CONCERNING THE ROLE OF GENETIC COUNSELLING IN DIAGNOSTIC SPECIFYING OF ANOMALIES OF SEXUAL DEVELOPMENT

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It is known that the period of individual development is of essential importance for the effectivity of genetic counselling (GC) in persons with anomalies of deter ination and/or differentiation of sex. In our opinion, GC effect varies considerably in 6 age groups as follows: Ist — up to 1 year, IIrd — between 1 and 14 years, IIIrd — between 15 and 20 years, IVth — between 21 and 35 years, Vth — between 36 and 50 years, and VIth — over 50 yearsu. Because of these considerations, an excerpt of 142 individuals from the whole contingent of GC in the city of Varna (2) having a preliminary diagnosis of congenitally or hereditarily determined anomaly of sexual development was analyzed according to the age groups mentioned above.

Results and discussion

Patients' distribution according to sex and age during GC realization is demonstrated on table 1.

The comparison of the number of probands aged over and below 14 years reveals a ratio of 2.5 : 1, i. e. the group of persons entered pubertal age predominates considerably. It is established that in our material female patients aged

Tablo 1

Patients' distribution according to age and sex

Age, (years)	Civil sex					
	male		female		total	
	n	%	n	%	n	%
0 — 1	7	10.9	5	6.4	12	8.4
1 — 14	16	25.0	11	14.1	27	19.1
15 — 20	10	15.6	11	52.6	51	35.9
21 — 35	23	35.9	18	23.1	41	28.9
36 — 50	5	7.8	2	2.6	7	4.9
Over 50	3	4.7	1	1.3	4	2.8
Total	64	45	78	55	142	100

between 15 and 20 years are most frequently consulted in relation with defects of sexual development. Second, both males and females aged between 21 and 35 years come in our contingent studied. It stresses that boys in the age group up to 20 years are one and a half times more seldom orientated to GC in order to clarify the cause of the defect of sexual development. This fact is in discrepancy to the ratio between the most frequent genetically determined disturbances of sex development which are clinically manifested at puberty — the syndromes of Klinefelter and of Turner (1, 3, 4). We accepted that this fact is related to more demonstratively and alarmingly expressed clinical stigmata in jemale individuals with anomalies of the reproductive system. In our opinion, it is probable that some «boys up to 20 years of age» from our contingent are not correctly orientated during the process of specifying of the essence of the disease. This concept is confirmed by data indicating that mainly endocrinologists require our consulting aid when this age group is concerned.

There are only 19 per cent from the contingent oj GC in Varna who belong to the age group between 1 and 14 years although clinical symptomatics of numerous nosological units related with disturbances of sex development is already manifested at this age. We presume that most probably, this low percentage is a result from the incorrect idea of clinical specialists about GC activity.

In our contingent, 8 per cent of the cases studied belong to the age group up to one year when the most severe anomalies of sexual development appear and a high competence and preciseness of the physician — specialist of genetics is required. Usually, at this age the biological nature of the clinical diagnosis of hermaphroditism (in its different variants) is specified. So one can help the patient to obtain the most appropriate civil sex corresponding to his original and/or corrected phenotype.

In this study, 7.7 per cent of the patients orientated to our GC on the occasion of anomalies of sexual development are more than 35 years old and 1 per cent of them — even more than 50 years old. It is known that concerning the last group, clarifying of the biological nature of the disease has no practtical value for the patient any more. Our aspiration to use an adequate quantity of genetic investigations during our work on this contingent directed us to systematize initial patients' diagnoses. As a result, 4 groups were formed. The first group included clinical diagnoses (e. g. of hermaphroditism and gonadal dysgenesis without the cases of supposed Turner's syndrome) requiring the application of the whole set of genetic methods available. The second group consisted of the diagnoses (Klinefelter's syndrome, some cases of Turner 's syndrome, gynecomastia, cryptorchism, adreno-genital syndrome) the specifying of which insists on applying of reduced number of genetic examinations with low tendency towards changes. The third group included anomalies such as hypogonadism, some cases of Turner's syndrome, etc., requiring varying in number genetic methods in dependence on the results from preceding analyses. However, when the fourth group was concerned it has to be noted that preliminary diagnoses (amenorrhoea, azoospermia, hypogenitalism, hirsutism, etc.) presented designations of single symptoms of the anomaly of sexual development and incompletely reflected a given clinical examination. That is why they did not enable, practically, to determine an adequate optimal number of genetic examinations.

Material presented in the communication allows us to assume that GC can actively participate in the solving of complex problems of a good and timely correction of the anomalies of sexual development under condition of competent orientation of patients by corresponding clinical specialists and if necessary, of their further fruitful collaboration.

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О РОЛИ МЕДИКО-ГЕНЕТИЧЕСКОЙ КОНСУЛЬТАЦИИ ПРИ ДИАГНОСТИЧЕСКОМ ОПРЕДЕЛЕНИИ АНОМАЛИЙ ПОЛОВОГО РАЗВИТИЯ

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РЕЗЮМЕ

Посредством анализа шести соотносительных групп пациентов в зависимости от существенных в биологическом и социальном аспектах возрастных показателей, определялась роль медико-генетической консультации при уточнении диагноза у лиц с аномалиями в половом развитии. Для медико-генетической консультации в Варне установлено соотношение пробанд до и старше 14 лет — 2,5 : 1. Наиболее часто и в связи с отклонениями в половом развитии консультировались женщины в возрасте от 15 до 20 лет. Вторая группа мужчины в возрасте от 21 года до 35 лет. Молодые люди до 20 лет направлялись на 1,5 раз реже в медико-генетические консультации для установления причины аномалий в половом развитии. Лишь 19% пациентов, обратившихся к медико-генетическим консультациям в Варне составляют группу от одного года до 14 лет. Восемь процентов всего контингента составляют пациенты до одного года, 7,7% — старше 35 лет и 1% — старше 50 лет. По мнению авторов распределение по группам деформировано в связи с клиническими трудностями диагностики при работе с таким контингентом больных, с одной стороны, а с другой — в связи с недостаточной активностью при сотрудничестве специалистов из клиник и генетиков.

Проведено систематизирование предварительного диагноза с целью оценки их ведущего места при определении адекватного оптимального объема необходимых генетических исследований.