Obstetrica şi Ginecologia LXV (2017) 189-192 **Editorial**

IS A NATIONAL PRENATAL SCREENING PROGRAM ACHIEVABLE IN ROMANIA?

Alina Veduţă*, G. Peltecu*,**

** Filantropia Clinical Hospital, Bucharest*
*** Carol Davila University of Medicine and Pharmacy, Bucharest*

The progress of prenatal diagnosis has been remarkably fast. Doctors as well as national health care systems have to keep up with rapidly evolving technologies and medical paradigms, in the interest of patients (1).

The ideas of universal prenatal screening and of universal ultrasound evaluation in pregnancy are actually not new (2). Screening for common genetic anomalies and detection of major fetal structural defects are thought to improve pregnancy outcome, therefore they are considered good practice in prenatal care across Western Europe and in other developed countries (3-5). Earlier diagnosis (6-7) as well as prediction and prevention of medical complications in pregnancy (8-9) are nowadays being implemented in the routine prenatal care.

The usual and most accepted screening for fetal anomalies involves two ultrasound examinations in pregnancy, at 11 – 14 weeks of gestation and at 20 – 24 weeks of gestation (2).

The 11 – 14 weeks ultrasound examination aims to identify fetuses at risk for frequent genetic problems such as trisomy 21 (Down's syndrome) and to diagnose major structural defects (1). The standard method of evaluating the risk for trisomy 21 (Down's syndrome) and other related aneuploidies is the Fetal Medicine Foundation (FMF) combined test, at 11 – 14 weeks of gestation. In the near future, when the cost of test becomes lower, the analysis of cell free fetal DNA in maternal blood will increase patients' access to accurate screening for aneuploidies; cell free fetal DNA in maternal blood analysis might soon become a first - line screening test for trisomy 21.

At 20 – 24 weeks of gestation, a detailed survey of the fetal anatomy (morphology) is usually undertaken (2-5).

New methods of prediction and prevention of obstetrical complications (such as preeclampsia) are just being introduced as a standard of clinical care, in advanced medical systems. This would be an opportunity to set up in Romania, at a national level, a comprehensive system that reflects the state of the art in modern obstetrics.

Good progress has lately been made in prenatal diagnosis, in Romania. Local centers have successfully implemented the Fetal Medicine Foundation (FMF) first trimester combined test (10); their results can be used as pilot studies and their activity can be further developed at a national level. In a favorable context, we have to seize opportunities and to analyze if a national screening program for fetal anomalies could be achievable at this point in time, in our country.

Our understanding is that there is official acknowledgement of the need for national screening and prevention programs. At present, European funding could be a solution for setting up national screening programs, including a program of prenatal screening and a national registry for fetal anomalies (both genetic and structural).

CORESPONDENCE: Alina Veduţã, e-mail: alina.veduta@yahoo.com

A number of issues are to be addressed in order to achieve national implementation of prenatal screening.

1. (Under) funding: Funding is relevant to all other issues on resources availability. European funds could be a solution for implementing national screening programs, including a program of prenatal screening.

Funding can be asked for only if relevant regulation states that all pregnant women are entitled to specific prenatal diagnosis services. Unfortunately, up to date regulations are not available in our case, as later discussed. As well, we are not aware of a systematic cost analysis or of any recent health technology assessment for fetal screening services, in Romania.

2. Regulatory background and updating of the description of the medical services for pregnant women that are covered by the public health insurance: The antenatal care in Romania is regulated by a document of the Ministry of Health, issued in 2004 and last updated in 2008 (OMS 2004/ OMS 2008) (11-12).

For long, the specific mechanism of implementing the regulation of the Ministry of Health was the National Program for Women's and Children's Health (*PNS VI – Programul naţional de sănătate a femeii şi copilului*). The subtitle 3.3 (now 2.3) of the national program PNS VI refers to the prevention of congenital anomalies, by means of prenatal and postnatal diagnosis. In practice, the funding was originally directed towards the conventional diagnosis of common aneuploidies in older pregnant women. Funding was from the beginning insufficient for reaching the goal of completely investigating the selected subgroup of patients; nowadays, such an approach would face the challenge of the ever increasing average maternal age. Over time, although all effort has been made to improve results, the program has only reached a minority of patients.

A broader – reaching mechanism has to be developed in order to provide basic fetal ultrasound evaluation, screening for genetic anomalies and for obstetrical complications to all pregnant patients in Romania as well as genetic counseling and genetic testing in a well – selected subgroup of pregnancies. The existing regulations and guidelines have to be updated to the new paradigms and to the modern technologies of prenatal diagnosis (population screening, contingency screening, combined first trimester screening for aneuploidies, analysis of cell-free fetal DNA in maternal blood, molecular karyotyping, Next Generation genetic tests). Moreover, new procedures (fetal therapy interventions, for instance) have to be endorsed and, if possible, covered by the public health insurance. This would involve changing the existing list of diagnoses and procedures that are endorsed by the public health care system (a document known as *Contract Cadru*).

3. Lack of reporting results, of auditing and lack of a National Registry: Funding is needed in order to create a system of reporting and auditing the results of the prenatal diagnostic services. Basic data such as the incidence and prenatal detection rate for major fetal anomalies is unfortunately missing in Romania. Until this problem is solved, a cost – effectiveness analysis of existing or proposed prenatal screening protocols and programs is not possible. Compulsory reporting to a functional National Registry of Fetal Anomalies would serve the purpose to enable the auditing of our prenatal diagnostic activity.

4. Patient information and patient access to prenatal services paid by the public health insurance: If necessary funding is found, campaigns of information on prenatal care and prenatal diagnosis can be conducted. We think that effective patient information can be done inexpensively. Correct and pertinent information would increase patients' access to prenatal diagnosis services, if such services are available.

The availability of the services fundamentally depends on their cost and the number of specialized health care providers.

5. Availability of services providers (doctors and other trained medical professionals in the field of obstetrics, fetal medicine and genetics): The small number of health care providers, including obstetrician and geneticists, is a worsening problem in many places across Romania.

6. Training in prenatal diagnosis: Training in prenatal diagnosis (extended ultrasound training during the postgraduate training in obstetrics and gynecology, practical training and continuous medical education for doctors holding national certificates of competence) is the direct solution for increasing the availability of the providers of fetal screening services. This solution is the least dependent on supplementary funding, as there already is important public funding for specific postgraduate training – a national 5-year postgraduate training program in obstetrics and gynecology (the Obstetrics and Gynecology Residency Program). At present, there are up to 600 obstetricians in training in Romania; more than 100 doctors are enrolled in postgraduate obstetrics and gynecology training, every year. Updating, better supervision and auditing of the obstetrics and gynecology residency program are necessary in order for the new specialists to be able to consistently provide modern prenatal diagnosis services.

Foreseeable patients will find less and less acceptable the lack of access to basic services such as first trimester assessment with correct pregnancy dating, screening for major fetal defects, timely referral to tertiary maternal – fetal medicine centers of high risk or abnormal pregnancies.

Regardless of the practical difficulties, a national program of fetal screening (for common aneuploidies and major fetal structural defects) is, in our opinion, overdue. Even if not easily, solutions can be found. Good training in obstetrical ultrasound during postgraduate obstetrics and gynecology training is a way to improve the availability and the quality of fetal diagnosis. Specialized assessment in the beginning of the pregnancy may reduce overall medical costs (1). Considering the data that shows that the combined first trimester test for aneuploidies is a robust test, training programs in first trimester ultrasound could be developed while offering the test to the general population (10). The analysis of the cell free fetal DNA from maternal blood, an operator – independent test, might soon become part of a cost effective program for the prenatal detection of aneuploidies.

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