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PRENATAL DIAGNOSTICS IN THE GENETIC COUNSELING – VARNA

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ABSTRACT

The authors report data from the follow-up of 167 pregnancies in the Medical Genetic Counseling in Varna for the period 1996-2001. In 44% of the cases a primary prevention was aimed. It was established that parental age was the most common indication for prenatal diagnostics (PD) - in 29% of the pregnancies. It was followed by the effects of drugs and/or infectious agents (in 14%), impaired reproduction (in 12%), neural tube defects (in 11%), chromosomal diseases (in 8%), multiple anomalies (in 7%) and single gene disease (in 3%). PD under the indication of 'other diseases' was carried out in 16% of the pregnancies. Data were compared to those reported by the authors for the period 1983-1987. It is emphasized that PD is a unique preventive method of a high social value.

Key words: prenatal diagnostics, medical genetic counseling, prevention, Varna.

Prenatal diagnostics (PD) in the genetic counseling is a method for prevention of congenital anomalies and inherited diseases. It is well known that the incidence of the congenital anomalies in the general population is about 5% (3,4,7). Up to 50% of the children with congenital defects die during the first year of life (2). The incidence rate of the genetically determined diseases among the population in Bulgaria varies widely in different age groups (1,5). According to hospital records, 35% of the admitted patients have inherited disease (3). The substantial importance of the figured indexes makes it urgent to develop and apply PD.

The Medical Genetic Counseling in Varna works on applying PD and is a center for prevention of genetic diseases in North East Bulgaria. In this paper the results from the analysis of a five-year period (1996-2001) are presented and discussed. During this time PD was applied in 167 pregnancies. In 44% of the cases PD has been performed as a primary prevention while in the rest 56% as a secondary one.

PD has been carried out by using 3 basic approaches:
- Consecutive ultrasound examination of the development of the embryo and fetus
- Ultrasound follow-up and prenatal karyotyping
- Ultrasound follow-up and DNA analysis of the fetus.

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Forty percent of the pregnancies have been followed-up by applying combined ultrasound and cytogenetic or molecular genetic testing.

The distribution of the pregnancies according to the indications is the following:
- Multiple anomalies but not suspecting chromosomal aberration - 11 pregnancies (6,59%);
- Neural tube defects - 18 pregnancies (10,78%);
- Impaired reproduction not suspecting chromosomal etiology - 20 pregnancies (11,98%);
- Effects of drugs and/or infectious agents - 24 pregnancies (14,37%);

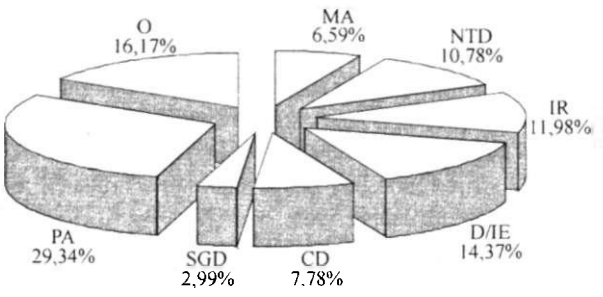

*Fig. 1. Case distribution according to indications. (**MA** multiple anomalies, not suspecting chromosomal aberration; **NTD** neural tube defects; **IR** impaired reproduction not suspecting chromosomal etiology; **D/IE** drug and/or infectious agents effects; **CD** chromosomal disease; **SGD** single gene disease where prenatal DNA analysis is available; **PA** parental age; **O** others)*

- Chromosomal disease - 13 pregnancies (7,78%);
- Single gene disease where prenatal DNA analysis is available - 5 pregnancies (2,99%);
- Parental age - 49 pregnancies (29,34%), and
- Others - 27 pregnancies (16,17%).

ary prevention – 56%. It is very close to the data for Bulgaria (55%) and Europe (52%) (3).

It can be concluded that PD in the genetic counseling practice is defined as a high-valued preventive method both for the family and the society.

Table1. Comparison of data from the investigations in 1983-1987 and in 1996-2001

Indications for PD	Distribution in %		Deviation in %
	1983-1987	1996-2001	
Multiple anomalies	17	12	- 5
Neural tube defects	16	20	+ 4
Impaired reproduction	27	22	- 5
Effects of environmental factors	9	26	+ 17
Chromosomal disease	25	14	- 11
Single gene disease	7	6	- 1

he most common indication for PD in the period 996-2001 is parental age followed by others, effects of en-ironmental factors, impaired reproduction, neural tube de-cts, chromosomal diseases, multiple anomalies and less ommon – single gene diseases (Fig. 1).

ese data are the reason to discuss the 'ageing' of the re-oducing couples as well as the inaccurate medical treat-ent of women in reproductive age and the living environ-ent in our villages.

previously published data (6) the indications of 'parental e' and 'others' have been of less importance for the pe-d 1983-1987. After some adaptation the figures for the iod 1996-2001 are compared to those for the period 83-1987 (Table 1). Though perfect correlation is not pos-le, Table 1 will be useful when discussing the factors that e led to the change in the distribution of the indications.

important to note the reduced percentage of the cases in ich PD is asked in the presence of 'hot' indications - omosomal disease, impaired reproduction and multiple malies. This fact points out the negative change in the tions between the family physician, woman's counsel-and genetic counseling in a situation of a structural re-in the health care services.

results of our observations show a slight prevalence of ases in when PD is done for the purposes of a second-

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