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PSYCHOSOCIAL ASPECTS OF GENETIC COUNSELING

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HEMOPHILIA AND THE USE OF GENETIC COUNSELING AND CARRIER TESTING WITHIN FAMILY NETWORKS

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INTRODUCTION

s paper discusses the diffusion of genetic counseling and carrier testing among potential clients, and the influence of the social network on this diffusion. The personal social network is the whole of relations an individual has. These may be relations with kin, with friends, neighbors or colleagues.

Social networks are associated with the diffusion of medical or other technologies in more than one way:

1)interpersonal communications within the network provide the individual with **information** on new technologies that he might otherwise have missed.

2)the knowledge that others in the network have considered the technology or made use of it provides the individual with **legitimation** and **support** to make use of it also.

3)the individual may be exposed to deliberate **influence attempts** concerning acceptance or disapproval of the technology. (Becker, 1970)

This summarizes the influence of social networks on the use of new technologies in an elegant way. The different stages in the decision to make use of a technology are distinguished: 1) being aware of its existence, 2) forming a positive or negative attitude toward the technology, and 3) making use of it. Furthermore the influence of the network in terms of social support and social pressure is stressed.

In this paper special attention is paid to the influence of family networks on genetic counseling. Genetic counseling is concerned with hereditary diseases and these are by defi n a "family issue". When someone in a family is affected by a genetic disorder, communication within the family is a prerequisite for knowledge about the possibility that others may also have an affected child.

GENETIC COUNSELING AND CARRIER TESTING FOR HEMOPHILIA

Hemophilia is a bleeding disease, caused by a lack of clotting factor in the blood. Depending upon the residual clotting factor a distinction is made between severe, moderately severe, and mild hemophilia. The degree of severity is the same for all the patients within one

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family. Hemophilia patients have spontaneous bleedings or bleedings after trauma, especially in joints and muscles. Bleedings may be life- threatening, and in the long run they may cause disability. Hemophilia can not be cured, but bleedings can be treated adequately by administering concentrated blood products. Compared with a few decades ago, this treatment has improved the medical and social situation and the life expectancy considerably (Smit et al., 1989).

Hemophilia is an X-linked recessive genetic disorder. Female relatives of patients may be carriers of the hemophilia gene, which they may transmit to their sons and daughters. Carrier testing for hemophilia is offered since about 1970 and carrier testing based on DNA-analysis since 1985. Prenatal diagnosis is available since 1970, though until 1978 only the fetal sex could be determined.

METHODS

We conducted a survey on genetic counseling among women who were obligate or possible carriers of hemophilia (women with an affected father, brother, son, uncle, cousin or nephew). Hemophilia patients in The Netherlands supplied us with the addresses of these women. Thirty- one women were removed from the file of addresses because these women also participated in an evaluative study on the quality of genetic counseling and we did not want to ask them to participate in two studies within one year. We sent a mail questionnaire to 620 women aged 18 to 38 years. Fourteen respondents were found to fall outside the age-limits. The response rate was 86% (518/604 women). When we define the obligate and possible carriers as the women with the above- mentioned family relation to the patient, we estimate to have reached about a quarter of the possible carriers in The Netherlands (Varekamp et al., 1990). Eighteen women who were tested for carriership after the birth of a hemophiliac son were excluded from the analysis because either their child was the first hemophilia patient in the family and they had no reason to be tested beforehand (12x), or because they were tested shortly after their son's birth and we assumed that this happened somewhat routinely when their son was diagnosed (6x). This brings the number of respondents on 500. The sample is probably not representative. We assume that women who are not in touch with their hemophiliac relative, will be underrepresented, and that these women are at the same time less well informed about their (possible carriership and about the possibility of carrier testing.

Several aspects of the social network were examined. As interactional aspects, the contents and frequency of communications within the family were assessed. Furthermore, the size of the personal network was measured by asking the respondents with whom they talked when they were worried about personal matters, and to whom they turned for advice about important issues. On average, the respondents named 3.5 persons. Within this personal network the family network was distinguished from the network consisting of friends, colleagues and neighbors. The family network was in general much larger than the friends network, on average 2.7 for family, including the partner, against 0.7 for friends. More than half of the respondents did not name any friends or others apart from kin. This may be due to the concise way in which we assessed the network; probably we measured only the most intimate personal network. In the results the association between various variables is illustrated by means of cross-tabulations. These associations could possibly be explained by a third variable, severity of hemophilia. We controlled for severity of hemophilia.

RESULTS

Table 1 shows the general characteristics of the survey population. The women were 18 to 38 years old. Some lived with their parents. However, the majority were married or lived with a partner. About one third had children. Most of the respondents had a nearby hemophiliac relative: a father or a brother or a son. For the others it concerned a more distant relative.

Table 1. General Characteristics of the Survey Population (18-38 Years, N=500).

	%
Family situation:	
Living with parents	24
Living alone	14
Married	47
Living with a partner	14
Children	37
Family relation to patient:	
Mother	7
Sister	40
Daughter	15
Aunt	8
Niece	12
Cousin	18

Most women knew about the possibility of carrier testing. Almost everyone had a positive attitude toward carrier testing and genetic counseling. Almost half of the respondents were tested. One third was not opposed to abortion because of hemophilia and half was opposed (Table 2).

Table 2. Carrier Testing and Prenatal Diagnosis (N=500).

	%
Acquainted with carrier testing	86
Attitude towards genetic counseling and carrier testing:	
Very useful	70
Useful	28
Not so useful	2
Not useful at all	-
Use of carrier testing	45
Attitude towards abortion because of hemophilia:	
Not opposed	32
No opinion	18
Opposed	50

The majority of the respondents stated that in their parental homes the hereditary nature of hemophilia had once or more than once been discussed (table 3).

Table 3. Communication About the Hereditary Nature of Hemophilia in Parental Home.[a]

	%
Often	9
Sometimes	68
Once	4
Never	19

[a] Question: Has the hereditary nature of hemophilia been discussed at your parental home? (Before you made use of genetic counseling)

However, in 19% of the families it had never been discussed, at least not before the respondent herself was tested for carriership. When the 'nearest' patient was a brother or a father 10% stated that the hereditary nature had never been discussed in their parental home (n=272), when it was a more distant relative (n=190) 30% stated that it had never been discussed (not in table).

Most of the women, related to a hemophiliac uncle or cousin or nephew, stated that they met their hemophiliac relative more than once a year. Only 50% had ever discussed with him the fact that hemophilia is a hereditary disease (not in table).
Table 4 shows by what source the respondents had received **most** information about genetic counseling and carrier testing.

Table 4. Sources that Provide Information About Genetic Counseling and Carrier Testing

	Most important source of information
	%
parent	30
sister	11
hemophilia patient	6
other family	5
friends, colleagues	1
doctors	16
newspapers, magazines, radio and tv	3
other	6
nobody/this mail questionnaire	23

Kin, especially the parents and sisters, were the most important source of information for potential carriers. Mass media were mentioned by no more than 3%, notwithstanding the fact that many respondents stated that they had heard about carrier testing and genetic counseling on television, or had read about it in papers and magazines (not in table).

When asked with whom the respondents generally talked about respectively the hereditary nature of hemophilia, and about genetic counseling and carrier testing, it was found that the role relations most mentioned were the partner and the mother. These matters were discussed more often with the mother (resp. 62% and 45%) than with the father (resp. 38% and 27%) and much more often with sisters (resp. 48% and 35%) than with brothers (resp. 23% and 12%). Even when the father or the brother had hemophilia these matters were not always discussed with them (father resp. 64% and 49%; brothers resp. 34% and 20%). As stated before, a person's social network may promote the diffusion of a new technology by providing the person with information he/she would otherwise have missed. Table 5 shows the association between frequency of communication in the parental home about the hereditary nature of hemophilia and knowledge about carrier testing.

Table 5. Communication in Parental Home and Knowledge of Carrier Testing

Acquainted with carrier testing	Communication about heredity nature in parental home: never/once (n = 115) %	more often (n = 381) %
yes	70	90
no	30	10

Pearson's r = 24
partial r, controlled for severity, = 23
p < 0005 (one sided)

When the hereditary nature of hemophilia was more than once discussed 90% of the respondents knew about carrier testing. When hemophilia was never discussed, or just once only 70% knew about it. The Pearson's correlation coefficient between these two variables was 24 The magnitude of this coefficient did not change when controlled for severity of hemophilia.

Table 6 shows the association between frequency of communication and use of carrier testing for women that were acquainted with carrier testing. For these women, more communication about hemophilia was associated with a higher use of carrier testing. (The percentage distribution in the table does not change when the obligate carriers are omitted from the analysis).

Table 6. Communication in Parental Home and Use of Carrier Testing.[a]

Use of carrier testing	Communication about hereditary nature in parental home: never/once (n = 81) %	more often (n = 343) %
yes	40	55
no	60	45

[a] Only respondents acquainted with carrier testing are included.
Pearson's r = .13
partial r, controlled for severity, = .14
p <.005 (one sided)

This illustrates the point made in the introduction, that the network may influence the individual by giving support to use an innovation, or by putting pressure on the individual to do so. In fact it is difficult to say whether it is a matter of social support or social pressure. Individuals often do not feel social pressure or, more generally, social control unless they deviate from the norms that are prevalent in their environment.

The family may influence possible carriers directly to make use of carrier testing, but it may also happen in a more indirect way by influencing attitudes that are related with carrier testing. It was found that respondents, who were opposed to abortion because of hemophilia, made less often use of carrier testing (table 7).

Table 7. Attitude Towards Abortion and Use of Carrier Testing (Severe Hemophilia).

	Attitude towards abortion		
Carrier testing	not opposed (n=71) %	no opinion (n=35) %	opposed (n=73) %
yes	63	49	43
no	37	51	58

X2 = 6.5, df = 2, $p < .05$ (one sided)

We also compared the attitude of respondents towards abortion because of hemophilia with the attitude - as they perceived it - in their parental home. Table 8 shows the attitude in their parental home towards abortion because of hemophilia. A considerable number of respondents did not know what the opinions were about abortion in their family of orientation. Besides, about 20% stated that there were different opinions.

Table 8. Attitude at Parental Home Towards Abortion Because of Hemophilia (Severe Hemophilia, N=189).

Attitude	%
Not opposed	19
No opinion	3
Opposed	20
Different opinions	25
Do not know	33

In Table 9 the cross-tabulations of attitude of the respondent and attitude in their parent home are shown

Table 9. Attitude at Parental Home and Attitude of Respondents Towards Abortio Because of Hemophilia (Severe Hemophilia)

Attitude respondent	Attitude at parental home towards abortion		
	not opposed (n=30) %	opposed (n=34) %	other (n=110) %
not opposed	83	3	38
no opinion	10	9	26
opposed	7	88	36

X2 = 59, df = 4, p < 0005 (one sided)

When the parental home was opposed to abortion, most of the daughters were al opposed. And when the parental home was not opposed, most of the daughters were not. Tl attitude at home in this respect was often reflected in the use the daughters made of carri testing. When the family of orientation was opposed to abortion the daughters made less oft use of carrier testing (Table 10). The Tables 7, 8, 9 and 10 show only figures for seve hemophilia; for moderately severe and mild hemophilia we found about the same associ tions.

Table 10. Attitude at Parental Home Toward Abortion and Use of Carrier Testing (Severe Hemophilia).

Carrier testing	Attitude of parents toward abortion		
	not opposed (n=32) %	opposed (n=35) %	other (n=114) %
yes	72	57	48
no	28	43	52

X2 = 5.8, df = 2, p < 05 (one sided)

We have explored whether the parental influence was less penetrating in some circumstances than in others. In the literature on social networks it is often stated that the more close-knit the network is, that is, the more people in a network know each other, the more the social network develops consensus on norms and exerts social control to adhere to these norms (Bott, 1971; McKinlay, 1973). Networks consisting of kin are more close-knit than networks also consisting of friends. The respondents' attitude toward abortion was compared with the attitude toward abortion in their parental homes, apart for women who had only kin in their personal networks (the close-knits) and for women who also had friends in their networks (the loose-knits). The hypothesis was that the first group resembled their parental home more in opinion on abortion than the second group. In Table 11 the cross-tabulations of attitudes of the respondent and attitudes in their parental home are shown for the close-knit networks and the loose-knit networks separately.

Table 11. Close-Knit and Loose-Knit Networks and Attitudes Toward Abortion.

	Attitude towards abortion (parental home)		
Attitude toward abortion (respondent)	not opposed (n=28) %	opposed (n=86) %	other (n=131) %
Not opposed	93	8	27
No opinion	-	4	24
Opposed	7	88	49

Close-knit network (n=245)

	Attitude towards abortion (parental home)		
Attitude toward abortion (respondent)	not opposed (n=19) %	opposed (n=58) %	other (n=119) %
Not opposed	74	5	45
No opinion	16	10	24
Opposed	11	85	32

Loose-Knit network (n=196)

The respondents with a close-knit network had more often the same opinion as their parents than the respondents with a loose-knit network.

DISCUSSION

One out of every five women stated that in their parental home the hereditary nature of the disease their father, brother, uncle, nephew or cousin suffered from, was never discussed. Perhaps the family does not know that it concerns a genetic disorder. However, it is more likely that the risk for female relatives of being a carrier is highly underestimated or that it

is more or less a taboo in these families to talk about the disease and its hereditary nature. Feelings of guilt or shame of the parents or the patient himself or feelings of being the messenger of bad news may impede discussion. The avoidance of communication about the hereditary nature of the disease will probably be a more common phenomenon than appears from our data, because our data will not be representative in this respect (see Methods). As we have shown, this lack of discussion is a main factor causing lack of knowledge about the possibility of carrier testing and as a consequence non-use of this health care service.

The social network is more than a means for the spread of information. As is found in other research on the diffusion of technologies, we found that interpersonal relations, in this case family relations, are important when people form a positive or negative attitude toward a technology (Rogers, 1981). Of course it is not surprising that women resemble their parental home in their attitudes. They are socialized by their parents, which means that they adopt most of their parents' norms. However, in discussions on reproductive decision making, it is sometimes stated that women at risk should be free of social pressure, as if social beings are completely free to make their own decisions, even on matters in which strongly felt attitudes are involved. In a study on reproductive decision making it was found that individuals do consider what other people think of their decisions (Lippman-Hand, 1979). This also holds, although presumably in a lesser degree, for the decision to have genetic counseling and carrier testing. It would be interesting to examine by means of in-depth interviews how people form attitudes on genetic counseling and prenatal diagnosis, what norms they are confronted with, whether they feel uneasy about conflicting norms, and how all this is related to the structure of their social network. And it would be interesting to explore not only how the social network influences use of reproductive technologies but also how reproductive technology may change the social relations, especially family relations, for the better or the worse.

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