
The Evaluation of Genetic Counseling

A Committee Report

A COMMITTEE of human geneticists and behavioral scientists met in Colorado Springs, Colo., on February 28 and March 1, 1975, to consider appropriate methods for evaluating genetic counseling. The meeting was requested by the National Institute of General Medical Sciences of the National Institutes of Health.

The committee recognized the need for a definition of genetic counseling and adopted the definition published in 1975 by the Ad Hoc Committee on Genetic Counseling of the American Society of Human Genetics (1):

Genetic counseling is a communication process which deals with the human problems associated with the occurrence, or the risk of occurrence, of a genetic disorder in a family. This process involves an attempt by one or more appropriately trained persons to help the individual or family (1) comprehend

the medical facts, including the diagnosis, the probable course of the disorder, and the available management; (2) appreciate the way heredity contributes to this disorder, and the risk of recurrence in specified relatives; (3) understand the options for dealing with the risk of recurrence; (4) choose the course of action which seems appropriate to them in view of their risk and their family goals and act in accordance with that decision; and (5) make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder.

This definition, in a slightly modified form, was published in 1974 together with a discussion of the state of the art of genetic counseling (2).

The following concepts were formulated as guidelines for groups who wish to evaluate the outcomes of their counseling methods, whether proband or non-proband oriented.

Evaluations

Most published evaluations of genetic counseling entail recontacting persons previously counseled in order to assess retention of information and attitudinal and behavioral changes as a result of counseling. This type of retrospective evaluation is often inadequate because of inaccurate records of the persons' precounseling knowledge, attitudes, and beliefs and insufficient data on the

exact transmission of messages by the counselor. These pitfalls in retrospective evaluation suggest that prospective evaluations are preferable whenever possible. Nonetheless, in certain situations retrospective studies may be useful, for example, as baseline information for generating designs for prospective studies. Additionally, ethical considerations may sometimes preclude the development of meaningful prospective studies involving, for example, a comparison group which did not receive counseling.

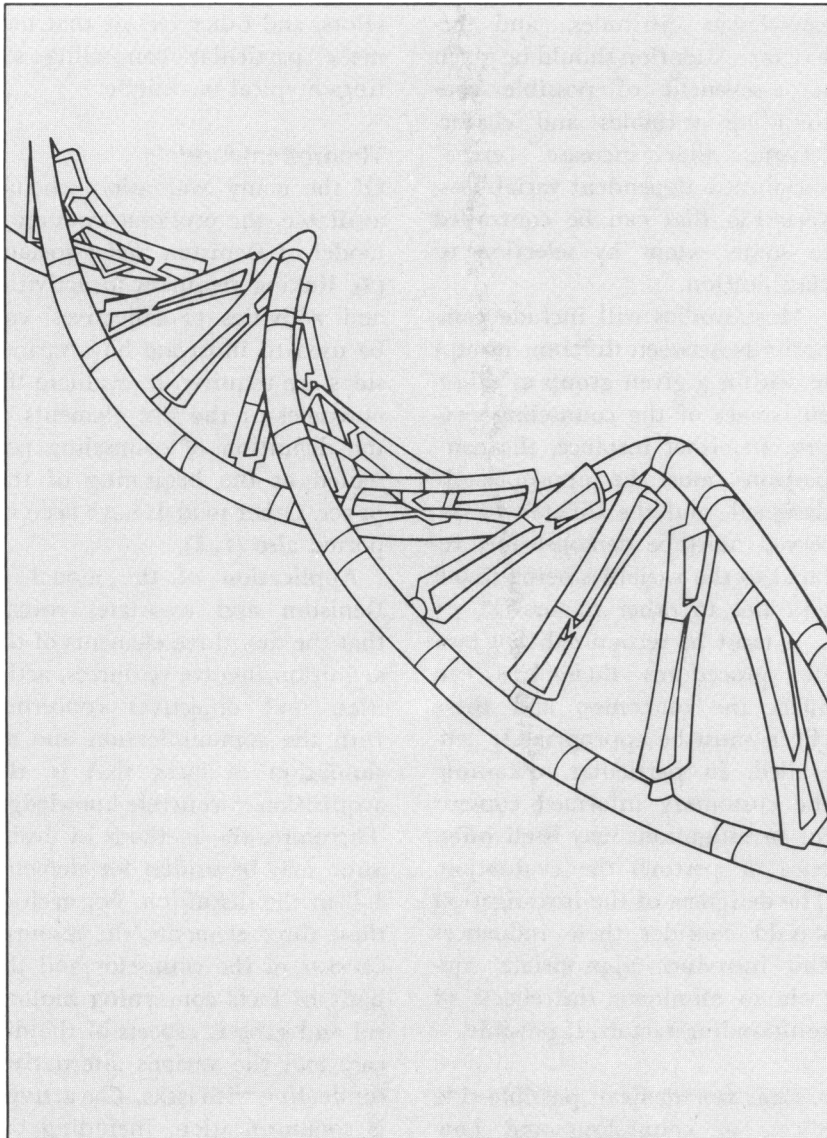
In sum, the investigator should justify the use of retrospective instead of prospective studies. He should acknowledge and deal with the limitations of experimental design imposed by a retrospective study.

Study Plans

In general, the following six areas should be covered in study plans for evaluating either retrospective or prospective counseling.

A clear statement of objectives of the study. All proposed studies must include a clear statement of study objectives. The objectives should be based on verifiable, testable hypotheses or aimed at measuring significant parameters.

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These objectives should be stated in terms susceptible to valid forms of measurement.

A clear statement of the methods used to counsel and how they will be measured. This statement should include the content of the information to be transmitted, the identification and qualifications of the counselor, the philosophical approach to counseling (for example, directive, participatory, or nondirective), the interventions (if planned), and the time schedules for the precounseling, counseling, followup counseling, and postcounseling evaluation sessions.

As a general principle, it is preferable that the evaluator of the counseling process not be a member of the counseling team, although both evaluator and counselors should participate in the design of the investigation. The evaluator should have demonstrated competence in the area of behavioral science required for the purposes of the study (for example, decision-making models, ego-adaptation models, transactional models, and measurement of attitudes). The relationship between the evaluator and the person or team doing the counseling must be defined, and potential

conflicts of interest and sources of bias introduced by this relationship must be identified.

A clear statement of the means by which each aspect of counseling will be evaluated. This statement includes the characteristics and size of the sample, the choice of instruments, the statistical techniques, and the temporal aspects of the design. Consideration should be given to the appropriate use of factorial designs using multiple analysis of variance with an explicit statement of appropriate sample size for a given expected magnitude of effects.

With regard to selecting the sample, its size will depend on the key variables in the study, the levels of measurement to be used, the analytic techniques to be used, and the levels of statistical significance desired. The sample chosen should be one that will give the best information on the specific questions being posed. In some instances, this might necessitate the choice of persons counseled for a single disorder, but in others the sample might better be composed of persons counseled for several specific disorders or even for a large variety of conditions. In all instances, it is necessary to define precisely the population to be studied. Further, the chosen sample should be randomly drawn from the population of reference, or, if it is necessarily unrepresentative, the biases of selection should be appropriately handled. The process by which the sample is chosen should be consonant with sampling theory.

The measuring instruments employed in the study must have demonstrated validity and reliability. Consultation by psychometricians may be sought, if needed. Internal and external threats to the validity of the data

should be identified. The data-gathering process should be structured with collaboration of both the evaluator and counseling team so that the data will be accumulated in a maximally useful form with a minimum of subsequent translation.

A clear statement of the variables or biases which might influence the results and how these will be handled. Explicit consideration should be given to numerous independent variables (such as sex, age, training of counselor, and counseling setting and methods) and dependent variables (such as knowledge, attitudes, and behavior). Attention should be given to assessment of possible confounding variables and characteristics that increase "error" variance in dependent variables—variation that can be controlled to some extent by selection or classification.

Most studies will include comparisons between different groups or within a given group at different stages of the counseling process. In either instance, the comparisons must be appropriately designed, and the differences observed must be demonstrably related to the variables being tested and not to other factors.

It must be recognized that testing procedures themselves can affect the outcomes, and these effects must be appropriately controlled. In particular, obtaining the customary informed consent for investigations may itself influence or perturb the evaluation. The designers of the investigation should consider these influences and introduce appropriate controls to eliminate the effects of confounding factors, if possible.

A clear statement of possible side effects of counseling and how

these will be measured and handled. Assessments should be made of possible negative side effects, such as generation of undue anxiety. Also, beneficial side effects—such as family planning alternatives for those couples who intend to have more children but wish to space them—should be noted.

A clear statement of the extent to which the results may be generalized. Evaluators should consider the limits of generalizability of their results in the light of the size and representativeness of their study sample, the number of counselors, and other factors that may make particular counseling settings atypical or unique.

Theoretical Models

Of the many evaluation schemes available, the program evaluation model of Deniston and associates (3), linking resources to activities and activities to objectives, can be used to illustrate how various steps are required to evaluate the outcomes of the five elements of the definition of counseling presented at the beginning of this paper. Other models have been reported also (4, 5).

Application of the model of Deniston and associates reveals that the first three elements of the definition involve resources, activities, and objectives concerned with the communication and assimilation of facts, that is, the acquisition of concrete knowledge. Therefore, the methods of evaluation may be similar for elements 1–3 in the definition. For each of these three elements, the resource consists of the counselor and the body of facts concerning biological and genetic aspects of the disease and the various alternatives for dealing with risks. The activity is communication, including the

various counseling methods and media, and can be evaluated by independent experts outside the counseling process; for example, to what extent and how skillfully the person counseled was exposed to the necessary information. The objective is seen as the acquisition of knowledge and understanding, and it can be measured by scores on tests of comprehension.

With regard to element 2 of the definition, the special problem of distinguishing knowledge of the precise mathematical risk of recurrence in specified relatives from its subjective meaning for the counselee should be recognized. It seems likely that the counselee's interpretation of the risk is more motivating than the actual mathematical risk.

Elements 4 and 5 in the definition relate the acquired knowledge to the complex process of individual or family decision making and can be anticipated a priori to be difficult to evaluate. The difficulties lie in the less reliably measurable factors of "family goals," "adjustment," and especially the hazards of ascertaining what courses of action would be "appropriate" in a specific family setting. Personal goals, adjustment, and appropriateness cannot be expected to be easily defined either by the family or by the counselor. Moreover, even if appropriateness could be judged at a particular time, changes could occur in perceived appropriateness; for example, the decision not to have a child could change over time.

It is possible, however, to assess the relative values, for the individual person, of a series of issues related to desired family size, to selection of a mate, to the possibility of, or willingness to have, a child with a genetic defect. It is also possible to assess the acceptability of various courses of

action for dealing with the problem. Judges could rate the extent to which decisions reflect a reasonable relationship to the values and means of the person counseled.

Thus, the real possibility exists that success with regard to attainment of elements 4 and 5 of the definition cannot be evaluated. As a family tries to balance old attitudes with new knowledge, for example, a dilemma could arise leading to paralysis of action. The inability to measure the confounding variables of coping mechanisms was acknowledged. Perhaps the definition of element 4 could be changed to read ". . . in accord with their knowledge and consistent with their value system."

Despite the problems posed, possible approaches that could be useful in evaluating elements 4 and 5 are as follows.

- The definition and use of appropriate control or comparison groups:

1. To assess the effects of pre-testing, half of the prospective counselees could be pretested and then counseled, and the other half could be counseled only. Then, both groups could be tested. Even here, however, emotional problems existing before counseling may confound the evaluation of initial knowledge, attitudes, and behavior.

2. Counselees could be tested for changes in attitudes toward alternatives for dealing with identified risks.

3. Persons not responding to the initial invitation for counseling could be followed up to measure any changes occurring in the absence of counseling.

- The measurement of frequency or change of actual or intended attitudes or behavior.
- The correlation of results or

acquired knowledge (objectives 1-3) with consequent attitudes and behavior.

Single or Multiple Centers

No blanket statement can be made regarding the relative merits of studies at single centers versus multiple centers. The alternative chosen must be justified on the basis of the objectives of the study. In general, a question that could be answered by evaluation of counseling at a single center should not be investigated by a multicenter study. However, if a multicenter study is required for the accumulation of larger amounts of data, such as in cases of rare genetic diseases, it must be demonstrated that the counseling processes at the different centers are comparable and that effective cooperation exists between the participating centers and the evaluators.

Recommendation

Because the guidelines outlined here deal with both genetic and behavioral matters, we suggest that a review process for the research proposal should include evaluation by both geneticists and behavioral scientists.

Authors' note: Names and affiliations of members of the Committee on Genetic Counseling are available from the authors.

References

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