cardiovascular system detection of defects is the highest at 20–29 weeks gestation (Fig. 3), which is consistent with the literature data.

In the urinary tract characteristic increase in the number of diagnosed defects within 35–40 weeks gestation (Fig. 4) results from detection of hydronephrosis and urinary tract obstructions in the third trimester of pregnancy.

Numeric distribution of diagnosed CNS malformations confirms the beneficial effect of screening at 20–22 weeks gestation (Fig. 5). In the third trimester dilations of the cerebral ventricular system are predominant – ventriculomegaly and hydrocephalus, that may become visible only during this stage of pregnancy (postinfectious, neoplastic). First trimester screening resulted in earlier detection of lethal defects of the CNS – acrania, holoprosencephaly.

In ICD-10 system musculoskeletal defects consist of a wide variety of anomalies, which include, above all, hernia, gastroschisis, achondroplasia, etc. There are two characteristic peaks, as seen in Fig. 6 – around 13 weeks gestation, when gastroschisis and umbilical cord hernia are primarily diagnosed (marker of

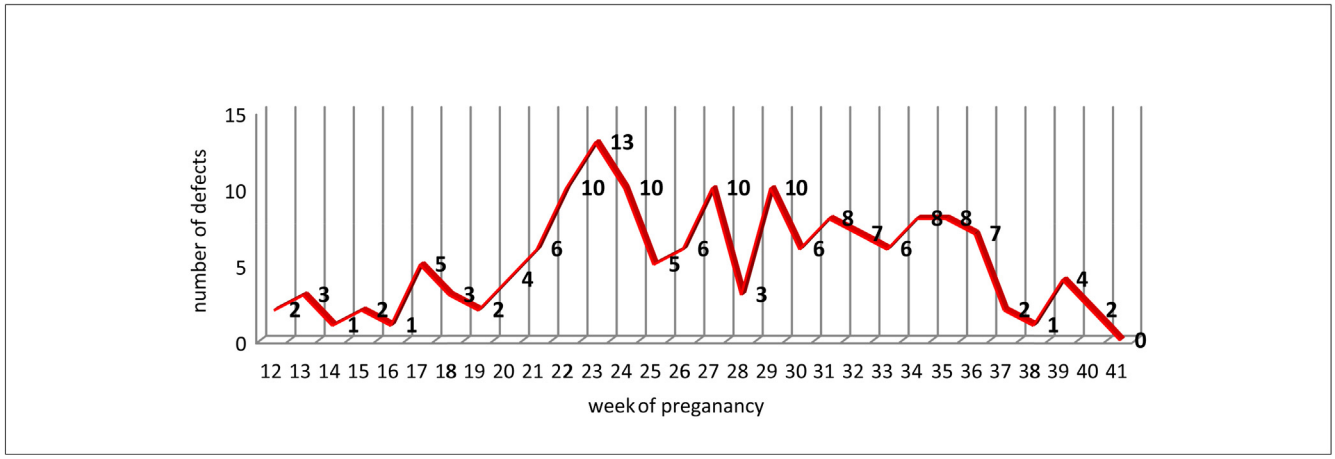

Fig. 3 – Identification of heart defects and week of pregnancy.

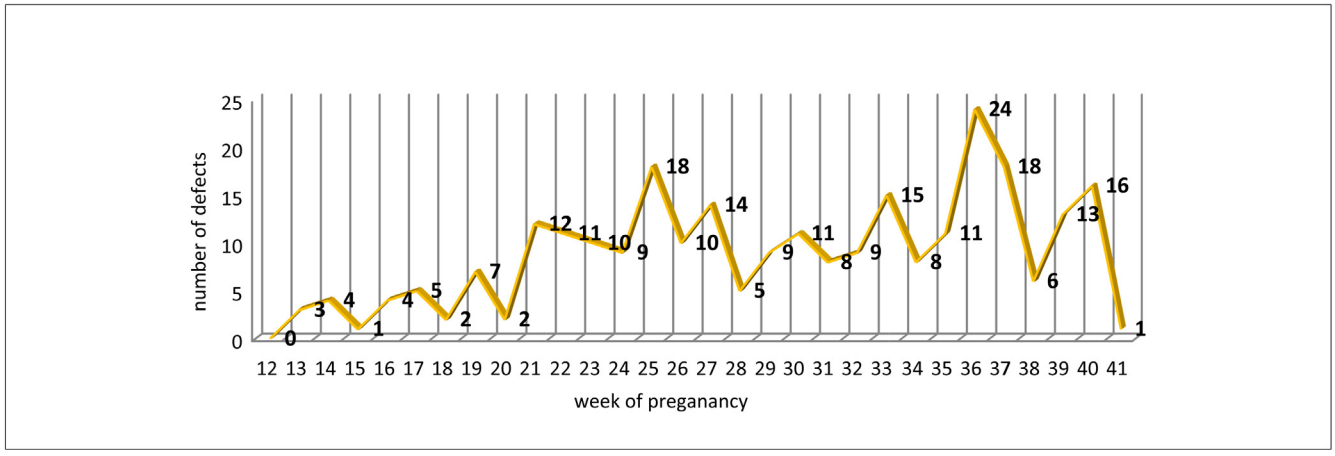

Fig. 4 – Urinary tract defects and week of pregnancy.

chromosomal aberrations!) and around 20 weeks gestation, when skeletal defects and other abnormalities from this group are readily visible.

Data gathered by MFMC (Table 3) reflect the demographic situation in Poland (current fertility rate approximately 1.3)[7]; thus, most defects were diagnosed in the first pregnancy.

Predominance of urban patients (Table 4) appears to result from the greater availability of medical services in the urban population.

Table 3 – Number of pregnancies and identification of defect.

Number of pregnancies	Cardio-vascular system	Urinary tract	Central nervous system	Musculoskeletal system	Other	Total
1	61	115	95	54	49	374
2	42	83	66	27	23	241
3	23	46	19	14	19	121
4	20	10	9	4	12	55
5	7	10	3	0	0	20
6	5	4	2	1	1	13
7	2	1	2	1	0	6
8	1	0	0	0	0	1
9	0	1	1	0	0	2
10	0	0	1	0	1	2
No data	1	1	1	1	1	5
Total	162	271	199	102	106	840

Table 4 – Identification of defect and place of residence – urban/rural.

Urban/rural	Cardio-vascular system	Urinary tract	Central nervous system	Musculoskeletal system	Other	Total
Rural	55	94	84	46	41	320
Urban	105	175	114	55	63	512
No data	2	2	1	1	2	8
						840

Table 5 – Number of defects per district.

District	Number of defects
Olsztyn	232
Ostróda	70
Szczytno	52
Iława	51
Kętrzyn	50
Bartoszyce	49
Elbląg	41
Mrągowo	39
Pisz	38
Nidzica	29
Ełk	28
Lidzbark Warmiński	26
Nowe Miasto Lubawskie	26
Giżycko	21
Braniewo	15
Olecko	15
Gołdap	14
Działdowo	10
Węgorzewo	8
Outside	12
No data	14
Total	840

Table 6 – Heart defects identified antenatally.

Diagnosis	Number of cases
Q21 Congenital malformations of cardiac septa	58
Q20 Congenital malformations of cardiac chambers and connections	33
Q24 Other congenital malformations of heart	32
Q22 Congenital malformations of pulmonary and tricuspid valves	22
Q23 Congenital malformations of aortic and mitral valves	15
Q25 Congenital malformations of great arteries	2
Total	162

Table 7 – Heart defects identified and referral centers.

Referred to	Number
Łódź ICZMP	27
Warszawa Karowa	43
Total	70

Most defects were diagnosed in pregnant women living in Olsztyn and geographically closest districts, and least defects were diagnosed in patients living on the eastern edge of the province (Table 5).

In the analyzed period, in MFMC 161 fetal congenital heart defects were diagnosed. This number comprises both isolated heart defects and defects coexisting with congenital malformation syndromes. In the analyzed material, heart defects most frequently coexisted with CNS malformations (Table 6).

Among 161 defects diagnosed in MFMC, in 70 cases pregnant women were referred to referral centers, as presented in Table 7. These were serious, complex malformations that required cardiosurgical treatment after delivery that is currently unavailable in Warmia-Masuria Province.

Cardiac defects diagnosed antenatally have to be reported to the Polish National Registry of Fetal Cardiac Pathology. In the analyzed period MFMC reported 82 defects to this registry.

5. Discussion

Diagnosis of fetal congenital defects has been possible since the introduction of ultrasound in medicine. Prenatal diagnosis of defects, in particular cardiac defects, has significantly improved pregnancy care, optimized perinatal management and allowed the appropriate treatment at the optimum time. All of this factors resulted not only in the reduction of mortality in case of defects that require fast surgical intervention (cardiac defects!), but also in the improvement of the quality of life of newborns and children with malformation, in whom diagnosis and treatment were implemented before the central manifestation and its complications (e.g. urinary tract defects).

The presented analysis refers to a group of patients for which there is no control group. The authors are also not aware of the existence of a site with a similar range and mode of action (the only one in the region), with which results could be compared. Conclusions of this analysis indicate directions that might improve the effectiveness of prenatal diagnosis of congenital malformations in our region.

Table 1 confirms the validity of the existence of specialized sites that perform prenatal diagnosis on a regional reference level. Diagnosis of 840 defects in the analyzed period allowed an adequate pregnancy, fetal and neonatal care, which is the principal purpose of MFMC.

The analysis provides arguments for continuation of the NHF program in relation to the entire population of patients of a childbearing potential, not only above the age of 35 (Table 2). Figs. 2–6 indicate the need to improve first trimester diagnosis, which should result in increased number of early diagnoses of serious defects, as well as rapid identification of high-risk pregnancies for obstetric reasons (the inverted pyramid principle[8]). The improvement of communication system in our province (public transport, roads) should also have a positive effect on the detection of defects. Tables 4 and 5

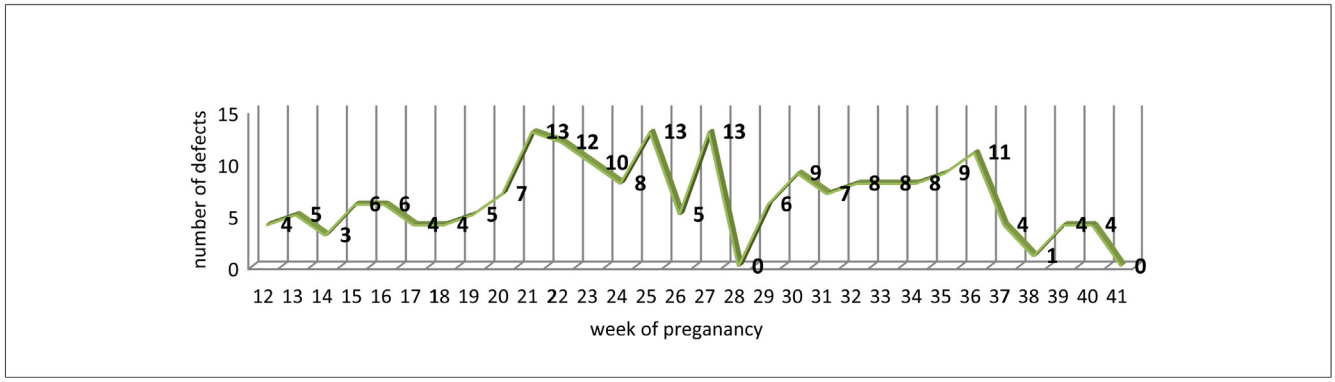

Fig. 5 – Central nervous system defects and week of pregnancy.

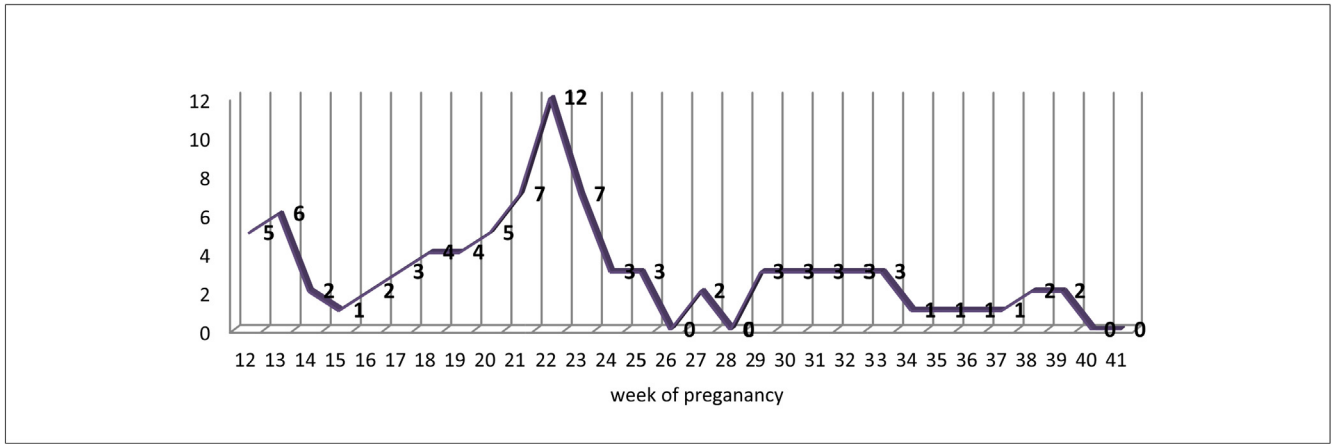

Fig. 6 – Musculoskeletal system defects and week of pregnancy.

illustrate the lower number of malformations identified in patients from rural and distant districts.

Fetal echocardiography has a significant place in the prenatal diagnosis, due to the required experience, difficulty of the examination itself, and necessary equipment. Data presented in Fig. 3 demonstrate that one of the ways of improving detection of cardiac defects in our region includes increasing the availability of reference fetal echocardiography in pregnant women above 35 years of age.

Further improvement of congenital malformation diagnosis is certainly dependent on a stable, effective reference system in the region and regular improvement of fetal ultrasound evaluation skills of general gynecologist.

It appears that results of the above analysis confirm that our regional reference site accomplishes all of the above aims.

6. Conclusions

1. Prenatal diagnosis of congenital malformations has allowed implementation of the adequate pregnancy, fetal and neonatal care.
2. Extending the NHF program to the entire pregnant population seems rational.
3. Improvement of first trimester diagnosis and increasing the availability of reference fetal echocardiography in pregnant women above 35 years of age should be important.

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