Am J Hum Genet 36:677–689, 1984

Screening and Genetic Counseling for Beta-Thalassemia Trait in a Population Unselected for Interest: Comparison of Three Counseling Methods

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SUMMARY

We have assessed the effects of screening and genetic counseling for beta-thalassemia trait on knowledge, attitudes, and behavior in a prospective, controlled study of randomly selected adult members of a health maintenance organization. We report here that knowledge of manifestations and of inheritance of thalassemia, previously reported to be high immediately after counseling, were well maintained at 2 and 10 months following counseling. There was no detectable impairment of self-concept. Marital adjustment improved, and sexual activity increased significantly. Mood, assessed immediately before and after counseling, showed no undesirable changes. A patient-structured counseling method, designed to minimize negative psychological effects via discussion of feelings, was not superior to conventional and programmed methods, described in our previous reports, in terms of learning or attitude change.

INTRODUCTION

To what extent can an individual selected at random be expected to benefit from unsolicited identification as a carrier of a gene for a serious recessive disease?

Received October 6, 1983; revised January 19, 1984.

This study was supported by grant R01-HL-17465 from the National Institutes of Health and grants 146 and 664 from the New York State Health Research Council. This study was presented in part at an international symposium entitled "Thalassemia: Recent Advances in Detection and Treatment" held in Sardinia, Italy, June 7–11, 1981.

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In a prospective controlled study in the population of a health maintenance organization, we have assessed the effects of screening and genetic counseling for beta-thalassemia trait on an individual's knowledge and attitudes.

We previously reported that identification as a beta-thalassemia carrier in such a setting, when followed by carefully monitored genetic counseling, can result in significant overall learning without negative effects on mood [1]. Demographic, attitudinal, personality, or intellectual information elicited prior to counseling did not predict learning or mood assessed immediately following counseling [2]. Counseling provided by a trained physician (conventional counseling) and counseling provided by a videotape containing the same information followed by an opportunity to question a trained physician (programmed counseling) resulted in similar learning and comparable mood changes [3].

Our present report expands our previous reports in three ways. First, we compare a third counseling method, called "patient-structured," with the previous two methods. Second, we provide the results of follow-up evaluations performed 2 and 10 months after counseling utilizing all three methods. Finally, we present the impact of counseling on selected attitudes.

Patient-structured counseling involves letting clues derived from observation of the patient's behavior determine the sequence and character of the counseling session. Individuals seeking genetic counseling because of the diagnosis of a serious genetic disorder in the family commonly manifest shock, denial, anxiety, anger, guilt, depression, and impaired self-esteem [4–13]. A genetic diagnosis may disrupt a marriage [14, 15] and can impair sexual relations [16], but genetic counseling has been documented to reduce anxiety and depression [10, 17, 18]. In addressing the concerns that patients bring to the genetic counselor, the quality of the relationship established between counselor and counselee may be as important as the transfer of information [17]. In the past, genetic counseling has commonly employed a neutral educator model that neglects psychological factors and may be determined more by the needs and values of the counselor than those of the counselee [19]. A nationwide study of genetic counseling revealed that, while genetic counselors do well dealing with the medical concerns patients bring to counseling, they often do poorly dealing with psychosocial concerns [18].

Individuals identified by population screening as carriers of a gene for a serious recessive disease may also have undesirable emotional responses [20]. Even in a carefully organized Tay-Sachs screening program, nearly half the carriers and their spouses expressed some degree of shock, anger, anxiety, or sense of alienation or imperfection [21]. However, McCrae et al. [22] noted that, among parents of children with cystic fibrosis, guilt was much less evident among those couples to whom a physician had spelled out explicitly that it was inappropriate for them to feel guilty because of the randomness of genetic events.

Desiring to minimize adverse psychological effects of identification by genetic screening, we have added a third counseling method: patient-structured counseling. Patient-structured counseling is based on the hypothesis that greater individualization of counseling and more attention to psychological aspects may enhance desirable and reduce undesirable reactions. Developed by Engel and his colleagues [23], this counseling strategy permits the content and affective tone of counseling

to be continuously modulated according to the verbal and nonverbal responses of the patient. We hypothesized that, by using cues offered by the patient, the session might be made more cognitively and emotionally fulfilling and, consequently, undesirable effects of the experience might be reduced.

Follow-up is critical to the evaluation of the effects of genetic counseling because patient response is a dynamic process. Nevertheless, most studies evaluating genetic counseling report only a single assessment, either immediately after or at a variable interval after counseling [24]. In this study, we report data from before, immediately after, 2 months after, and 10 months after counseling, utilizing an uncounseled population to control for effects of the testing procedure and using objective assessment methods (questionnaires) rather than subjective reports.

METHODS

A flow diagram of the study is shown in figure 1. The details of most of the methods have been previously presented [1]. Briefly, the adult population (18–65 years) of the Genesee Valley Group Health Association, a health maintenance organization (HMO) in Rochester, New York, was screened for beta-thalassemia trait by testing blood samples drawn for any reason. Positive individuals were notified by mail of an "abnormal laboratory result" and asked to come in for an explanation. Prior to receiving further information, patients were asked to participate in a study of patient-physician communication and 92% consented. Subjects were randomly assigned on the basis of age, sex, marital status, and parental status to three counseling methods called programmed, conventional, and individual patient-oriented. Each method presented the same information, but in different ways. Just prior to counseling, the patient was asked to complete a symptom checklist, a medical-attitude and -experience questionnaire, a knowledge of probability questionnaire, a marital-adjustment scale, the Tennessee Self-concept Scale [25], and the Nowlis Mood Adjective Checklist [26]. A thalassemia-knowledge questionnaire tested knowledge of the manifestations of thalassemia. Finally, a genetics questionnaire tested knowledge of its inheritance.

Immediately following counseling, the mood adjective checklist and the thalassemia- and genetics-knowledge questionnaires were repeated and the patient was given the California Personality Inventory to complete later and mail to us. At 2 and 10 months after counseling, the patient was asked to return to determine what effects thalassemia-trait identification had had on his or her life and was asked to complete the medical-attitude and -experience questionnaire, thalassemia- and genetics-knowledge questionnaires, probability questionnaire, marital-adjustment scale, and symptom checklist. In addition, an IQ scale was given at 2 months and a self-concept scale at 10 months after counseling.

A control population was selected to establish that any changes occurring in counseled subjects were due to counseling and not to the psychological testing, the passage of time, or information from other sources. Control subjects were selected from subjects simultaneously screened from the same population but found not to have thalassemia trait. For the control subject, a film dealing with general health maintenance was substituted for counseling about thalassemia. Methods for evaluation were identical to those for counselees with thalassemia.

Counseling provided the various names for the condition, the characteristics of thalassemia trait, the characteristics of thalassemia major, and the expectations about offspring type by mates who were normal or had thalassemia trait. The content of counseling and a description of the programmed and conventional counseling methods has been presented [1]. In both cases, counseling was provided entirely on an individual, not on a group basis. In brief, the *programmed* counseling method employed a specially produced 40-min color-sound videotape showing the counseling of a young adult patient by a physician. Following viewing, a physician-counselor entered to answer questions. For the *conventional*

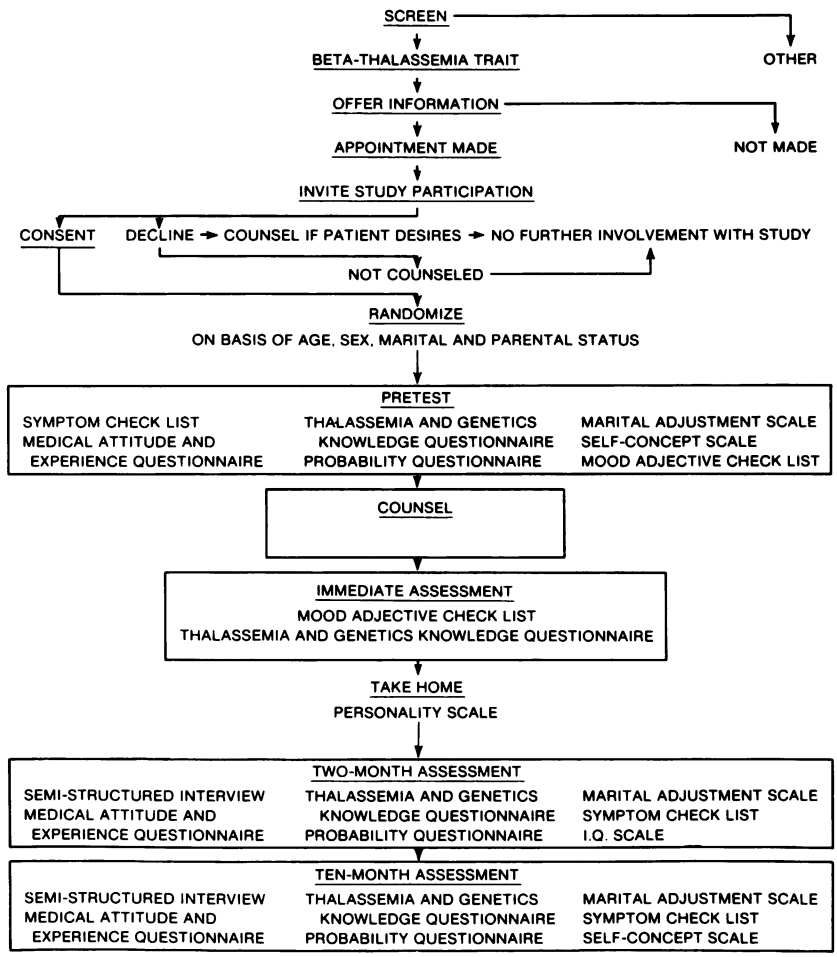

FIG. 1.—Flow diagram of project to evaluate screening and genetic counseling for beta-thalassemia trait.

counseling method, general internists were trained to present the same information as presented by the videotape. The third counseling method, referred to above, is called the *patient-structured* method. The counselors were internists trained via a 1- or 2-year fellowship in psychosocial medicine and nondirective counseling. The counselor attempted not only to teach but also to encourage expression of feelings and to assist with adjustment to the new information in light of the counselee's experience and values. This method is an adaptation of the interviewing method more fully described elsewhere [23]. The three counseling methods are compared in table 1.

RESULTS

Subject Groups

The counselees and control subjects in each group were very similar in demographic characteristics, as shown in table 2.

TABLE 1

COUNSELING METHODS COMPARED

	Programmed	Conventional	Patient-structured
Content	Same	Same	Same
Sequence	Fixed	Fixed	Determined by counselee
Dealing with facts/feelings	Facts only	Facts only	Facts and feelings
Interview style	Instructional videotape	Structured	Nondirective, open-ended
Discussion of alternatives	Structured but complete	Structured but complete	Determined by counselee

TABLE 2

DEMOGRAPHIC CHARACTERISTICS OF COUNSELEE AND CONTROL GROUPS

	PROGRAMMED		CONVENTIONAL		PATIENT-STRUCTURED	
	Counselees	Controls	Counselees	Controls	Counselees	Controls
No.	43	40	53	50	46	33
Age	38.79 ± 12.80	38.33 ± 12.74	35.60 ± 13.72	35.54 ± 13.27	31.37 ± 11.21	32.03 ± 10.68
Sex	M = 23; F = 20	M = 21; F = 19	M = 26; F = 27	M = 27; F = 23	M = 22; F = 24	M = 15; F = 18
Education*	4.83 ± 1.48	5.08 ± 1.31	4.40 ± 1.29	4.74 ± 1.19	4.33 ± 1.30	4.30 ± 1.40
Social class*	3.29 ± 0.90	3.08 ± 0.83	3.37 ± 0.88	3.40 ± 0.86	3.57 ± 0.94	3.06 ± 1.00
Marital status:						
Single	4	4	12	11	12	7
Married	36	33	38	37	25	19
Divorced	3	3	3	2	9	7
No. children	1.63 ± 1.50	2.20 ± 1.80	1.43 ± 1.68	1.78 ± 2.14	1.17 ± 1.34	1.15 ± 1.15
No. previously unaware of thalassemia trait status	24	. . .	39	. . .	29	. . .

NOTE: Shown are means ± SD.

* Social class was assessed by the Hollingshead and Redlich scale [28], which is based on occupation (7-point score, based on prestige) and on education (7-point score, based on no. of years), weighting the two (occupation weight 7, education weight 4), and conversion to a scale of 1.0 (lowest) to 5.0 (highest).

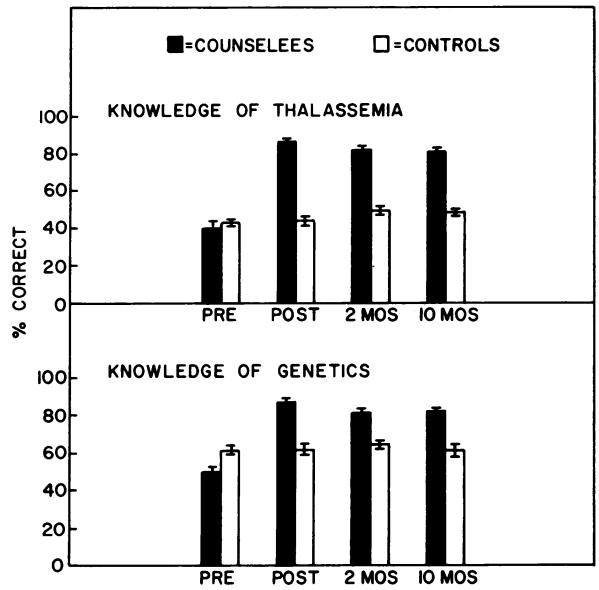

FIG. 2.—Effects of genetic counseling for beta-thalassemia trait on knowledge: comparison of counselees and control subjects. Shown are means ± SE of the mean for scores on questionnaires testing knowledge of manifestations and inheritance of thalassemia, respectively, as described [1].

Effects on Knowledge

Figure 2 shows the effect of counseling on knowledge of genetics and of thalassemia over time for all counseling methods combined. Compared with the controls, who do not show any significant change, there is, immediately after counseling, a striking increase in knowledge of both manifestations ($P < .01$) and inheritance ($P < .01$) of thalassemia, as shown by an analysis of variance. There is no statistically significant change in these levels at 2 months or at 10 months after counseling; knowledge is well retained.

All subjects identified as having beta-thalassemia trait were provided genetic counseling unless their physicians objected, regardless of whether or not they had been identified or counseled before. Figure 3 shows the effect on knowledge of prior awareness of having thalassemia trait. Not surprisingly, prior to counseling, previously aware individuals had more knowledge of both the manifestations and the inheritance of thalassemia than did previously unaware subjects ($P < .01$). Following counseling, however, the knowledge scores for the two groups were not significantly different.

Figure 4 presents knowledge scores for each of the three counseling groups. For each counseling method, there was a significant increase in both knowledge of thalassemia and knowledge of genetics immediately after counseling and no significant change between immediately after and 2 months after counseling or between 2 and 10 months after counseling. A Neuman-Keuls student's test of significance was used to assess differences in scores between counselees and

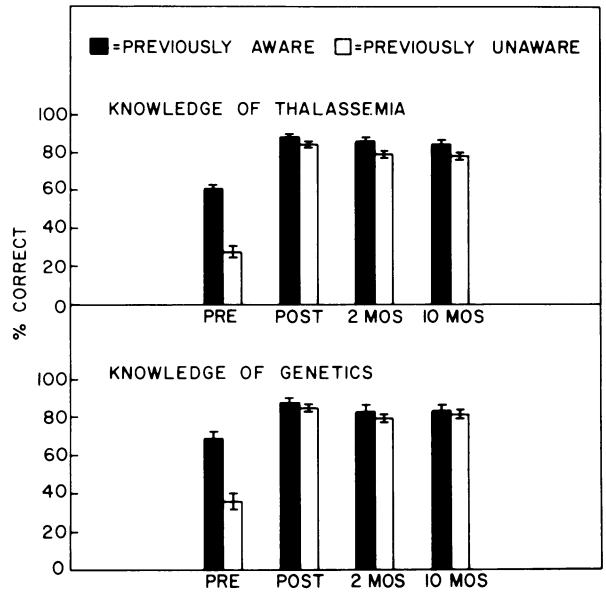

FIG. 3.—Effect of prior awareness of diagnosis of beta-thalassemia trait on knowledge. Shown are mean ± SE of the mean for scores on questionnaires testing knowledge of manifestations and inheritance of thalassemia, respectively, as described [1].

controls for each group. For both knowledge of thalassemia and knowledge of genetics, there was no significant difference between groups before counseling. However, at each time after counseling, counselees scored significantly higher than controls (not shown). There was no signficant difference among the three methods with regard to knowledge of either thalassemia or genetics at any of the four times. Thus, counselees and controls were relatively similar prior to counseling; counselees, regardless of method, displayed significant learning after counseling, relative to controls. This learning was retained over time across methods, relative to controls.

Effects on Mood

Table 3 shows the effect of genetic counseling on mood for counselees (all methods) vs. controls before and immediately after counseling. The increase in startle and the decreasse in deactivation (increased alertness) were expected because new information was provided. The increase in pleasantness and the decreases in skepticism and in anxiety may reflect satisfaction with the explanation of the "abnormal test result."

The most important finding, however, with regard to mood is the lack of change, particularly in those parameters in which some adverse change might have been anticipated. The lack of significant change in depression or aggression (anger),

coupled with decrease in anxiety, provides no evidence of an untoward effect of our unsolicited identification of the subject's thalassemia trait status on mood.

Some differences in mood were noted depending on whether subjects were or were not previously aware of having thalassemia trait. Subjects who were previously aware understandably manifested less startle ($P < .05$), as might be expected, but also more concentration ($P < .04$) and activation ($P < .05$) than did the group who were not previously aware. There were no significant differences among the three counseling groups in their mood scores before or immediately after counseling.

Effect on Attitudes

Table 4 shows the effects of counseling on various attitudes of counselees (all methods) vs. controls. Attitudes measured included self-concept, marital adjustment, and sexual attitudes. Self-concept did not significantly change. Marital adjustment showed a significant improvement across time. Sexual activity, reflecting sexual frequency and sexual satisfaction, also showed a significant increase across time. In each case, the statistical significance depends on a decrease in the controls as well as on an increase in the experimentals. Thus, the data on attitudes provide no evidence of adverse effects of identification and counseling. More important, there was no significant difference in attitude change among the three counseling groups (not shown).

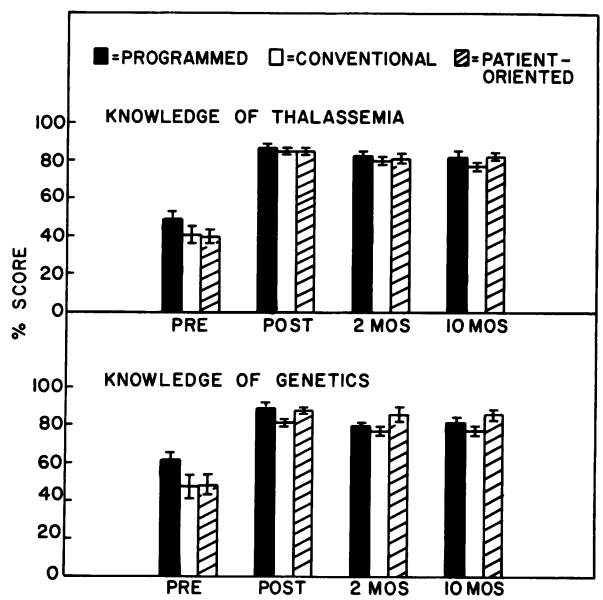

FIG. 4.—Effects of genetic counseling for beta-thalassemia trait on knowledge: comparison of three counseling methods. Shown are means ± SE of the mean for scores on questionnaires testing knowledge of manifestations and inheritance of thalassemia, respectively, as described [1].

TABLE 3

EFFECT OF GENETIC COUNSELING ON MOOD

	MEANS				
	COUNSELEES		CONTROLS		
	Before	After	Before	After	P*
Startle	2.70	12.11	1.00	1.17	.001
Deactivation	23.20	17.13	17.00	20.23	.002
Pleasantness	16.07	21.28	27.05	22.34	.002
Skepticism	22.00	6.35	11.94	6.30	.002
Anxiety	17.92	11.49	6.12	6.13	.01
Activation	26.61	21.65	46.46	36.67	NS†
Aggression	2.19	2.13	1.52	1.76	NS
Concentration	65.97	56.29	79.30	71.66	NS
Depression I	2.68	3.48	1.46	2.91	NS
Depression II	3.17	3.47	1.09	1.35	NS
Egotism	2.27	2.20	2.32	1.85	NS
Nonchalance	12.68	7.89	18.87	11.96	NS
Social affection	25.38	16.98	30.82	23.62	NS

NOTE: Mood was evaluated using the Nowlis Mood Adjective Checklist [26].

* For each scale, analysis of variance was used to assess the significance of the difference in scores before and after counseling for counselees vs. controls.

† NS = not significant.

DISCUSSION

This report adds information to our previously reported results in three respects: the results of a third counseling method; data concerning health, marital, and sexual attitudes; and evaluation at 2 and 10 months after counseling.

The third counseling method, the patient-structured method, although presenting the same information as the other two methods, differed in that the counselor was intensively trained in a medical psychiatric liaison fellowship and specifically

TABLE 4

EFFECTS OF COUNSELING ON ATTITUDES

	MEAN SCORES						
	COUNSELEES			CONTROLS			
PARAMETER	Before	2 mos	10 mos	Before	2 mos	10 mos	P*
Self-concept†:							
Total positive	48.38	· · ·	48.68	50.16	· · ·	50.01	NS‡
Total variability	45.84	· · ·	44.58	43.29	· · ·	42.76	NS‡
Marital adjustment§	66.26	68.00	69.89	72.03	69.53	66.87	< .01
Sexual activity§	53.01	52.46	59.45	58.32	58.52	54.01	< .01
Consistency of sexual responses§	34.85	35.52	39.15	51.16	49.26	45.42	NS
Abortion knowledge§	43.13	44.02	47.21	45.67	46.71	49.51	NS

* For each attitude, analysis of variance was used to assess the significance of change in scores across time for counselees vs. controls.

† Evaluated by the Tennessee Self-concept Scale [23].

‡ NS = not significant.

§ Each attitude was scored on the basis of multiple items from questionnaires designed for this study.

directed to elicit patient feelings and to help the patient make an adjustment to the new information. At the initiation of the study, we anticipated that unsolicited screening might evoke intense feelings since patients had not had an opportunity to select themselves out, as occurs with public genetic-screening programs. Aware of the possibility of negative effects, we therefore included a counseling method that was devised to minimize them.

No immediate negative mood effects were seen. The principal emotion reported was anxiety between the time the patient received the letter notifying him of "an abnormal laboratory result" and his visit to the center to learn its nature. The explanation generally relieved the anxiety. Using the counseling information specified, a psychiatrically trained physician did not surpass in this regard a physician without special psychiatric training, nor did counseling employing a carefully constructed videotape. A potential explanation for the lack of superior results with the patient-structured method, considering the literature on psychological reactions cited above and the generally positive mood data, relates to the setting in which the counseling occurred. Patients received all of their individual and family health care at this facility, and, by and large, most had come to trust the staff and to feel secure and supported in this setting. This sense of trust and familiarity may have provided sufficient influence to overcome the potentially adverse effects reported by others.

We had considered the possibility that a subject who learned that he was genetically different might experience a deterioration in sexual activity or marital adjustment. The measures used reflected no adverse effects in these areas, but instead, an unexpected and unexplained improvement in all three measures. From analyses to be reported later, we know that subjects generally told their spouses the result of their screenings. One might speculate that this information sparked a discussion of their marriages or their reproductive plans and that such discussion enhanced their relationships. An improvement in self-concept has been noted following proband-related genetic counseling [12, 27].

Finally, an encouraging aspect of these data is the finding that the information provided is well retained for the 10-month period of follow-up. We tried to enhance learning by presenting only essential information, relating new concepts to familiar ones, utilizing charts, and encouraging questions. The level of retention observed is particularly significant since the patient had not requested being tested for thalassemia.

Taken together, unsolicited screening and genetic counseling for thalassemia trait in a health maintenance organization can result in highly satisfactory learning and excellent retention without detected adverse effects on sexual behavior, marital adjustment, or self-concept.

ACKNOWLEDGMENTS

We thank Dr. James S. Roberts, Medical Director of the Joseph C. Wilson Health Center, GVGHA; physician-counselors Michael Buck, Richard Morris, Dennis Novak, John Robbins, Michael Udkow, and Porter Welbourne; and the entire medical and ancillary staff for their generous cooperation. We thank Dr. Paul Grover, previously of the Division of Medical Education, and Dr. Sanford Meyerowitz (deceased), Departments of Psychiatry

and Medicine, for wise counsel; and Starlene Dick, project coordinator, and Judith Lee and Carol J. Sutera, research assistants, contributed immeasurably.

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