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Short Communication

THE ATTITUDE OF JAPANESE PHYSICIANS REGARDING GENETIC SERVICE FOR AUTOSOMAL DOMINANT POLYCYSTIC KIDNEY DISEASE (ADPKD)

Shinsuke NOMURA,* Shin-ichiro KAWAI, Tatsuo FUKUSHIMA, Hirotaka KATOH, and Gengo OSAWA

Nephrology Division, Department of Medicine, Kawasaki Medical School, Kurashiki 701-01, Japan

Summary A questionnaire carried out among Japanese physicians revealed that a strong demand for genetic counseling among patients and families with autosomal dominant polycystic kidney disease (ADPKD). In contrast, the awareness of physicians regarding genetic counseling for the disease seemed to be low. For example, 66.0% of the respondents to the questionnaire revealed a negative attitude to providing genetic counseling for patients, and 30.7% of the respondents did not know that most types of polycystic kidney disease are inherited disorders. With the advance of scientific research, the demand for genetic counseling among patients is bound to increase. Therefore, the providers of genetic counseling including the physicians are now pressed to improve their services.

Key Words genetic services, genetic counseling, bioethics, autosomal polycystic kidney disease

Autosomal dominant polycystic kidney disease (ADPKD) is one of the most common inherited diseases in man (Gabow, 1990). The defective gene for this disease was mapped on chromosome 16 (Reeders, 1985), and recently the ADPKD gene has been identified (The European Polycystic Kidney Disease Consortium, 1994) so that the linkage study within each family and presymptomatic or prenatal diagnosis have been established while the another candidate gene exist on chromosome 4 (Kimberling, 1993). Thesesci entific advances place upon physicians dealing with ADPKD patients the responsibility to provide accurate information and adequate counseling.

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* Reprint request should be addressed.

General practitioners stand in the forefront of clinical medicine and must always deal with inherited disorders. In Japan, however, it seems that general practitioners pay little attention to inherited diseases, especially, with regard to genetic counseling (Okura *et al.*, 1981). If this is the case, it is unlikely that the advantages resulting from the scientific advances would be of much benefit to ADPKD families.

This situation prompted us to evaluate the attitude of Japanese clinicians regarding genetic services by conducting a survey as to how they deal with ADPKD patients.

We acquired a list of 1,599 physicians in the fields of internal medicine, surgery, pediatrics, urology and obstetrics and gynecology from a register kept at the Okayama branch of the Japanese Medical Association (Nihon-Ishi-Kai in Japanese). Every third name was selected from the list, and these 533 physicians were sent a questionnaire, a covering letter explaining the purpose of the survey, and a stamped addressed envelope for return of the questionnaire. The respondents were not required to identify themselves. This study was carried out in November 1992. The respondents were asked about their professional status, the kind of institution they worked at, and 10 questions about their knowledge of and attitude to ADPKD and inherited disorders.

The completed questionnaire was returned by 265 physicians (49.7%) within two months. Most respondents were male (85.7%, 227/265). By speciality, 59.8% of them were internists, 11.7% were pediatricians, and 9.4% were obstetricians and/or gynecologists. Half of them (49.8%, 132/265) worked in their own private clinics.

In response to the question, "Are you aggressive about providing or interested in genetic services for not only ADPKD but also for other inherited diseases?," only 38 (14.3%) answered in the affirmative. One hundred and seventy-five (66.0%) answered in the negative, although they recognized the need for genetic services. The affirmativity for the genetic service among the internists (10.3%) was lower than that among the pediatricians (35.5%) and that among obstetricians and/or gynecologists (20.0%). Among the respondents, 77 (30.7%) did not know that most types of polycystic kidney diseases are inherited disorders (Table 1). The ratio of negative responses to the question increased as the age of the respondents increased (Table 1). One hundred and sixty (60.4%) had seen at least one patients with ADPKD. Of the physicians who did not know that these diseases are inherited disorders, 39.0% (30/77) had dealt with such patients before.

Asked what diagnostic terms they do or would use when seeing a patient with polycystic kidney disease, 69.9% of the respondents answered that they do or would use the term "polycystic kidney disease" (tahatsusei-noho-jin in Japanese) and do not or would not mention the mode of inheritance. Only 15.4% of them answered that they do or would attempt to differentiate the type of inheritance and

Table 1. Negative responses to the question "Are you aware that most polycystic kidney diseases are inherited disorders?"

Age of respondents	Negative responses	Total number	Ratio (%)
-39	5	49	10.2
40-49	15	62	24.2
59-59	17	52	32.7
60-	39	83	47.0
Unkwown	1	5	20.0
Total	77	251	30.7

to differentiate between autosomal recessive polycystic kidney disease and ADPKD.

Among the respondents, 33 (12.5%) had had at least one experience in giving genetic counseling. They had been asked some questions, such as the influence of the disease on the patient's children and on their marriages. Most questions were asked by the patients themselves, but some were by parents and spouses. Of the 33 physicians who had had the opportunity to provide genetic counseling before, two had not known that most polycystic kidney diseases are inherited disorders. Thirteen of the 33 (39.4%) including these two physicians indicated that they could not provide their clients with satisfactory answers, mostly due to their lack of genetical knowledge. When the physicians in this survey were asked about the best facility for genetic counseling, 77.7% answered that such counseling could be best carried out at specialized institutions, such as in the clinical departments of university hospitals, while 10.2% answered that the private clinics of general practitioners would be better. The answer recommending public centers (hoken-sho in Japanese) was quite low (1.1%).

Only 18.1% (48/265) knew that a defective gene of ADPKD had been mapped on Chromosome 16 and that a linkage study within each family had been reliably established. Most of them were under forty years of age. While 25.8% of pediatricians and 24.0% of obstetricians and/or gynecologists knew it, only 17.4% of internists knew it.

Thirty-six of the respondents (13.6%) had the clients who were now waiting for the latest information on the disease and advances in molecular genetics. Forty-seven physicians (17.7%) were against prenatal testing of the fetus, either because they did not consider ADPKD to be a critical disorder (30 physicians) or because they disapproved of artificial abortion for any reason (nine physicians). The other 138 respondents (52.1%) said that they would honor the opinion of patients and families who wished to have the intrauterine prenatal testing and artificial abortion if the tests were positive.

This study revealed a strong demand for genetic services in Japan among patients and families with ADPKD. One-eighth of the respondents had given

some genetic counseling for the disease, and many respondents had patients who should be immediately informed of the recent advances in molecular genetics.

When patients or their families in Japan visit physicians for genetic counseling, can these physicians handle their requests? Based on the results of our study, it appears they cannot. Over one-fourth of the respondents did not know that most polycystic kidney diseases are inherited disorders, and only one-tenth of them indicated willingness to provide genetic counseling. In addition, most physicians thought that their private clinics were not appropriate for genetic counseling. Not all physicians must do genetic counseling, of course. If they have insufficient knowledge, they should refer the patients to appropriate facilities where genetic counseling is available. The medical community in Japan needs to develop systematic genetic services, which would include the availability of genetic counseling at local facilities, but this is not happening.

In general, it is said that Japanese people like new things more than old things, for example, they have a great interest in new technology and frequently buy new-model cars. Another survey using a questionnaire revealed that a prenatal genetic screening system is supported by a higher percentage of the population in Japan than in any other country (Macaer, 1993). Our study also revealed that Japanese physicians are no exception. They are not reluctant to utilize new techniques resulting from the latest biochemical advances as tools to resolve clients' requests for help. It reflect the fact that artificial abortion may not be a controversial issue, as it is in some Western countries. The respondents said that there were many patients who needed genetic information. These attitudes of physicians and the Japanese population can lead to a situation in which prenatal genetic screening system would be easily carried out even in any facility. The medical community seems to be standing at a folk in the road in dealing with this issue. Action is required within the Japanese medical community itself or even nationally now so that Japan will take the right road leading to fuller and better management of inherited disorders and systematic genetic services.

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