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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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ABSTRACT

Introduction: Congenital malformations are morphological abnormalities acquired in intra-uterine 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 MFMC 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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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 pre-implantation 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.^{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

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