February 2007 · Vol. 9 · No. 2

Genetics and genetic counseling: Practices and opinions of primary care physicians in Turkey

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Purpose: The purpose of this study was to assess the educational needs of physicians relating to genetics and genetic counseling in the Denizli region of Turkey. **Methods:** Data were collected by questionnaire about physicians' approaches to genetics and genetic counseling. **Results:** A total of 60 (60.0%) of 100 physicians working in Denizli province returned a questionnaire. Physicians described "their most knowledgeable subjects" in basic genetic information as chromosome abnormalities (41.8%), in genetic disorders as xeroderma pigmentosum (80.0%), and in genetic counseling as directing the parents of and couples with a risk for having a child affected by a genetic disease to an expert or a genetic counseling center (94.8%). Only 20.7% knew the ethical regulations and techniques related to genetic counseling. Physicians thought that they did not have sufficient knowledge about genetics or genetic counseling, and 83.9% would like to attend an educational course. **Conclusions:** As a result of this study, a genetics course is planned for physicians so they can actively participate in the prevention and early diagnosis of genetic diseases. ***Genet Med* 2007:9(2):130–135.**

Key Words: *Genetics, physicians, genetic conseling, primary care*

The Human Genome Project is widely believed to be leading to a profound transformation in the practice of medicine and public health by facilitating the identification of genes causing or predisposing one to a host of human disorders, both common and rare. For better or worse, depending on one's point of view, medicine and health care are becoming "geneticized," and it is expected that genetic considerations will become important in all aspects of disease diagnosis, treatment, and prevention.[1] Over recent years there has been much debate about what role primary care physicians will play in genetics, but the field remains wide open. Initially they are likely to be involved in educational activities with their peers.[2] In many European countries, general practitioners (GPs) are the primary care providers and act as gatekeepers in the referral of patients to specialist care.[3] Several medical specialties serve as primary care providers in the United States, including family practitioners, general internists, general pediatricians, obstetrician-gynecologists, and primary care nurse practioners.[4] Genetic diseases are increasingly being recognized in developing countries, such as Pakistan, Iran, Indonesia, Tunisia, Turkey, and India.[5] Primary health care practitioners in lower-resource countries undertake a broad range of responsibilities, often in relatively isolated and difficult circumstances, with limited or distant contact with secondary or tertiary services.[6] In our country the increased awareness of the importance of early diagnosis of genetic illnesses has given those illnesses priority in primary health care.[7] Physicians who work in primary care in Turkey ensure that preventive and therapeutic services are provided. In particular, the physicians work together with midwives to provide maternal–child health care.[8] In their mother–child monitoring, midwives refer at-risk cases to physicians.[9] The majority of practicing physicians in Turkey's primary care facilities have a knowledge deficit about basic genetics and genetic counseling, which is an important reason why these services are limited.[10–17]

Therefore, education in genetics is an indispensable base on which to introduce programs to control genetic diseases and congenital disorders.[18] The purpose of this study was to assess the educational needs of physicians related to genetic diseases and genetic counseling in the Denizli region of Turkey. The findings from this study will help inform the development of an educational strategy on genetics for primary care.

MATERIALS AND METHODS

Permission was obtained from the Province of Denizli Health Ministry to conduct this study, and they were informed about the contents of the questionnaire. Only volunteers participated in the study, and the data were collected anonymously. The items on the questionnaire were developed on the basis of an article by Erdemir,[15] and a study by Mertens et al.[19] In addition, basic genetic information and disorders defined

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The authors declare no conflict of interest.

A supplementary Appendix is available via the ArticlePlus feature at www.geneticsinmedicine.org. Please go to the February issue and click on the ArticlePlus link posted with the article in the Table of Contents to view this material.

Submitted for publication April 10, 2006.

Accepted for publication November 12, 2006.

DOI: 10.1097/GIM.0b013e3180306880

by the World Health Organization (WHO) as "point-of-entry" disorders that can be prevented were also included. Questions related to genetic counseling were developed on the basis of WHO criteria in a public health study by Gökkoca[10] (see Appendix).

The questionnaire's validity and reliability were not researched because our goal was not to create a tool.

Procedures

All 26 of the health clinics in Denizli province were included in the study, and questionnaires were used. Physicians who worked at these health clinics were included in the study. The self-report method was used for data collection.

Measures

The following categories of information were asked on the questionnaires: sociodemographic information, basic genetic information, theoretical/practical information about some genetic diseases/disorders, genetic counseling experience, and sources for information.

Data analysis

In this cross-sectional and descriptive study, data were analyzed using the Statistical Package Program for the Social Sciences (version 13.0; SPSS Inc., Chicago, IL). Descriptive statistics, including mean ± standard deviation, frequencies, and percentages, were calculated in the current study. To determine the relationship between two categorical variables, the chi-square test was used. The statistical significance was set at the 5% level ($P \leq 0.05$).

RESULTS

Response rate and demographic characteristics

A total of 60 of the 100 physicians who work full time in the 26 health clinics in the province answered the questionnaire (response rate of 60.0%) and were included in the study. The majority of our respondents were male (56.7%). The age of the 60 physicians who answered the questionnaire ranged from 28 to 58 years, with a mean of 37.4 ± 6.11 years. The length of time in their profession ranged from 3 to 26 years with a mean of 12.64 ± 5.62 years (Table 1).

Table 1
Sociodemographic variables about physicians (n = 60)

Variable category	N	%
Age		
20–30 y	6	10.0
31–40 y	39	65.0
≥41 y	15	4.6
Years (mean ± SD)	37.40 ± 6.11	
Gender		
Female	26	43.3
Male	34	56.7
Profession		
Physicians	60	100.0
Attained educational degrees		
University (6 y)	60	100.0
Years working as a provider		
0–5	6	10.0
6–10	19	31.7
11–15	19	31.7
16–20	10	16.7
≥21	6	10.0
Years (mean ± SD)	12.64 ± 5.62	
Length of time in current job		
Years (mean ± SD)	11.04 ± 6.08	

SD, standard deviation.

Genetic knowledge

The examination of basic genetic information showed that the highest percentage (41.8%, n = 23) knew about chromosome abnormalities and that the lowest percentage (3.8%, n = 2) knew about polygenic inheritance (Table 2).

The examination of the sources of information about some genetic abnormalities and illnesses showed that the highest percentage (80.0%, n = 44) knew about xeroderma pigmentosum and that the lowest percentage (12.7%, n = 7) knew about diabetes mellitus (Table 3).

Knowledge associated with genetic counseling

For answers related to genetic counseling the highest percentage was 94.8% (n = 55) for directing at risk couples or parents to an expert or a genetic counseling center. The lowest was 20.7% (n = 12) for those who knew the ethical regulations and techniques related to genetic counseling. In addition, 21.1% were able to develop a family tree by learning the genetic history of individuals in whom a genetic disorder was suspected, 22.8% were able to organize a screening program for genetic diseases, 27.3% knew about the genetic counseling center in Denizli, and 55.4% stated that they knew about genetic diseases common in the region. The majority of the physicians (83.9%) stated that they would like to participate in an educational program (Table 4).

Sources of knowledge

The distribution of answers to the multiple-choice question about physicians' sources of information was, in order: class in medical school (n = 51), book (n = 49), press (n = 12), word-of-mouth (n = 2), course (n = 20), seminar (n = 22), conference (n = 23), and the Internet (n = 9).

Table 2
Physicians' answers related to basic genetics information

	Knowledgeable		Need more information		No information		
Topic	N	%	N	%	N	%	Respondents N
Chromosome abnormalities	23	43.8	31	56.4	1	1.8	55
Autosomal recessive disorders	22	40.0	33	60.0	—	—	55
Sex-linked defects	21	30.9	33	60.0	1	1.8	55
Mendelian inheritance	19	35.2	31	57.4	4	7.4	54
Human genetic diseases	17	30.9	38	69.1	—	—	55
Mitosis and meiosis	16	29.1	38	69.1	1	1.8	55
Autosomal dominant disorders	16	29.1	37	67.3	2	3.6	55
Prenatal diagnosis of genetic disease	16	29.6	35	64.8	3	5.6	54
Nucleic acids in protein synthesis	12	22.6	34	64.2	7	13.2	53
Principles of probability	11	20.8	31	58.5	11	20.8	53
Mitochondrial inheritance	8	14.8	26	48.1	20	37.0	54
Genes and mental retardation	7	12.7	45	81.8	3	5.5	55
Ethics of human genetics	7	13.7	30	58.8	14	27.5	51
Genes and the environment	6	11.3	36	67.9	11	20.8	53
Genetic screening	4	7.4	40	74.1	10	18.5	54
Genetic counseling	4	7.5	43	81.1	6	11.3	53
Genes, race, and intelligence (IQ)	4	7.4	42	77.8	8	14.8	54
Recombinant DNA	3	5.7	36	67.9	14	26.4	53
Genetic engineering	3	5.9	32	62.7	16	31.4	51
Polygenic inheritance	2	3.8	34	64.2	17	32.1	53

IQ, intelligence quotient.

DISCUSSION

Deficiency of genetic knowledge

Basic genetic counseling is both feasible and increasingly necessary in primary health care (WHO 1999).[20] Minimally, physicians will need sufficient knowledge about genetics to answer questions, identify at-risk patients, and refer appropriately.[21] In this study, most of the participants reported having insufficient knowledge about ethics of human genetics, genetic screening, counseling, engineering, and polygenic inheritance. Practicing physicians do not think they have enough information about genetic diseases or genetic counseling and recognize their need for more education. McGovern et al.[22] found that 79% of genetic counselors participated in the general education of physicians about genetic testing. The need for this education and its benefits in primary care have been defined by Kolb et al.[23] and Kirk.[24] In another study by Tomatır et al.,[9] the need for this education in primary care was emphasized.

With the rapid development of many potential gene therapies just on the horizon, all physicians will have major continuing medical education needs in the area of genetic diseases.[25] Primary care physicians need more education about the genetic component of many diseases to directly provide and appropriately refer for genetics services.[26]

Primary care providers and GPs are under increasing pressure from professionals involved in the delivery of genetics services to become more knowledgeable about genetics and more aware of the need to counsel and refer patients for genetic testing when appropriate. There have been several studies of GPs' knowledge of genetics and genetic services and of their receptiveness to an increased emphasis on genetics and genetic counseling in primary care. The majority of these studies have been undertaken in the United States and United Kingdom.[27] Watson et al.[28] believe there is a need to develop and evaluate a model for the delivery of genetic services that incorporates a realistic role for primary care and takes the views of primary health care professionals into account. Hunter et al.[29] found that a majority of physicians considered their knowledge of genetics to be adequate, but that a minority were confident to provide genetic counseling in simple genetic scenarios. Relatively few had actually made use of DNA diagnostic services, and there was relatively poor knowledge of what services were available. Menasha et al.[30] found that further education for physicians is required for them to accurately convey the risks and benefits of genetic testing to their patients. No similar studies conducted in Turkey were found in a review of the literature.

Table 3
Physicians' sources of information about genetic diseases and disorders

	Classroom		Clinical		No information		Classroom and clinical		
Disease/defect	N	%	N	%	N	%	N	%	Respondents N
Xeroderma pigmentosum	44	80.0	2	3.6	8	14.5	1	1.8	55
Turner syndrome	43	79.6	2	3.7	7	13.0	2	3.7	54
Galactosemia	43	79.6	2	3.7	6	11.1	3	5.6	54
Achondroplasia	43	78.2	1	1.8	8	14.5	3	5.5	55
Tay-Sachs disease	42	76.4	1	1.8	11	20.0	1	1.8	55
Hunter syndrome	42	76.4	1	1.8	12	21.8	—	—	55
Alcaptonuria	41	75.9	1	1.9	11	20.4	1	1.9	54
Hurler syndrome	41	74.5	1	1.8	13	23.6	—	—	55
Huntington disease	40	72.7	3	5.5	9	16.4	3	5.5	55
Porphyria	40	72.7	2	3.6	11	20.0	2	3.6	55
Klinefelter syndrome	39	72.2	1	1.9	8	14.8	6	11.1	54
Cri du chat syndrome	39	70.9	1	1.8	12	21.8	3	5.5	55
Patau syndrome	38	69.1	1	1.8	15	27.3	1	1.8	55
Edwards syndrome	37	68.5	2	3.7	14	25.9	1	1.9	54
Cystic fibrosis	37	67.3	2	3.6	7	12.7	9	16.4	55
Brachydactyly	37	67.3	2	3.6	10	18.2	6	10.9	55
Muscular dystrophy	36	66.7	2	3.7	7	13.0	9	16.7	54
Sickle cell anemia	36	66.7	4	7.4	—	—	—	—	54
Fabry disease	36	65.5	1	1.8	18	32.7	—	—	55
Osteogenesis imperfecta	35	66.0	2	3.8	9	17.0	7	13.2	53
Hemophilia A and B	35	63.6	4	7.3	2	3.6	14	25.5	55
Lesch-Nyhan syndrome	35	63.6	1	1.8	18	32.7	1	1.8	55
Spina bifida/anencephaly	33	60.0	2	3.6	2	3.6	18	32.7	55
Polydactyly	31	56.4	3	5.5	5	9.1	16	29.1	55
PKU	30	54.5	4	7.3	—	—	21	38.2	55
Albinism	29	52.7	2	3.6	4	7.3	20	36.4	55
Jacobs karyotype	29	52.7	1	1.8	25	45.5	—	—	55
Color-blindness	26	47.3	4	7.3	5	9.1	20	36.4	55
Cleft lip/palate	25	45.5	3	5.5	1	1.8	26	47.3	55
Rh factor	23	41.8	5	9.1	—	—	27	49.1	55
Cooley's anemia	22	40.0	4	7.3	—	—	29	52.7	55
Down syndrome	20	36.4	3	5.5	1	1.8	31	56.4	55
Diabetes mellitus	7	12.7	4	7.3	—	—	44	80.0	55

PKU, phenylketonuria.

Limitations of the study

Our response rate of 60% was below our expectations. Nevertheless, the number and distribution of our responses were sufficient for analysis. In Turkey, practicing physicians do not have the role of "gatekeeper" because of differences in the health care systems. Because there is inadequate knowledge about screening and counseling even in areas with abundant genetic services in Turkey, intervention at the primary care level is limited.

CONCLUSIONS

In this study, physicians who are actively giving genetic counseling recognize their knowledge deficits, and a large percentage request participation in an educational program. The prevention of genetic diseases before birth and the early diagnosis after birth could be a result of educational programs on this subject. This education could be provided by the Health Ministry in cooperation with uni-

Table 4
Physicians' answers related to genetic counseling

Topic	Yes		No		No information		Respondents *N*
	N	%	*N*	%	*N*	%	
Directing the parents of and couples with a risk for having a child affected by a genetic disease to an expert or a genetic counseling center	55	94.8	3	5.2	—	—	58
Recommending ultrasound during pregnancy	55	93.2	4	6.8	—	—	59
Recommending use of folic acid during pregnancy	53	91.4	5	8.6	—	—	58
Recommending maternal serum screening test for mothers 35 years and over	53	89.8	4	6.8	2	3.4	59
Knowing about some of the genetic counseling centers in Turkey	16	88.9	—	—	2	11.1	18
Encountering babies with congenital anomalies	47	87.0	6	11.1	1	1.9	54
Determining carriers of or individuals with inherited diseases such as β-thalassemia	49	86.0	3	5.3	5	8.8	57
Request participation in an educational course about genetic diseases and genetic counseling	47	83.9	9	16.1	—	—	56
Recording and informing individuals who have married a relative	42	76.4	6	10.9	7	12.7	55
Making recommendations to decrease the risk of diseases with genetic tendencies such as Type 2 diabetes	42	72.4	10	17.2	6	10.3	58
Routinely informing individuals about subjects such as risky pregnancies and marriage with relatives	41	71.9	13	22.8	3	5.3	57
Informing and recommending testing of carriers and next of kin	40	70.2	11	19.3	6	10.3	58
Recognizing genetic diseases common in the region	31	55.4	19	33.9	6	10.7	56
Giving genetic counseling about the maternal serum screening test	27	46.6	25	43.1	6	10.3	58
Knowing about the genetic counseling center in Denizli	15	27.3	8	14.5	32	58.2	55
Organizing a screening program for genetic diseases	13	22.8	39	68.4	5	8.8	57
Developing a family tree by learning the genetic history of individuals suspected of having a genetic disorder	12	21.1	43	75.4	2	3.5	57
Knowing the ethical regulations and techniques of genetic counseling	12	20.7	33	56.9	13	22.4	58

versity instructors and clinical geneticists. International cooperation on this subject could also be arranged. The WHO works with various nongovernmental organizations and collaborating centers that support implementation of genetics approaches to disease control in countries. In addition to the recognition of genetic services in the region and the country, consideration of medical genetics services and the application of their supporting genomic technologies should be an integral part of education in genetics at all levels. Genetic education needs to be provided not only to the primary care providers but also to the medical students who will soon join their ranks. Finally, the most efficient and effective methods for providing information and heightening awareness need to be determined through additional research.

ACKNOWLEDGMENTS

We thank all the physicians who participated in the survey. We also thank the Turkish Ministry of Health for giving us permission to use the questionnaire and make this work possible.

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Erratum

Miller TE, You L, Myerburg RJ, Benke PJ, Bishopric NH. Whole blood RNA offers a rapid, comprehensive approach to genetic diagnosis of cardiovasular diseases. *Genet Med* 2007;9(1):23–33.

Figures meant to be published in color in the above article were printed in black and white in error. For color versions of these figures, please vist www.geneticsinmedicine.org and open the PDF link of this article in January 2007 Table of Contents. We regret the error.