Experiences of Public Health Nurses in Obtaining Family Pedigrees

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THE FAMILY PEDIGREE as a systematic and productive method of gathering family health histories appears well suited to public health use whether for casefinding and treatment purposes or for epidemiologic research. The family pedigree is a tool with which public health workers have had little experience. Therefore, the methodology used and the experiences of public health nurses with this tool may be helpful to others contemplating an activity which uses a genetic approach.

The impetus to initiate a program based on family pedigrees in the Contra Costa County (Calif.) Health Department began with the finding that the majority of the county's crippled children caseload represents conditions in which measurable hereditary factors have been described in the literature. In the area of chronic disease, the contribution of heredity appeared impressive. So did the opportunities for casefinding, preventive treatment, and research that are offered by the genetic approach. Moreover, other than a few projects concerned mainly with specific rare diseases, there appeared to be little or no genetically oriented effort, either service or research, being carried on by other public health agencies.

Plans for the Contra Costa program were first described in a paper presented at the 1962 meeting of the American Public Health Association, excerpts of which appeared in the *Journal of the American Medical Association* (*1*) and *Public Health Reports* (*2*). The program went into actual operation in May 1963, following two orientation sessions on pedigree-taking for the health department's 45 nurses. Preliminary results of this program were included in a paper given at the American Public Health Association meeting in October 1964.

Briefly, the program objectives are to provide genetic counseling or information to health department clients through their family physicians (or through a health department physician if the family does not have one), opportunities for early casefinding and prophylactic treatment among these families, and a collection of family pedigrees as a basis for research activities.

Pregnancy histories of certain women (household cases, mothers of household cases) are taken, and a note is made of which family members have been examined for the same disorder or disorders.

Pedigrees are taken by the nurses on families with chronic or congenital disorders or defects who are in or are entering the nursing caseload. In taking the pedigree, the nurse asks the family informant about the health status of each relative as he is listed, so that all health problems are recorded on the pedigree. (Instructions for taking pedigrees appear on p. 45 and the symbols used are shown in figure 1.)

Completed pedigrees are sent to the health department physician in charge of the program for an interpretation based on what the pedigree and the pertinent literature suggest. The interpretation consists of whatever general genetic information is available on the stated dis-

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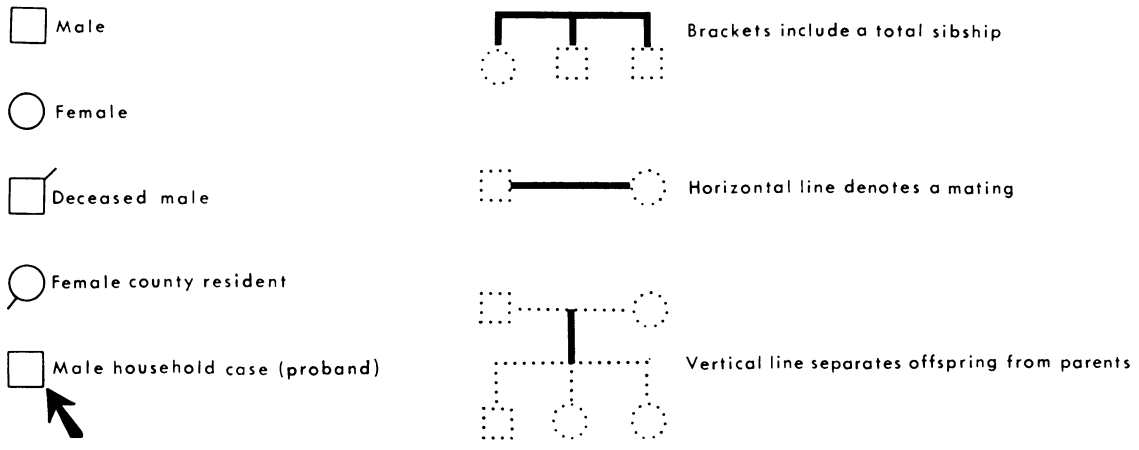
Figure 1. Symbols used in taking family pedigrees



order in the way of usual mode of transmission, risk to relatives and future offspring, methods of identifying susceptibles or carriers of the genetic trait, and availability of prophylactic measures. It includes an assessment of the given pedigree, stating risks where possible as they pertain to specified individuals on the pedigree. A typical pedigree and its interpretation are shown in figure 2.

Copies of the pedigree and interpretation are supplied to the family physician for his use in counseling if the family requests it and to the nurse for her information and reinforcement of the physician's counsel.

By May 1964, when 250 pedigrees had been taken, the nurses had volunteered a sufficient number of interesting experiences in taking pedigrees to justify a closer look at this particular aspect. The results of this are described below. The response of family physicians as well as other project findings will be presented in a future report.

Because spontaneity of expression was desired, an unstructured questionnaire was used in which each nurse was asked to describe her experiences in taking family pedigrees, the observed reactions of the families she approached, and her reactions to the pedigree interpretations she had received. While all information obtained was essentially volunteered and therefore difficult to quantitate, certain predominating responses emerge. Replies to questions asked of about one-third of the nurses individually, after they completed the questionnaire, suggested these responses were shared by a majority of the nurses.

Experiences and Reactions of Nurses

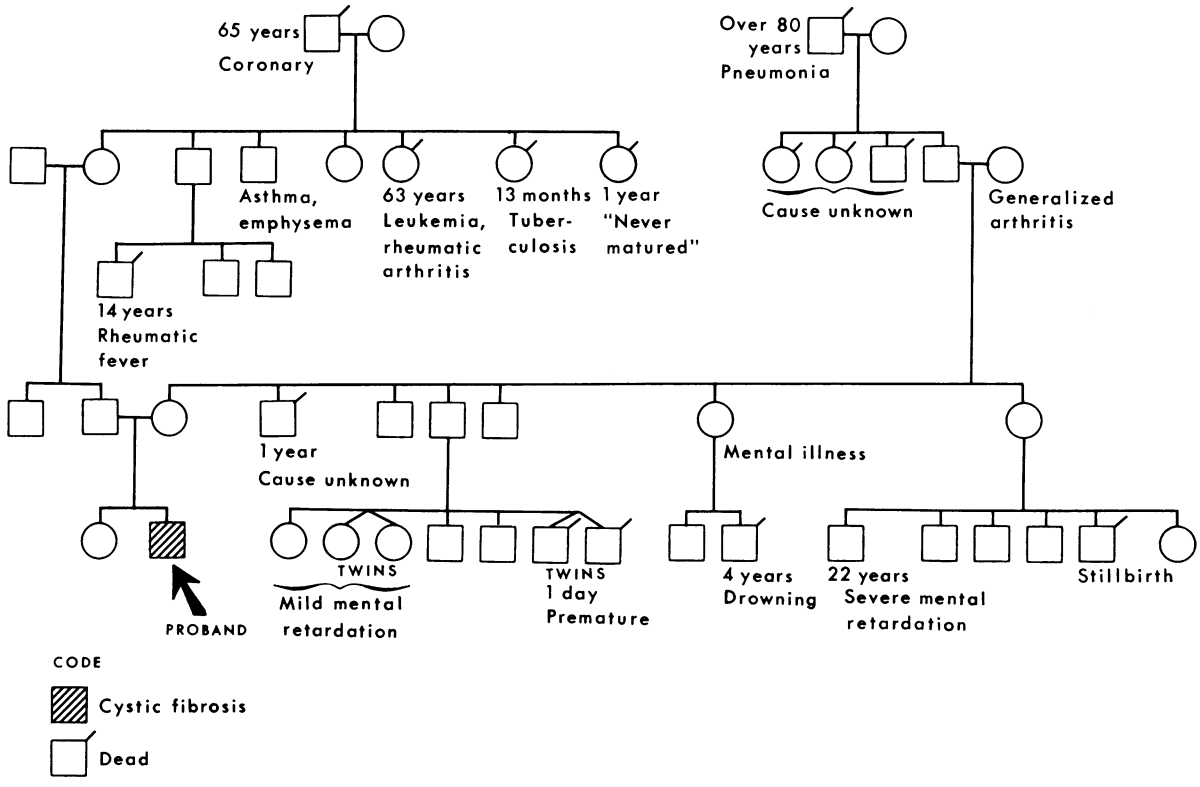
Although the proposed activity required mastery of a new technique as well as an added workload, general nurse response has been enthusiastic. Previous emphasis in the department on the significance of genetics in so many of the conditions with which nurses deal undoubtedly helped to trigger this enthusiasm as did the anticipation among nurses of "a chance to explore something new in working with families."

In the first few weeks two or three nurses admitted to some anxiety that the emphasis on heredity might alarm families, but they reported that this anxiety tended to disappear as they gained experience and as the favorable reaction of families became evident.

Eleven kinds of response were obtained from the questionnaire. Six of these were volunteered a total of only seven times. Each of the remaining five responses was volunteered by 20 percent or more of the 45 nurses. These five, described below, were therefore considered major responses.

- Obtaining a pedigree helps clarify not only the family health picture but family relationships, including social structure and behavior . . . "increases my awareness of the family's ability to recognize, evaluate, and accept their problem" . . . "gives insight into why a family lives and acts the way they do and how

Figure 2. Typical family pedigree and its interpretation



INTERPRETATION

This interpretation is based on the information given by the family, and its validity consequently depends on the validity of the information given.

Diagnosis: Cystic fibrosis (confirmed through crippled children's program).

General Genetic Information

Cystic fibrosis appears to be due to a pair of recessive genes. Presence of the disease, therefore, depends on the inheritance of the abnormal gene from both parents who, therefore, must both be carriers of the abnormal gene. For such carrier couples the probability of an infant with cystic fibrosis with each pregnancy is 1 in 4, of a carrier infant 1 in 2, and of a completely normal infant 1 in 4.

Variations in the rapidity of the clinical course and in the predominating symptoms (pancreatic vs. pulmonary) suggest the modifying effect of other genes or of the environment.

Effect of one gene (carrier state) may not yet be fully appreciated.

Both sexes are equally affected. Whites have a greater incidence than nonwhites. About 1 in 33 persons is estimated to be a carrier. As yet, there is no reliable test for carrier detection.

Assessment of the given family: Because of the recessive nature of the gene, the presence of an isolated case in a family is not unusual. The risk of cystic fibrosis in every future offspring these parents may have is 25 percent. Having already had a fibrotic infant does not decrease or increase this risk for any subsequent pregnancy. The patient's normal sister may be either completely normal (⅓ chance) or a carrier like her parents (⅔ chance). The chance of cystic fibrosis, therefore, among each of her own offspring someday appears to be around 1 in 200 (her ⅔ chance of being a carrier × her 1/33 chance of marrying a carrier × her ¼ chance of having a child with cystic fibrosis with each pregnancy if she is a carrier and if she does marry one).

Significance: The information on risks may have value to carrier couples in planning for the future. Knowing which newborns are at risk of having cystic fibrosis is important for early diagnosis and early institution of therapy.

Additional Comments

The familial picture of mental retardation is interesting and raises the question of whether or not a genetic factor for retardation may also exist in some of the members of this family. Lacking an underlying diagnosis, this, of course, is only speculation.

one needs to approach them in order to help them with their problems."

• The technique improves the nurse's rapport with the family . . . "enhances the relationship because of the total family concept" . . . "families who had done pedigree studies with the previous nurse in the district seemed extremely well informed and had a very strong positive feeling for the nurse, which in most cases was transferred to the next nurse, myself" . . . "makes families feel I really care, not about just one of them but about all of them."

• The technique provides opportunities for health education . . . "is a marvelous teaching tool" . . . "in obtaining the pedigree one is always able to do health teaching, including correcting people's misunderstanding of diseases" . . . "elicits health problems in the family the nurse hadn't been aware of."

• Taking a pedigree is time consuming, averaging 1 to 1½ hours, but it is a fertile source of information and may, in some instances, actually save time . . . "took quite a bit of time but time was well spent. Opened up areas that would otherwise not have been uncovered" . . . "time consuming but worth it if family is interested" . . . "learned in one afternoon's visit what would ordinarily have taken me a year of visits to uncover."

• Even when a pedigree is not being taken there is useful carryover of the technique . . . "improved my interviewing skill in general" . . . "has helped me to develop good interviewing habits even in situations where I'm not actually taking a pedigree."

Of the 45 nurses, only 3 have actually disliked taking pedigrees. The remaining 42 have had sufficiently pleasant and profitable experiences to consider the technique a valuable nursing tool if not for "all or most families" at least for the "cooperative" families who are "willing to follow-through."

Family Reaction

The majority of families have reacted favorably. Only two or three families have refused to participate. The following family reactions were most commonly noted by the nurses.

• Amazement and interest that such a project is being done . . . "all my families have felt that this study is something quite amazing." . . . "The reaction of families has been a mixture of surprise and amazement that medical science has progressed to the point where hereditary disease can be studied and help given."

• Eagerness to contribute to research. "Families have expressed a good deal of interest in participating in a research project" . . . "being part of a research project frequently seems to please people."

• Active cooperation, occasionally to the extent of writing letters, making long-distance telephone calls, and holding family gatherings. . . . "interested and conscientious in collecting information" . . . (the process) "has helped to draw families together in a common search for information" . . . "alerts them to the importance of trying to find out about their family's health history and the significance of health problems in the family."

• Less interest, especially regarding health aspects, among families with economic or marital problems and with educational or language handicaps . . . "In my caseload taking pedigrees is difficult. There is much illegitimacy among these families, and they are therefore disinterested in pedigrees. Culture and educational background also enter the picture" . . . "Those in the lowest socioeconomic group were not always interested in it from a health point of view but did enjoy talking about their families . . . liked seeing their families organized as a family tree." While families with economic problems have often been less impressed with the health implications of the process in contrast, for example, to "many middle-class families in the crippled children caseload" and occasionally, therefore, a point of frustration with the nurse, some of the most detailed pedigrees have come from the lower socioeconomic groups with large families, extended family groups nearby, and closely maintained ties.

• Most families are interested in any feedback of information that might be forthcoming from the study of their own and similar pedigrees and request that any such information be shared with them.

Instructions for Taking Pedigrees

Persons Included

1. Begin pedigree with the household case (identified by an arrow), then include as a minimum the following living or dead relatives of the case, in this order: siblings, parents, father's siblings, all descendants of father's siblings, father's parents, mother's siblings, all descendants of mother's siblings, mother's parents, and all descendants of the case and his siblings.

2. If another member in the household has another qualifying disorder, extend the pedigree to include the relatives listed above to this member or, if easier, make a separate pedigree.

3. Include parents of all persons in the oldest generation shown who have the same affliction as the household case.

4. Do not include spouses of case's relatives unless relatives' offspring are afflicted.

5. Include stillborn relatives.

Information Required

1. As each relative, living or dead, is added to the pedigree, inquire of the informant whether or not this relative has or had (*a*) the same affliction as the case and (*b*) any other chronic disorder or defect. Asking this for one person at a time ("does he have . . .") is often more productive than inquiring for a whole sibship at a time ("do any of these . . . ").

2. All disorders noted on the pedigree, whether or not the same as the case's, should be as specific as possible. For example, qualify the site and nature of congenital anomalies, the basis of acquired heart lesions (rheumatic), primary site of malignancies (breast, stomach), and type of arthritis (rheumatic, gouty).

3. When asking about the health status of each person, keep in mind that a condition may be present in less severe form in some persons; for example, as might be suggested by a higher than usual incidence of "kidney infection" among adult relatives of a child with obvious and severe anomalies of the urinary tract.

4. Distinguish, if possible, between identical and fraternal twins.

5. Enter the current age of all afflicted persons.

6. For nonliving persons, record where possible the age (or approximate age) at death and the cause.

7. Ask routinely and note on pedigree if there is any known consanguinity among forebears.

8. Ask routinely if the household case is colorblind; if so, ask this for all others in the pedigree.

9. If convenient, symbols may be used to designate different or partial disorders. Enter your code on the pedigree sheet if symbols are used.

10. Ask if disorder in question is or was present in more distant relatives than those mentioned. If so, identify these relatives on the pedigree.

• Many families have anxieties about hereditary factors, and their participation in the construction of a family pedigree often serves as an opportunity to air these anxieties. . . . "There is evidence that hereditary disease is a cause of much concern to families" . . . "most (of my) families have had an air of relief" . . . "for some it's been a relief to express their concerns and to have someone really work out the facts."

Response to Pedigree Interpretations

The value of the pedigree interpretation in increasing their store of information was expressed by nearly all nurses. This material, it was noted by nursing supervisors, is being "filed for future use so that all nurses in each office have access to each pedigree and its interpretation."

The initial disappointment of two or three nurses at the lack of definite conclusions that can be drawn from certain pedigrees portraying disorders of obscure genetics has apparently been alleviated by the explanation that it is from the study of such pedigrees that more conclusive answers may ultimately come.

Interpretations have also been noted to affect subsequent pedigree-taking in a striking if not surprising way. Receipt of interpreted pedigrees by nurses results in an immediate increase in pedigree submissions. Likewise, a lag in interpretation reduces the number of pedigrees submitted. Nurses feel strongly that pedigree interpretations should be done without too much delay. Interested families want "prompt answers."

Administrative Viewpoint

In the opinion of the nursing administrative staff, "nurse participation in obtaining pedigree histories has been a valuable expansion of nursing service to families." As summarized by the director of nursing services, "Two aspects merit special mention: First, adequate preparation for genetic counseling has not, in most instances, been part of public health nurse instruction. The pedigree approach has increased the nurse's knowledge and awareness of inherited characteristics so that she can answer more effectively queries made by families about hereditary fac-

tors in disability and disease. These questions are of major concern to many families in the nurse's caseload. Her ability to provide reliable scientific information greatly enhances her helpfulness to these families.

"Second, the approach has the potential of increasing family physician-nurse teamwork in meeting family needs. Skilled pedigree interpretations available to physician and nurse at the point of family interest and active participation promote a coordinated physician-nurse approach to family counseling."

Summary and Conclusions

The predominant reaction of nurses and families to the process of taking family pedigrees has been enthusiastic. They both find pedigrees interesting, useful, and educational. Aside from its use to accomplish the objectives of the local public health program in genetics, pedigree-taking is commonly found by nurses to increase their understanding of family relationships, improve their rapport with the family, provide new opportunities for health education, and improve their general interviewing skills.

Common family reactions include appreciation of nurse attention to total family, eagerness to participate in research, active cooperation in obtaining further family information, desire for feedback information, and use of the process as an opportunity to air anxieties about heredity. From a nursing administrative standpoint, the pedigree process directly enhances the nurse's helpfulness to families and leads to a more coordinated physician-nurse approach to family counseling.

REFERENCES

(*1*) "Family pedigree" will be used to help congenitally disabled. JAMA 182: 36, Nov. 24, 1962.
(*2*) Health department plans genetic counseling. Public Health Rep 78: 118, February 1963.

Physical Exercise for Heart Patients

A research project at the Donolo Institute of Physiological Hygiene in Jaffa, Israel, backed by the Vocational Rehabilitation Administration, Department of Health, Education, and Welfare, seeks to get heart patients back on the job by having them engage in physical exercise.

Daniel Brunner, M.D., its director, says that surveys in Israel have indicated that incidence of heart attacks in sedentary workers was three times greater than among nonsedentary groups subjected to similar diets and living conditions on an Israeli kibbutz, or communal farm. Followup studies, he says, show that ex-patients in hard-labor jobs had a greater survival rate than those who were sedentary after their attacks.

Researchers in the present project are doing larger-scale followup to buttress earlier findings. In addition, the project is developing a course of gradual physical training for about 100 heart patients, first with exercises and subsequently with sports.

Mary E. Switzer, U.S. Commissioner of Vocational Rehabilitation, comments that "This project, like many others the Vocational Rehabilitation Administration is backing in nine foreign countries, is a good example of how VRA can work to benefit the people of foreign lands, while, at the same time, gaining insight into problems which are of the utmost importance in rehabilitation work here at home."