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INVESTIGATIONS OF THE CONTENT OF PRENATAL DIAGNOSIS IN THE CONTINGENT OF GENETIC COUNSELLING IN NORTH-EASTERN BULGARIA

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Diseases enabling a prenatal diagnosis (PD) are of varying incidence rate in human populations from different geographic areas [1-3]. Having in mind that fact we decided to investigate on the basis of the contingent of Varna Genetic Counselling (GC) the specificity of prenatal diagnosis required in North-Eastern Bulgaria.

MATERIAL AND METHODS

Our study covered a five-year period - from 1983 till 1987. It was done prior to the introduction of mass screening for neural tube defects. According to our material, there were 104 families (or 34 per cent of a total of 310 ones) who required confirmation or denial of the corresponding prenatal prophylaxis of a given disease or anomaly. Our examination covered a total of 784 individuals belonging to these 310 families with inherited diseases and/or malformations.

RESULTS AND DISCUSSION

Distribution of this contingent according to the occasion for GC is presented on table 1. However, the occasion is complex in some families. That is why sum percentage of single groups on table 1 increases illusively.

Table 1

Distribution of families considered for prenatal diagnosis corresponding to the cause for genetic counselling

No	Cause for genetic counselling	Distribution (in %)
1.	Reproductive anomalies	29
2.	Down's syndrome	27
3.	Multiple inborn developmental malformations	18
4.	Neural tube defects	17
5.	Monogenic diseases	8
6.	Environmental factors affecting pregnancy	10

It is evident that families with reproductive anomalies occur most frequently (in 29 per cent of the cases). They are cases with several spontaneous abortions predominantly in the first trimester of pregnancy where results from the obstetric-gynaecologic, andrologic and genetic examinations form the idea of necessary prenatal prophylaxis. Reproductive failures affect only marital couples which have visited GC, in 62 per cent of the cases while there is a familial history in the rest 38 per cent. No aberrations from the normal karyotype are observed in marital partners by means of cytogenetic analysis. Nevertheless, on the basis of the results from the whole genetic study and literature data available a realization of prenatal diagnosis in a subsequent pregnancy is indicated in 40 per cent of the families (with more than three spontaneous abortions).

Families with children suffering from Down's disease come second in our sample. Mean age of mothers is 24 years and of fathers - 29,5 years. The ill child is first-born in 64 per cent of the families, second-born for the corresponding marital couple - in 32,4 percent and third-born in the same family - in 3,6 per cent of the cases. Diagnosis of Down's syndrome is confirmed in the half of probands examined by karyotype determination. It is denied in 25 per cent of the cases and not controlled in the rest cases due to lethal outcome or some other objective reasons. Down's syndrome is presented by the following cytogenetic variants: a regular trisomy - in 64 per cent of the cases; translocation forms (inherited pat t 14/21 and new t 21/21) - in 21,4 per cent, and mosaic combinations of the type 46/47, +21 - in 14,3 percent of the cases. Absolute indications for prenatal prophylaxis of Down's disease are established in 6,3 per cent of the families (cases of proved balanced pat t 14/21 and mat mosaic combination).

About 18 per cent of the families are examined on the occasion of relation to various multiple inborn developmental anomalies. Forty per cent of the cases are associated with anamnestic data about an action of harmful exogenic factors chronologically related to the corresponding pregnancy. Besides in two thirds of multiple inborn developmental defects present sporadic nonclassified complexes. This creates difficulties for the subsequent genetic prognosis of a next pregnancy in corresponding marital couples and for the determination of PD necessity for every concrete case.

Neural tube defects (anencephaly, spina bifida occuring alone or in combination together or with other defects) are diagnosed in the offspring of 17 percent of marital couples. A hereditary history of spina bifida on the father's side is anamnestically proved in 5,6 per cent of them. PD necessity in a subsequent pregnancy in the same family is recognized not only in sporadic but also in familial cases with neural tube defects.

Of the families examined, 8 per cent show indications for PD on the occasion of degenerative diseases of the nervous system or cystic fibrosis. However, monotony of cases in this group which includes, in fact, monogenic diseases as well as the small number of families does not enable us to obtain reliable data. PD is required and possible in 37,5 per cent of the cases in the group although there exists a risk of diagnostic mistakes.

GC is realized because of the increased risk of embryonal affection by environmental factors of various kind and nature in 10 per cent of the families. Risk is elevated because of x-ray application for diagnostic purpose in 50 per cent of the cases. Danger of offspring anomalies is due not only to the affecting action of a given factor but also to pregnant woman's advanced age in 33 per cent of the cases. Interpretation of data concerning a possible mutagenic and teratogenic effect enables to consider an indicated pregnancy interruption in 30 per cent of the cases and a required PD in the rest 70 per cent.

In some families PD realization requires a combined examination and there are absolute indications for PD application in about 50 per cent of all families where PD is indicated.

We have calculated interfamilial correlations determined by the indicated prenatal diagnostic methods separately for cases with extended and with absolute indications for PD. Table 2 reflects the significant differences between these two digital series.

Table 2

Interfamilial correlations according to the indications and the appropriate method for prenatal diagnosis

Indications	Interfamilial range according to the method required for PD (in %)			
	Ultrasound fetal examination	Karyotyping the fetus	Maternal AFP	Biochemical fetal examination
Extended	76	62	17	5
Absolute	42.8	46.9	32.6	10.2

We accept that by the present study some indicative data are obtained about PD content in the region as well as about the activity of PD realizing sectors. In our opinion, these results present an appropriate basis for territorial, staff and supply planning of these divisions of public health service and for the preparation of their working programmes.

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ИССЛЕДОВАНИЕ ОБЪЕМА ПРЕНАТАЛЬНОЙ ДИАГНОСТИКИ КОНТИНГЕНТА МЕДИКО-ГЕНЕТИЧЕСКОЙ КОНСУЛЬТАЦИИ СЕВЕРОВОСТОЧНОЙ БОЛГАРИИ

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РЕЗЮМЕ

С целью исследования специфики необходимой в Северовосточной Болгарии пренатальной диагностики были определены соответствующие индикации для оценки этой диагностики. Исследование проводилось среди контингента медико-генетической консультации в Варне в течение пятилетнего периода (с 1983 по 1987 г.) до введения в практику массового скрининга для установления дефектов при закрытии невральной трубы. Устанавливается, что 50 % всех семей, при которых необходимо проведение пренатальной диагностики, имеют абсолютные индикации для осуществления этого профилактического подхода. 47 % семей нуждаются в цитогенетических исследованиях, 33 % семей показывают потребность в прослеживании сыворотки крови на АФП уровне. У 10 % всех семей имеются данные о биохимическом анализе зародыша. В некоторых случаях пренатальная диагностика требует комбинированного применения некоторых диагностических методов.