GENETIC COUNSELLING AND ADULT POLYCYSTIC KIDNEY DISEASE: PATIENTS' KNOWLEDGE, PERCEPTIONS AND UNDERSTANDING

VOLUME TWO: APPENDICES

by Patricia A. Wilkie, M.A.

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Department of Psychology

University of Stirling

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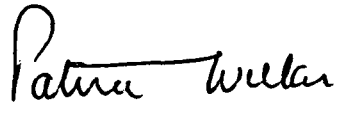
DECLARATION

I declare that this work is my own and has not appeared in any other thesis.



ABSTRACT

Adult Polycystic Kidney Disease (APKD) is a genetic disease transmitted in an autosomal dominant fashion. There is no cure. Treatment is of the symptoms as they appear usually in adulthood. Patients affected by APKD may receive genetic counselling from renal physicians.

The aims of genetic counselling can be described through paradigms which reflect the current understanding of genetics and knowledge of the illnesses. The availability of new diagnostic techniques creates a new paradigm concerned with the ethical issues of genetic testing and counselling.

An investigation into patients' knowledge, perceptions and understanding of genetic counselling was undertaken at the Renal Unit of Glasgow Royal Infirmary, prior to the establishment of a screening and counselling service for those at risk for APKD.

The main findings of the study were: the majority of patients had received some genetic counselling from renal physicians; the majority of patients had relatively good knowledge of the symptoms of and treatments for APKD; nevertheless patients believed that the two most important items to be included in genetic counselling were information about the symptoms and the treatment of APKD; patients did not fully understand the genetic inheritance of APKD; they described the risk of transmission of APKD (50-50) as a medium risk; almost all patients recommended that their at risk relatives and their children be tested for APKD; prior to the availability of prenatal diagnosis, patients thought that their children should be tested between the ages of 16 and 20.

A secondary study, including spouses of those with APKD and also haemophiliacs and their spouses, found that respondents favoured prenatal testing without termination of pregnancy and that both diseases were rated as being of medium severity.

These findings raise ethical issues for those giving genetic counselling, and have implications for the content of genetic counselling.

ACKNOWLEDGEMENTS

There are several people that I should like to thank.

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There is a very special thank you to all the patients who gave of themselves and of their time so generously and enthusiastically. It was a privilege to know them.

In particular I would like to thank all four generations of my family and especially David who has been such a loyal friend.

I should also like to thank Dr Keith Simpson for providing the photographs of kidneys reproduced in Figure 3.1, and the American Map Co, Inc, for the diagram of a kidney reproduced in Figure 3.2

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APPENDIX A: THE QUESTIONNAIRES

A.1 QUESTIONNAIRE 1

POLYCYSTIC KIDNEY DISEASE STUDY

FIRST INTERVIEW QUESTIONNAIRE

I IDENTIFICATION

Study No. ☐☐☐ 1 - 3

Hospital Number ☐☐☐☐☐ 4 - 9

Date of Interview ☐☐☐☐☐☐ 10 - 15

Video/Tape Record ☐ 16

Card Number ☐☐☐ 17 - 19

1. Present surname _____
2. Birth surname _____
3. Prenames _____
4. Address _____

_____

_____

5. Post code ☐☐☐☐☐☐☐ 20-26
6. Telephone No. _____
7. Sex ☐ 27
   Date of Birth ☐☐☐☐☐☐ 28 - 33
9. Marital Status ☐ 34
10. Year of marriage ☐☐ 35 - 36
11. Age at marriage ☐☐ 37 - 38
12. Number (live) children ☐☐ 39 - 40
13. Year of birth of children ☐☐ 41 - 42
14. Number of children deceased ☐ 43
15. Year of death ☐☐ 44 - 45
16. Cause of death _____

QUESTIONNAIRE 1

II EDUCATION

1.	How old were you on leaving school	☐☐ 46-47
2.	What qualifications did you get at school	☐ 48
3.	What qualifications have you received since leaving school	☐ 49
4.	What was the main employment of your Father _____	☐ 50
5.	What were the educational qualifications of your Father	☐ 51
	Age on leaving school	☐☐ 52-53
6.	What is the main occupation of spouse _____	☐ 54
7.	If unemployed, last employment of spouse _____	☐ 55
8.	Educational qualifications of spouse	☐ 56

QUESTIONNAIRE 1

III OCCUPATION AND EMPLOYMENT

1. What is the name of your present employment?_____ ☐ 57

2. Describe briefly what you do? _____ ☐☐ 58-59

3. How long have you been in present job?

4. Details of previous employment

Occupation	Period of employment	Reason for leaving

5. Does your employer know about your condition? ☐ 60

6. In what ways is your employer considerate? _____ ☐ 61

7. In what ways is your employer not considerate? _____ ☐ 62

8. Can you describe any special facilities or privileges you need in your job? _____ ☐ 63

9. If unemployed, length of time since last employed. ☐☐ 64-65

10. Reason for unemployment. ☐ 66

11. Have you been looking for work? ☐ 67

12. If not, why are you not looking for work? _____ ☐ 68

QUESTIONNAIRE 1

IV HOUSING STUDY NUMBER ☐☐☐ 1-3

1. What is your relationship to head of household

Child
Spouse
Parent
Sib.
Lodger
Head of Household.

☐ 4

2. Is the accommodation

privately owned
rented from local authority
rented from private landlord
rented from housing association or charitable trust.
tied with job
rented unfurnished
other

☐ 5

3. On what floor is your own front door ☐ 6

4. Is the accommodation

rooms house hostel boarding house
flat caravan institution hotel
other

☐ 7

QUESTIONNAIRE 1

V Knowledge of Inheritance and transmission

1. Can you describe how you got this condition? ☐ 8

2. Does this disorder run in the family? ☐ 9

3. If yes, which relatives are affected? ☐ 10

4. Is this an inherited condition? ☐ 11

5. If yes, how is it inherited? ☐ 12

6. Can it be passed on to children? ☐ 13

7. If yes, how is it passed on? ☐ 14

8. Is this a genetic disorder? ☐ 15

Questions 9 and 10 to be asked only if respondent understands that disorder is genetic.

9. Do you know what is the risk or probability for you to inherit polycystic kidney disease? ☐ 16

10. Do you know what is the risk or probability for your children to inherit polycystic kidney disease? ☐ 17

QUESTIONNAIRE 1

VI Knowledge of Disorder and Treatment

1. When were you diagnosed as having polycystic kidney disease? ☐☐ 18-19

2. Who told you? ☐ 20

3. What were you told about the condition?.....
..... ☐ 21

4. Were you surprised by the diagnosis? ☐ 22

5. Explain why you were or werenot surprised?..... ☐ 23
.....

6. Are you currently having treatment? ☐ 24

7. If yes, what treatment are you having? ☐ 25

8. If no, do you know what treatment is available? ☐ 26

9. What other forms of treatment are available? ☐ 27

10. What do you know about:
   (a) hypertension ☐ 28
   (b) renal transplant ☐ 29
   (c) haemodialysis ☐ 30
   (d) C.A.P.D. ☐ 31
   (e) dietary restriction ☐ 32

QUESTIONNAIRE 1

VII Social and Statutory Support

1. Is your income derived mainly from: ☐ 33
   - employment
   - occupational pension
   - state pension
   - social security
   - sickness benefit
   - other (specify)

2. Do you get help with rent and rates? ☐ 34

3. Do you receive any form of disability allowance? ☐ 35

4. How do you normally get about? ☐ 36
   - on foot
   - bus
   - train
   - own car
   - other

5. If on dialysis, how do you get to the Hospital? ☐ 37

6. Are you a car owner? ☐ 38

7. Do you get a mobility allowance? ☐ 39

8. Do you have a home help? ☐ 40

9. If yes, how often? ☐ 41

QUESTIONNAIRE 1

VII Social and Statutory Support continued

10. Do you have regular visits from:

Health visitor	☐ 42
District nurse	☐ 43
Social worker	☐ 44
General practitioner	☐ 45

11. Do you rely on others to take you out? ☐ 46

spouse

other relatives (specify)

neighbours

friends

12. Do you have a life insurance policy? ☐ 47

13. Did you have any difficulties getting life insurance? ☐ 48

14. If yes, describe..... ☐ 49

.....

15. Which, if any, Church do you belong to? ☐ 50

16. Are you a member of the Kidney Patient Association? ☐ 51

QUESTIONNAIRE 1

VIII Problems associated with Kidney disease.

Which of the following do you see as problematic?

If, for example, you do not have sickness, tick in the not applicable box.
If, on the other hand, you think it quite important, tick in that box.
Give ONE tick only for each of the categories.

	PROBLEM	Not applicable	Not at all important	Slightly important	Quite important	Very important	Extremely important
1.	Lethary						
2.	Headache						
3.	Abdominal pain						
4.	Itchiness						
5.	Nausea						
6.	Lack of concentration						
7.	Sleepiness						
8.	Sickness						
9.	Moodiness						
10.	Back pain						
11.	Other (specify)						
12.	Dependence on hospital						
13.	Restriction on what you eat						
14.	Restriction on what you drink						
15.	Feeling different from others						
16.	Difficulty at work						
17.	Fear of being unable to continue work.						
18.	Inability to remain the bread-winner.						
19.	Loss of job through ill health						
20.	Loss of income through ill health						
21.	Reduction in standard of living.						

QUESTIONNAIRE 1

VIII Which of the following do you see as problematic? (Cont...)

	Not applicable	Not at all important	Slightly important	Quite important	Very important	Extremely important
22. Restriction on physical activity eg. walking to shops and sport.						
23. Illness puts strain on marriage.						
24. Illness causes tension in the family.						
25. Illness makes it difficult to make plans.						
26. Difficulties in keeping friends.						
27. Difficulties in making relationships with opposite sex.						
28. That it is a family/genetic disorder.						
29. Illness makes it difficult to .... (please fill in your own category.						

A.2 QUESTIONNAIRE 2

POLYCYSTIC KIDNEY DISEASE STUDY

Second Interview Questionnaire NAME:

Study Number | | | Hospital Number | | | | | | |

Date of Interview | | | | | | Video/tape Record |

I. Experience of Genetic Counselling

In this section, we would like you to tell us about your experience of genetic counselling. Genetic counselling is a discussion about the inheritance of Polycystic Kidney disease, the effect on you and your children and what you can do about it.

This discussion may have been especially arranged or you may have obtained the information on other occasions e.g. when you were having treatment or during a check up.

1. Have you ever received information about the inheritance of polycystic kidney disease? ____

2. If so, who gave you this information? ____

3. And when? ____

4. Have you ever received any information about other problems related to polycystic kidney disease e.g. effect of the condition on family life? ____

QUESTIONNAIRE 2

5. If yes, who gave you this information?

6. And when.

7. If you have had genetic counselling, which of the following apply:

   (a) Was it your own idea to seek it?

   (b) Was it your husband/wife's idea to seek it?

   (c) Did you receive counselling during a routine medical visit?

   (d) Were you specially sent for?

   (e) Did your GP suggest you had counselling?

   (f) Did other family members suggest you had counselling?

   If so, specify:

   (g) No counselling?

8. Were any of the following discussed with you?

   The risk to you of inheriting polycystic kidney disease.

   The risk to your children of inheriting polycystic kidney disease.

   The advantages of testing those at risk of developing polycystic kidney disease.

QUESTIONNAIRE 2

3.

The disadvantages of testing those at risk of developing polycystic kidney disease.

How to tell those at risk.

Screening of Brothers, Sisters, Cousins in child-bearing age groups.

Possibility of adoption.

Possibility of fostering.

Voluntary childlessness - deciding to have no children.

Deciding to have no more children.

Available family planning methods.

Sterilisation.

Vasectomy.

Artificial insemination by donor. (A.I.D.)

Prevention of polycystic kidney disease.

Telling boyfriends/girlfriends that there is an inherited condition in the family.

Telling inlaws/future inlaws that there is an inherited condition in the family.

Information about the symptoms of polycystic kidney disease.

Information about treatment available.

QUESTIONNAIRE 2

9. Was your husband/wife present when you received counselling?

10. If not, would you have liked him/her to have been present?

11. Were any other members of the family given counselling?

12. If yes, specify.

13. How many times did you have discussions about the inheritance of polycystic kidney disease and its effect on you and your family?

14. Have you taken any decisions as a result of genetic counselling?

15. If yes, describe.

QUESTIONNAIRE 2

II. Attitudes to Genetic Counselling

1. Number of children.

2. How many children would you like to have?

3. Has your knowledge about polycystic kidney disease affected this?

4. If yes, describe.

5. Do you think those 'at risk' should be told of their risk of having polycystic kidney disease?

6. If yes, why should they be told?

7. Do you think those 'at risk' should be tested for polycystic kidney disease?

8. Why do you think this?

9. Do you think your children should be screened for polycystic kidney disease?

10. Why do you think this?

QUESTIONNAIRE 2

11. Is this a difficult decision to come to?

12. Do you think that knowledge of polycystic kidney disease could make any difference to the lives of those at risk?

13. If yes, describe.

14. If you have had genetic counselling, how valuable was it for you in the following respects?

    (a) Relieved stress and anxiety.

    (b) Gave information about the risk of inheritance.

    (c) Helped in deciding whether to have a family.

    (d) Gave information about symptoms of polycystic kidney disease.

    (e) Gave information about the available treatment.

    (f) Other - describe

QUESTIONNAIRE 2

15. Would you consider?

    (a) Not having children.

    (b) Sterilisation.

    (c) Vasectomy

    (d) A.I.D. (when applicable).

16. When (age) do you think children of a parent who has polycystic kidney disease, should be told of their risk?

17. Who should tell them?

18. How should they be told?

19. When (age) do you think those 'at risk' should be tested for polycystic kidney disease.

20. What kind of person should give genetic counselling?

    (a) General Practitioner

    (b) Social Worker

    (c) Nurse

    (d) Doctor in renal unit

    (e) Specialist genetic counsellor

QUESTIONNAIRE 2

(f) Other, specify ____

____

____

(g) No counselling should be given. ____

21. If you have not had any genetic counselling, but it is now available, what sort of information would you like included?

QUESTIONNAIRE 2

III. How important is it to discuss the following in any counselling about polycystic kidney disease, its inheritance and its effect on you and your family?

Please tick in the appropriate box depending on how important you think an item is.

	Not applicable	Not at all important	Slightly important	Quite important	Very important	Extremely important
1. The risk to you of inheriting polycystic kidney disease						
2. The risk to your children of inheriting polycystic kidney disease						
3. The advantages of testing those at risk of developing polycystic kidney disease						
4. The disadvantages of testing those at risk, of developing polycystic kidney disease						
5. How to tell those at risk						
6. Screening of brothers, sisters, cousins in childbearing age groups.						

QUESTIONNAIRE 2

	Not applicable	Not at all important	Slightly important	Quite important	Very important	Extremely important
7. Possibility of adoption						
8. Possibility of fostering						
9. Voluntary childlessness - deciding to have no children						
10. Deciding to have no more children						
11. Available family planning methods.						
12. Sterilisation						
13. Vasectomy						
14. Artificial insemination by donor (A.I.D.)						
15. Prevention of polycystic kidney disease						
16. Telling boy-friends/ girlfriends that there is an inherited condition in the family						

QUESTIONNAIRE 2

	Not applicable	Not at all important	Slightly important	Quite important	Very important	Extremely important
17. Telling in-laws/ future in-laws that there is an inherited condition in the family						
18. Information about the symptoms of polycystic kidney disease						
19. Information about the treatment available						

A.3 QUESTIONNAIRE 3

POLYCYSTIC KIDNEY DISEASE STUDY

Third Interview Questionnaire

Study Number: Name:
Date of Interview:

SECTION 1

Some of thse questions may not seem relevant to you. Could you please try to complete all the questions.

1. What is the name of your kidney problem?
2. How did you get it?
3. What can be done to help?
4. What are the medical problems?
5. What other (if any) problems are associated with it?
6. How is it discovered?
7. Can you catch it?
8. Can it be prevented?
9. Is it serious?
10. Where can you get information about APKD?

QUESTIONNAIRE 3

SECTION 11

1. How many children do you have?

2. Did you plan your children?

3. Did your knowledge of APKD affect this?

4. Would you like to have children?

5. How many children would you like to have?

6. Does your knowledge of APKD affect this?

7. Would you like to have grandchildren?

8. Would you like to have grandchildren if they might have APKD?

9. Do you think that your children should have children?

QUESTIONNAIRE 3

SECTION 111

1. Do you think that your children should be tested for APKD?

2. Do you want to know whether your children have not or have APKD?

3. What are the advantages of testing those at risk?

4. What advantages are there for you of having your children tested?

5. What are the disadvantages of testing those at risk?

6. Would you rather not know if your children were affected?

7. Is (was) it a relief to tell your child that he/she is at risk?

8. Do you think that those 'at risk' have a right to know of their ris

9. Do you think that parents should withhold from children informatior about this risk?

10. To whom does the information that APKD is inherited belong:-
    a Affected person
    b Doctor
    c Child of an affected person

11. Should children be given the information that APKD is inherited regardless of views of parents?

QUESTIONNAIRE 3

SECTION IV

1. What, if any, difference to your life has APKD made? Describe.

2. Does APKD cause any problems for you? Describe.

3. How long have you felt like this?

4. Do these difficulties make any difference at home?

5. Do these difficulties make any difference at work?

6. Have you had to make any changes in your life as a result of APKD?

7. What were your feelings when you were told you had APKD?

8. Do you worry in case one of your children is affected?

QUESTIONNAIRE 3

SECTION V

Which of the following apply

1. APKD is
   - a Passed from generation to generation ☐
   - b Can only be passed from female to female ☐
   - c Can only be passed from male to male ☐
   - d Can only be passed from female to male ☐
   - e Can only be passed from male to female ☐

2. The risk of inheriting APKD is
   - a A big risk ☐
   - b A medium risk ☐
   - c A small risk ☐

3. The risk of passing on APKD is
   - a A big risk ☐
   - b A medium risk ☐
   - c A small risk ☐

4. APKD
   - a Sometimes skips a generation ☐
   - b Always skips a generation ☐
   - c Never skips a generation ☐

5. The risk of inheriting APKD is
   - a 1 in 20 ☐
   - b 50-50 ☐
   - c 1 in 4 ☐
   - d 1 in 2 ☐

QUESTIONNAIRE 3

Which of the following apply	True	False
1. All children of a person with APKD will develop the condition	___	___
2. On average half the children of a person with APKD will develop the problem	___	___
3. On average half the children of a person with APKD are at risk of developing the problem	___	___
4. All children of a person with APKD are at risk of developing the problem	___	___
5. A person with APKD will sometimes have a parent with APKD	___	___
A person with APKD will always have a parent with APKD	___	___
A person with APKD will never have a parent with APKD	___	___
6. Does APKD always have symptoms	___	___
Does APKD sometimes have symptoms	___	___

QUESTIONNAIRE 3

SECTION VI

	YES	NO
1. What, if any, of the following symptoms may be associated with APKD?		
Obesity	___	___
Headache	___	___
Kidney Stones	___	___
Infection in urine	___	___
High blood pressure	___	___
Heartburn	___	___
Cloudy urine	___	___
Tiredness	___	___
Digestive problems	___	___
Pain	___	___
Itchy skin	___	___
Swollen ankles	___	___
2. Which, if any, of the following could help someone with APKD?		
Water tablets	___	___
Blood pressure tablets	___	___
Kidney machine (dialysis)	___	___
Exercise	___	___
Diet	___	___
Kidney transplant	___	___
Rest	___	___

QUESTIONNAIRE 3

SECTION VIII

Problems Associated with Kidney Disease

Which of the following do you see as problematic?
If, for example, you do not have sickness, tick in the not applicable box.
If, on the other hand, you think it quite important, tick in that box.
Give ONE tick only for each of the categories.

PROBLEM	Not applicable	Not at all important	Slightly important	Quite important	Very important	Extremely important
1. Lethargy						
2. Headache						
3. Abdominal pain						
4. Itchiness						
5. Nausea						
6. Lack of concentration						
7. Sleepiness						
8. Sickness						
9. Moodiness						
10. Back pain						
11. Other (specify)						
12. Dependance on hospital						
13. Restriction on what you eat						
14. Restriction on what you drink						
15. Feeling different from others						
16. Difficulty at work						
17. Fear of being unable to continue work						
18. Inability to remain breadwinner						
19. Loss of job due to ill-health						
20. Loss of income through ill health						
21. Reduction in standard of living						

QUESTIONNAIRE 3

SECTION VIII (Cont...)

Which of the following do you see as problematic?

PROBLEM	Not applicable	Not at all important	Slightly important	Quite important	Very important	Extremely important
22. Restriction on physical activity eg. walking to shops, sport						
23. Illness puts strain on marriage						
24. Illness causes tension in the family						
25. Illness makes it difficult to make plans						
26. Difficulties in keeping friends						
27. Difficulties in making relationships with opposite sex						
28. That it is a family/genetic disorder						
29. Illness makes it difficult to ..... (please fill in your own category)						

QUESTIONNAIRE 3

SECTION VIII

How important is it to discuss the following in any counselling about polycystic kidney disease, its inheritance and its effect on you and your family?

Please tick in the appropriate box depending on how important you think an item is.

	Not applicable	Not at all important	Slightly important	Quite important	Very important	Extremely important
1. The risk to you of inheriting polycystic kidney disease.						
2. The risk to your children of inheriting polycystic kidney disease.						
3. The advantages of testing those at risk of developing polycystic kidney disease.						
4. The disadvantages of testing those at risk of devloping polycystic kidney disease.						
5. How to tell those at risk						
6. Screening of brothers, sisters, cousins in childbearing age groups.						

QUESTIONNAIRE 3

	Not applicable	Not at all important	Slightly important	Quite important	Very important	Extremely important
7. Possibility of adoption.						
8. Possibility of fostering.						
9. Voluntary childlessness deciding to have no children						
10. Deciding to have no more children.						
11. Available family planning methods						
12. Sterilisation						
13. Vasectomy						
14. Artificial insemination by donor (AID)						
15. Prevention of polycystic kidney disease						
16. Telling boy/girl friends that there is an inherited condition in the family.						

QUESTIONNAIRE 3

	Not applicable	Not at all important	Slightly important	Quite important	Very importanat	Extremely importan
17. Telling in-laws/ future in-laws that there is an inherited condition in the family.						
18. Information about the symptoms of polycystic kidney disease.						
19. Information about the treatment available.						

QUESTIONNAIRE 3

VIII Problems associated with Kidney disease.

Which of the following do you see as problematic?

If, for example, you do not have sickness, tick in the not applicable box.
If, on the other hand, you think it quite important, tick in that box.
Give ONE tick only for each of the categories.

PROBLEM	Not applicable	Not at all important	Slightly important	Quite important	Very important	Extremely important
Lethary						
Headache						
Abdominal pain						
Itchiness						
Nausea						
Lack of concentration						
Sleepiness						
Sickness						
Moodiness						
Back pain						
Other (specify)						
Dependence on hospital						
Restriction on what you eat						
Restriction on what you drink						
Feeling different from others						
Difficulty at work						
Fear of being unable to continue work.						
Inability to remain the bread-winner.						
Loss of job through ill health						
Loss of income through ill health						
Reduction in standard of living.						

QUESTIONNAIRE 3

VIII Which of the following do you see as problematic? (Cont...)

	Not applicable	Not at all important	Slightly important	Quite important	Very important	Extreme important
22. Restriction on physical activity eg. walking to shops and sport.						
23. Illness puts strain on marriage.						
24. Illness causes tension in the family.						
25. Illness makes it difficult to make plans.						
26. Difficulties in keeping friends.						
27. Difficulties in making relationships with opposite sex.						
28. That it is a family/genetic disorder.						
29. Illness makes it difficult to .... (please fill in your own category.						

A.4 QUESTIONNAIRE 4: APKD

INSTRUCTIONS

Please read the whole questionnaire before answering the questions.

I hope that the questions are clear.

1. Most questions require only a tick in the appropriate box.

2. At first glance, one or two questions may not seem appropriate to you. For example, if your children have grown up please answer in the present as if you were considering grandchildren, or possible grandchildren.

3. Polycystic kidney disease affects men and women equally. So please, men, it is just as important to have your views on these subjects.

4. While I appreciate that you wish to help each other, I would like where possible to hear your own views. This is particularly important where couples are concerned.

All the information is strictly confidential to me alone.

QUESTIONNAIRE 4: APKD

A.

Q U E S T I O N N A I R E

Name: ..... Study Number: .....

1. Sex: Male ☐ Female ☐

2. Age Range:

15 - 19	☐
20 - 24	☐
25 - 29	☐
30 - 34	☐
35 - 39	☐
40 - 44	☐
45 - 49	☐
50 - 59	☐
60 and over	☐

3. Marital Status:

Married	☐
Single	☐
Widowed	☐
Divorced/Separated	☐
Other	☐

4. What is your relationship to adult polycystic kidney disease?
Please tick the appropriate box.

Affected	☐	Spouse of affected person	☐
Screened and unaffected	☐	Spouse of screened and unaffected person	☐
At risk (unscreened)	☐	Spouse of at risk person	☐
Unaffected	☐	Spouse of unaffected person	☐
Don't Know	☐		

QUESTIONNAIRE 4: APKD

B. Study Number .....

Q U E S T I O N N A I R E

Children and Grandchildren:

1. How many children have you ever had? .....

2. Please fill in one line in the following table for each living child.

Child Number	Age	Not at Risk	Affected	Screened and Unaffected	Not yet Screened
1	.....	☐	☐	☐	☐
2	.....	☐	☐	☐	☐
3	.....	☐	☐	☐	☐
4	.....	☐	☐	☐	☐
5	.....	☐	☐	☐	☐
6	.....	☐	☐	☐	☐

3. How many grandchildren have you ever had? .....

4. Please fill in one line in the following table for each living grandchild.

Grandchild Number	Approx. Age	Not at Risk	Affected	Screened and Unaffected	Not yet Screened
1	.....	☐	☐	☐	☐
2	.....	☐	☐	☐	☐
3	.....	☐	☐	☐	☐
4	.....	☐	☐	☐	☐
5	.....	☐	☐	☐	☐
6	.....	☐	☐	☐	☐
7	.....	☐	☐	☐	☐
8	.....	☐	☐	☐	☐
9	.....	☐	☐	☐	☐
10	.....	☐	☐	☐	☐

QUESTIONNAIRE 4: APKD

C. Study Number .....

Q U E S T I O N N A I R E

SCREENING FOR POLYCYSTIC KIDNEY DISEASE

1. Do you think that your children (or grandchildren) should be tested for adult polycystic kidney disease?
(If you do not have children, please answer as if you do, or if you have grandchildren, answer as if you are responsible for them).

Yes ☐ No ☐ Don't Know ☐ Not Applicable ☐ Have Already Been Tested ☐

2. At what age would you like to know whether or not your child (or grandchild) had adult polycystic kidney disease?

Prenatally (Before birth) ☐
0 - 4 years ☐
5 - 9 years ☐
10 - 14 years ☐
15 - 19 years ☐
20 years and over ☐
Don't Know ☐

3. Do you think that other members of your family, e.g. brothers, sisters, cousins, who may be at risk for adult polycystic kidney disease, should be told of their risk?

Yes ☐ No ☐ Don't Know ☐

4. Do you think that other members of your family, e.g. brothers, sisters, cousins, who may also be at risk for adult polycystic kidney disease, should be tested, i.e. have an ultrasound examination to show whether or not they have inherited adult polycystic kidney disease?

Yes ☐ No ☐ Don't Know ☐ Not Applicable ☐ Have Already Been Tested ☐

QUESTIONNAIRE 4: APKD

D. Study Number .....

Q U E S T I O N N A I R E

PRENATAL SCREENING

1. If, in the future, it becomes possible to tell during pregnancy whether or not a baby has polycystic kidney disease, do you think couples should take such a test?

   Yes ☐ No ☐ Don't Know ☐

2. Would you consider taking such a test to determine whether or not your baby had adult polycystic kidney disease?

   Yes ☐ No ☐ Don't Know ☐

3. Would you consider taking such a test to be followed by termination of the pregnancy, if it was shown that the baby had adult polycystic kidney disease?

   Yes ☐ No ☐ Don't Know ☐

4. Prenatal diagnosis is frequently carried out in the second three months of pregnancy as it takes time to carry out all the necessary tests.

   Would you consider having prenatal diagnosis for adult polycystic kidney disease followed by termination of an affected baby if that procedure could only be carried out in the second three months of pregnancy?

   Yes ☐ No ☐ Don't Know ☐

5. Recent advances in genetics should make it possible for prenatal diagnosis for adult polycystic kidney disease to be carried out in the first three months of pregnancy.

   Would you consider having prenatal diagnosis for adult polycystic kidney disease followed by termination of an affected baby if that procedure is carried out in the first three months of pregnancy?

   Yes ☐ No ☐ Don't Know ☐

QUESTIONNAIRE 4: APKD

E. Study Number .....

Q U E S T I O N N A I R E

TERMINATION OF PREGNANCY

1. Which of the following best reflect your views on abortion or termination of pregnancy?

   (a) I believe that it should be available on request ☐

   (b) I am totally against it ☐

   (c) I believe that it should be available sometimes ☐

2. Under which of the following circumstances would you consider termination.

   (a) For reasons of the mother's physical health ☐

   (b) For reasons of the mother's mental health ☐

   (c) Because the family is too large ☐

   (d) Because the mother is a young teenager ☐

   (e) Because the mother is unmarried ☐

   (f) Because the mother is over 40 ☐

   (g) Because one of the parents has AIDS ☐

3. Would you favour termination if it could be determined early in pregnancy that the baby would definitely have

   (a) Slight physical handicap Yes ☐ No ☐

   (b) Severe physical handicap Yes ☐ No ☐

   (c) Mild mental handicap Yes ☐ No ☐

   (d) Severe mental handicap Yes ☐ No ☐

   (e) Adult polycystic kidney disease Yes ☐ No ☐

   (f) A disease from which the child is likely to die before the age of 5 years Yes ☐ No ☐

   (g) A disease from which the child is likely to die between the ages of 6 and 10 years Yes ☐ No ☐

   (h) A disease from which the child is likely to die between the ages of 11 and 15 years Yes ☐ No ☐

   (i) The bleeding disorder haemophilia Yes ☐ No ☐

   (j) AIDS Yes ☐ No ☐

QUESTIONNAIRE 4: APKD

F. Study Number .....

Q U E S T I O N N A I R E

Could you please rank the following conditions in order of the degree of impairment likely to be caused to the individual:

	Very Mild	Mild	Moderate	Severe	Very Severe
1. Overweight					
2. Previous Heart Attack					
3. Epilepsy					
4. Cancer					
5. Adult Polycystic Kidney Disease					
6. Stomach Ulcer					
7. High Blood Pressure					
8. AIDS					
9. Diabetes					
10. Chronic Bronchitis					

QUESTIONNAIRE 4: APKD

G. Study Number .....

QUESTIONNAIRE

You have already been asked to assess the level of impairment of various conditions. Could you now think how others might view these conditions. To help you to do this, can you imagine that you are the manager of a Life Insurance Company and that people with the following conditions have applied for insurance. For each of the applicants, please say to what extent the manager might increase the cost above the normal level.

	Normal	Slight Increase	Moderate Increase	High Increase	Unacceptable
1. Overweight					
2. Previous Heart Attack					
3. Epilepsy					
4. Cancer					
5. Adult Polycystic Kidney Disease					
6. Stomach Ulcer					
7. High Blood Pressure					
8. AIDS					
9. Diabetes					
10. Chronic Bronchitis					

A.5 QUESTIONNAIRE 4: HAEMOPHILIA

INSTRUCTIONS

Please read the whole questionnaire before answering the questions.

I hope that the questions are clear.

1. Most questions require only a tick in the appropriate box.

2. At first glance, one or two questions may not seem appropriate to you. For example, if your children have grown up please answer in the present as if you were considering grandchildren, or possible grandchildren.

3. While I appreciate that you wish to help each other, I would like where possible to hear your own views. This is particularly important where couples are concerned.

The information is strictly confidential.

QUESTIONNAIRE 4: HAEMOPHILIA

A.

Q U E S T I O N N A I R E

Name: ..... Study Number: .....

1. Sex: Male ☐ Female ☐

2. Age Range:

15 - 19	☐
20 - 24	☐
25 - 29	☐
30 - 34	☐
35 - 39	☐
40 - 44	☐
45 - 49	☐
50 - 59	☐
60 and over	☐

3. Marital Status:

Married	☐
Single	☐
Widowed	☐
Divorced/Separated	☐
Other	☐

4. What is your relationship to Haemophilia?
Please tick the appropriate box.

Haemophilia Male	☐	Spouse of Haemophilia Male	☐
Male Unaffected	☐	Spouse of Male Unaffected	☐
Female Carrier	☐	Spouse of Female Carrier	☐
Female Not a Carrier	☐	Spouse of Female Not a Carrier	☐
Female Carrier Status Not Known	☐	Spouse of Female Carrier Status Not Known	☐
Don't Know	☐		

QUESTIONNAIRE 4: HAEMOPHILIA

B. Study Number .....

QUESTIONNAIRE

Children and Grandchildren:

1. How many children have you ever had? .....

2. Please fill in one line in the following table for each living child.

Child Number	Age	Male Haemophilia	Male Unaffected	Female Carrier	Female Tested Not A Carrier	Female Carrier Status Unknown
1	.....	☐	☐	☐	☐	☐
2	.....	☐	☐	☐	☐	☐
3	.....	☐	☐	☐	☐	☐
4	.....	☐	☐	☐	☐	☐
5	.....	☐	☐	☐	☐	☐
6	.....	☐	☐	☐	☐	☐

3. How many grandchildren have you ever had? .....

4. Please fill in one line in the following table for each living grandchild.

Grand Child Number	App. Age	Male Haemophilia	Male Unaffected	Female Carrier	Female Tested Not A Carrier	Female Carrier Status Unknown
1	.....	☐	☐	☐	☐	☐
2	.....	☐	☐	☐	☐	☐
3	.....	☐	☐	☐	☐	☐
4	.....	☐	☐	☐	☐	☐
5	.....	☐	☐	☐	☐	☐
6	.....	☐	☐	☐	☐	☐
7	.....	☐	☐	☐	☐	☐
8	.....	☐	☐	☐	☐	☐
9	.....	☐	☐	☐	☐	☐
10	.....	☐	☐	☐	☐	☐

QUESTIONNAIRE 4: HAEMOPHILIA

C. Study Number .....

QUESTIONNAIRE

SCREENING FOR CARRIER STATUS IN HAEMOPHILIA

1. Do you think that your daughter (or granddaughter) should be tested for carrier status?
(If you do not have a daughter, please answer as if you do, or if you have granddaughter, answer as if you are responsible for them).

Yes ☐ No ☐ Don't Know ☐ Not Applicable ☐ Have Already Been Tested ☐

2. At what age would you like to know whether or not your daughter (or granddaughter) is a carrier for haemophilia?

Prenatally (Before birth) ☐

0 - 4 years ☐

5 - 9 years ☐

10 - 14 years ☐

15 - 19 years ☐

20 years and over ☐

Don't Know ☐

3. Do you think that other members of your family, e.g. aunts, sisters, cousins, who may be at risk of being a carrier for haemophilia, should be told of their risk?

Yes ☐ No ☐ Don't Know ☐

4. Do you think that other members of your family, e.g. aunts, sisters, cousins, who may also be at risk of being a carrier of haemophilia, should be tested, for carrier status.

Yes ☐ No ☐ Don't Know ☐ Not Applicable ☐ Have Already Been Tested ☐

5. At what age would you like to know whether or not your son had haemophilia.

Prenatally (before birth) ☐

0 - 4 years ☐

5 - 9 years ☐

10 - 14 years ☐

15 - 19 years ☐

20 years and over ☐

Don't Know ☐

QUESTIONNAIRE 4: HAEMOPHILIA

D. Study Number .....

QUESTIONNAIRE

PRENATAL SCREENING

1. If, in the future, it becomes possible to tell during pregnancy whether or not a baby has haemophilia, do you think couples should take such a test?

   Yes ☐ No ☐ Don't Know ☐

2. Would you consider taking such a test to determine whether or not your baby had haemophilia?

   Yes ☐ No ☐ Don't Know ☐

3. Would you consider taking such a test to be followed by termination of the pregnancy, if it was shown that the baby had haemophilia?

   Yes ☐ No ☐ Don't Know ☐

4. Prenatal diagnosis is frequently carried out in the second three months of pregnancy as it takes time to carry out all the necessary tests.

   Would you consider having prenatal diagnosis for haemophilia followed by termination of an affected baby if that procedure could only be carried out in the second three months of pregnancy?

   Yes ☐ No ☐ Don't Know ☐

5. Recent advances in genetics should make it possible for prenatal diagnosis for haemophilia to be carried out in the first three months of pregnancy.

   Would you consider having prenatal diagnosis for haemophilia followed by termination of an affected baby if that procedure is carried out in the first three months of pregnancy?

   Yes ☐ No ☐ Don't Know ☐

QUESTIONNAIRE 4: HAEMOPHILIA

E. Study Number .....

QUESTIONNAIRE

TERMINATION OF PREGNANCY

1. Which of the following best reflect your views on abortion or termination of pregnancy?

   (a) I believe that it should be available on request ☐

   (b) I am totally against it ☐

   (c) I believe that it should be available sometimes ☐

2. Under which of the following circumstances would you consider termination.

   (a) For reasons of the mother's physical health ☐

   (b) For reasons of the mother's mental health ☐

   (c) Because the family is too large ☐

   (d) Because the mother is a young teenager ☐

   (e) Because the mother is unmarried ☐

   (f) Because the mother is over 40 ☐

   (g) Because one of the parents has AIDS ☐

3. Would you favour termination if it could be determined early in pregnancy that the baby would definitely have

   (a) Slight physical handicap Yes ☐ No ☐

   (b) Severe physical handicap Yes ☐ No ☐

   (c) Mild mental handicap Yes ☐ No ☐

   (d) Severe mental handicap Yes ☐ No ☐

   (e) Haemophilia Yes ☐ No ☐

   (f) A disease from which the child is likely to die before the age of 5 years Yes ☐ No ☐

   (g) A disease from which the child is likely to die between the ages of 6 and 10 years Yes ☐ No ☐

   (h) A disease from which the child is likely to die between the ages of 11 and 15 years Yes ☐ No ☐

   (i) AIDS Yes ☐ No ☐

QUESTIONNAIRE 4: HAEMOPHILIA

F. Study Number .....

QUESTIONNAIRE

Could you please rank the following conditions in order of the degree of impairment likely to be caused to the individual:

	Very Mild	Mild	Moderate	Severe	Very Severe
1. Overweight					
2. Previous Heart Attack					
3. Epilepsy					
4. Cancer					
5. Haemophilia					
6. Stomach Ulcer					
7. High Blood Pressure					
8. AIDS					
9. Diabetes					
10. Chronic Bronchitis					

QUESTIONNAIRE 4: HAEMOPHILIA

G. Study Number .....

QUESTIONNAIRE

You have already been asked to assess the level of impairment of various conditions. Could you now think how others might view these conditions. To help you to do this, can you imagine that you are the manager of a Life Insurance Company and that people with the following conditions have applied for insurance. For each of the applicants, please say to what extent the manager might increase the cost above the normal level.

	Normal	Slight Increase	Moderate Increase	High Increase	Unacceptable
1. Overweight					
2. Previous Heart Attack					
3. Epilepsy					
4. Cancer					
5. Haemophilia					
6. Stomach Ulcer					
7. High Blood Pressure					
8. AIDS					
9. Diabetes					
10. Chronic Bronchitis					

APPENDIX B: FURTHER DEMOGRAPHIC INFORMATION

B.1 Introduction

In this Appendix, the demographic, social, education and occupational information for each patient that was gathered during the first interview, but that was not used in the explanatory variables in Chapter 9 is described.

B.2 Age at marriage

The year and age at marriage was asked of all those who were ever married. The distribution by sex and age at marriage of the ever-married is shown in Table B.1. The ages at marriage ranged from 16 to 35 and the great majority (42 out of 56) were married between ages 20 and 29.

Table B.1.

First population: ever-married, subdivided by sex and age at marriage.

Age at marriage	Ever-married females	Ever-married males	Total
Up to 19	8	1	9
20-24	13	11	24
25-29	7	11	18
30-34	2	2	4
35-39	0	1	1
Total	30	26	56

B.3 Parent's qualifications

Many patients were asked about their parent's qualifications, though this was not felt to be an appropriate question for all these interviewed; many did not know what their

parents had done. Of the 52 who were asked, only two identified that their parent had had any qualifications at all, both of these being ordinary level school certificates.

B.4 PARENT'S OCCUPATION

Patients were also asked about the occupation of their parent; in most, perhaps all, cases the occupation of the patient's father was given. These occupations were classified in the same way as the patient's occupation had been. Table B.2 shows the occupations of patients (the 'permanent' occupation, as defined in Section 9.8) and of parents.

Table B.2

First population: occupations of patients, subdivided by sex, and of parents

Occupation	Female patients	Male patients	Total patients	Parents
Managerial	2	1	3	-
Self employed	-	-	-	3
Professional	-	2	2	4
Lesser professions	6	3	9	3
Clerical	18	4	22	6
Service jobs	4	1	5	-
Foremen	-	-	-	3
Skilled manual	-	5	5	14
Semi-skilled manual	5	7	12	19
Unskilled manual	5	1	6	14
Farmers	1	2	3	5
Agricultural workers	-	1	1	-
None	3	-	3	-
Total	44	27	71	71

It is clear that there has been considerable 'upwards mobility' in that the occupations of the patients include many more lesser professional and clerical jobs than

the parents, who had many more manual jobs. Among the parents 47 (66%) had occupations classified as skilled, semi-skilled or unskilled manual, with a further 8 (11%) in the manual jobs of foreman or farmer, whereas among the patients there were only 23 (32%) in the skilled, semi-skilled or unskilled manual jobs, with a further 4 (6%) in farming and agriculture. By contrast 31 (44%) of the patients were in lesser professions or clerical jobs, compared with 9 (13%) of their parents.

The contrast between the occupations of patients and their parents can be tested by grouping the occupations into 'non-manual' and 'manual', the former including: managerial, self-employed, professional, lesser professions, clerical and service jobs; and the latter including foremen, skilled, semi-skilled and unskilled manual, farmers and agricultural workers. Table B.3 shows compares the patients' and the parents' occupations on this basis. In the 2 by 2 table formed by omitting the 3 with no stated occupation Fisher's exact test showed that the probability of the top right hand cell having as few or fewer than 2 was 0.0096, and so significant at a 1% level.

Table B.3

First population: patient's occupation (across) by parent's occupation (down)

	Non-manual	Manual	None	Total
Non-manual	14	2	-	16
Manual	27	25	3	55
Total	41	27	3	71

The purpose of asking about the main occupation of the patient's parent was to make a comparison of occupation status between generations. It is possible that, in the presence of a chronic illness transmitted in an autosomal dominant pattern, there could be downward social mobility and this could be measured by comparing the occupations of parent and child. Downward mobility could happen in these families because the affected

parent was unable because of ill health or premature death to remain in employment with the result that opportunities for the family could be reduced. In other words in these circumstances not only is the illness transmitted from one generation to the other but social disadvantage may also have been transmitted (Paterson and Inglis 1975). The comparison of the occupation of the study respondents with the occupation of their parent shows that the reverse has occurred. This may reflect two factors: first, the study population included a high proportion of women, who are more likely to be in non-manual occupations than their fathers; and secondly there has been a changing pattern of available work in central Scotland with the decline of heavy industry. The shift to non-manual work evident in this population may be no greater than has happened in the population of this part of Scotland as a whole over a corresponding period, and might be even less, but at least it is in the same direction.

A further point to consider, however, is the extent to which those who suffer from APKD choose less strenuous occupations, and therefore move into non-manual occupations. It should also be noted that there is a difference between the study population and their parents. All of the study population apart from the six who were unaffected had APKD. On average only half of their fathers should have had APKD.

B.5 Spouse's qualifications and occupation

Those who were married were asked about the qualifications and occupation of their spouses. The qualifications were classified into the same four levels as were those of the population, and the occupations were likewise classified in the same way as the study population. For the 15 single respondents no information about spouses is applicable.

Table B.4 shows a comparison of spouse's and patients qualifications, including only those who were ever-married. Only one of the spouses was recorded as having any post-school qualifications, though 11 had school examination qualifications.

Table B.4

First population: ever-married only;
comparison of education levels of patients and spouses

Education level	Ever-married patients	Spouses
1	36	34
2	4	11
3	10	-
4	6	1
Not asked	-	10
Total	56	56

Table B.5 shows a cross-comparison of the educational levels of patient and spouse, excluding the 15 single patients. There is a tendency for those with higher educational levels to have spouses also with higher educational levels. For the 2 by 2 table formed by excluding the 'not asked' and grouping education levels 2, 3 and 4 together both for patients and for spouses Fisher's exact test shows that the probability that the patients in level 1 have would as few or fewer than 3 spouses in levels 2-4 is 0.0045, so significant at a 0.5% level.

Table B.5

First population: ever-married only;
educational levels of patient (across) and of spouse (down)

	Level 1	Level 2	Level 3	Level 4	Total
Level 1	25	2	6	1	34
Level 2	2	1	3	5	11
Level 3	-	-	-	-	-
Level 4	1	-	-	-	1
Not asked	8	1	1	-	10
Total	36	4	10	6	56

Table B.6 shows a comparison of the occupations of ever-married patients and of their spouses; many of the spouses were now retired or were housewives, and questions about their previous occupations were not asked. Table B.7 shows a cross-comparison of the occupations of ever-married patients and spouses, grouping occupations into the broad categories of non-manual and manual.

There is some similarity in the overall distributions of patients and their spouses, and a small, but not significant, tendency for patients and spouses to have the same broad category of occupation. In the 2 by 2 table formed from Table B.7 by omitting the row and the column each headed 'none', Fisher's exact test shows that the probability of the manual patients having as few or fewer than 8 non-manual spouses is 0.1571, not a significant value.

Table B.6

First population: ever-married only; patient's occupation and spouse's occupation

Occupation	Patients	Spouses
Managerial	2	2
Self employed	-	1
Professional	2	7
Lesser professions	8	10
Clerical	15	8
Service jobs	4	1
Foremen	-	2
Skilled manual	5	5
Semi-skilled manual	10	7
Unskilled manual	5	5
Farmers	2	1
Agricultural workers	1	-
None, housewife, retired	2	7
Total	56	56

Table B.7

First population: ever-married only;
patient's occupation (across) by spouse's occupation (down)

	Non-manual	Manual	None	Total
Non-manual	20	8	1	29
Manual	10	9	1	20
None	1	6	-	7
Total	31	23	2	56

B.6 Employment status

Patients were asked about their current employment. Only 39 out of the 71 (55%) were currently employed, the remaining 32 (45%) not being employed. The distribution

of current occupation and current reason for not being employed is shown in Table B.8, subdivided by sex and marital status.

Table B.8

First population: employment status and current occupation, subdivided by sex and marital status

Employment status and current occupation	Single females	Married females	Males	Total
Employed				
Managerial	1	-	1	2
Professional	-	-	1	1
Lesser professions	1	2	3	6
Clerical	5	3	3	11
Service jobs	1	1	1	3
Skilled manual	-	-	3	3
Semi-skilled manual	2	1	4	7
Unskilled manual	1	1	1	3
Farmers	1	-	2	3
Total employed	12	8	19	39
Not employed				
Housewife	1	16	-	17
Retired	-	5	6	11
Unemployed	1	-	-	1
Others	-	1	2	3
Total not employed	2	22	8	32
Total	14	30	27	71

Only 6 patients (5 females and 1 male who turned out to be unaffected) had reached the normal retirement ages of 60 for females and 65 for males, and all but one of these (a female) was retired. There were a further 6 (1 female and 5 males) who were described as retired, the youngest of whom was 51. All but one of these gave 'illness' as

the reason for leaving their last job, and only one of these was actively looking for work. One young single female was unemployed and also looking for work.

Since the symptoms of APKD are likely to have appeared by middle years it is not surprising that there are few older people in this population, and that many of them had had to retire early. This points to the need for patients with APKD to consider that they may have a shorter working life and, for those with occupational pension schemes, a smaller pension.

B.7 Does the employer know about the condition?

There is evidence from studies into chronic illnesses such as haemophilia (Markova and Forbes 1984) and epilepsy (Edwards et al 1986) and from evidence given by the Huntington's Chorea Association (Wilson 1992) of the reluctance of people with these illnesses to inform their employers of their illness for fear of discrimination. This may be pertinent in conditions such as APKD which can have a long symptom free period and in which a positive diagnosis can be made at an earlier age.

Patients were asked whether their employer knew of their condition and the results are shown in Table B.9. Of the 39 in this population in employment, 29 (74%) reported that their employer knew that they had APKD compared with 4 (10%) who had not told their employer; the question was not asked of 6 of the patients. This is in contrast to those with haemophilia (Markova et al 1977) where 50% of employers had not been informed that the employee had haemophilia.

Table B.9

First population: 'Does your employer know about your condition?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Yes	8	7	14	29
No	2	1	1	4
Not asked	2	-	4	6
Not employed	2	22	8	32
Total	14	30	27	71

B.8 Housing and type of accommodation

Patients were asked questions about their housing: first, what was their relationship to the head of the household; then how was the house owned (owner-occupied, local authority, ...); what type of building was it (house, flat, ...); and how many floors up was the entrance. House ownership has been discussed in Section 9.9. The results for the other questions are shown in Tables B.10(a), (b) and (c), subdivided by sex and marital status.

All the males described themselves as heads of household, and all but one of the married females (29 out of 30) described themselves as spouses of the head (including one of the widows, but not the other). A majority of the single women (8 out of 14, mostly but not all younger ones) were still living at home with their parents.

Unsuitable housing can cause problems in the management of patients with chronic illness. For example patients unable to climb stairs can become prisoners in their own home when the front door is not at ground floor level.

Table B.10

First population: subdivided by sex and marital status.

	Single females	Married females	Males	Total
(a) relationship to head of household				
Head of household	4	1	27	32
Spouse	-	29	-	29
Child	8	-	-	8
Other	2	-	-	2
(b) type of building				
House	11	24	22	57
Flat	3	4	3	10
Other	-	2	2	4
(c) 'on what floor is the front door?'				
Ground	11	25	25	62
First	1	3	1	5
Other	2	1	1	4
Total	14	30	27	71

Traditionally a high proportion of Scottish city dwellers live in flats with or without lifts. In this population 62 out of the 71 (87%) had ground floor accommodation with only 9 (13%) living in upper floor flats.

Two patients complained of having problems with their housing. In one case there was damp and in the other case the patient could not manage the stairs. In both instances the patients rented their houses from private landlords.

B.9 INCOME MAINTENANCE

Patients were asked a series of questions relating to their income and what sort of welfare benefits they received. The first of these was about the main source of their own

or their spouse's income; further questions asked about whether they received certain benefits such as help with rent and rates, or whether they received disability allowance. The answers to these questions are analysed in Tables B.11 and B.12.

Table B.11

First population: main source of income, subdivided by sex and marital status.

	Single females	Married females	Males	Total
Employment	10	23	16	49
State or occupational pension	1	6	5	12
State or occupational sickness benefit	-	-	2	2
Social security	2	-	1	3
Other	1	1	3	5
Total	14	30	27	71

The main source of income for 49 out of the 71 (69%) was their own or their partner's employment. A further 12 out of the 71 (17%) received occupation or state pensions, and the remaining 10 were in receipt of other benefits as shown in Table B.12.

Table B.12

First population: 'do you get help with rent and rates or disability allowance?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Neither	14	23	19	56
Rent and rates	-	4	2	6
Disability allowance	-	2	2	4
Both	-	1	4	5
Total	14	30	27	71

15 out of the 71 (21%) received help with rent and rates, or received disability allowance, or both, as shown in Table B.12. All those receiving help with rent and rates lived in local authority rented houses, but two of the 9 receiving disability allowance lived in their own houses.

This study did not examine the financial needs of patients in relation to benefits that they may be entitled to. The uptake of benefits may reflect the presence of a social worker or other member of staff who informs patients of the benefits that are available and how to apply. It may also reflect the presence of an active patient organisation. For example, the Huntington's Chorea Association in Edinburgh informs all members of benefits and how to apply and from time to time has speakers from organisations such as Disablement Income Group who can give advice. The corresponding group for patients with APKD is the Kidney Patients Association, which is a national organisation concerned with the welfare of all patients with a kidney problem. In the United Kingdom there is no organisation only for those with APKD.

B.10 Kidney Patients Association

Patients were asked whether they were members of the Kidney Patients Association (of which there was a branch in the hospital), and, if not, whether they knew of it. The answers are shown in Table B.13. Only 9 out of the 71 (13%) were members and 32 out of the 71 (45%) did not know about the organisation.

Table B.13

First population: membership and knowledge of Kidney Patients Association, subdivided by sex and marital status.

	Single females	Married females	Males	Total
Member	-	6	3	9
Not member, but knew about KPA	7	15	8	30
Not member, and did not know about KPA	7	9	16	32
Total	14	30	27	71

It is of interest to look at the answers to this question by severity of disease, and the results are shown in Table B.14. None of those who were unaffected know about the organisation.

Table B.14

First population: membership and knowledge of Kidney Patients Association, subdivided by severity of disease.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
Member	-	2	-	7	9
Not member, but knew about KPA	-	6	10	14	30
Not member, and did not know about KPA	6	6	11	9	32
Total	6	14	21	30	71

B.11 TRANSPORT

Transport to and from hospital can be problematic for patients when the hospital is not well served by public transport. Patients were asked about their normal method of transport and whether they owned a car. The results are shown in Table B.15.

Table B.15

First population: method of transport and car ownership, subdivided by sex and marital status.

	Single females	Married females	Males	Total
Car owner and user	4	14	19	37
Car owner, but uses other transport	1	6	-	7
Normally uses bus	7	7	7	21
Normally uses foot or other transport	2	3	1	6
Total	14	30	27	71

A majority of patients (44 out of 71 or 62%) owned and normally used a car. A further 7, all females, had a car in the family but normally travelled by other methods. 21 (30%) normally used bus, and a further 6 (8%) used other methods of transport (foot, bus or train).

For most patients getting to the hospital was not a problem. Two, however, both married females, normally used an ambulance for hospital visits.

Three patients (all male and including two car owners) received a mobility allowance, and one married female had applied for one.

B.12 CONTACT WITH OTHER AGENCIES

All patients attended renal clinics at Glasgow Royal Infirmary, but it was also of interest to assess the degree of medical and community support received by these patients. Questions were asked about what visits they had from other community health professionals. The results are shown in Table B.16. One patient received visits from all four professionals asked about, one received visits from their general practitioner and

from a district nurse, 5 received visits from their general practitioner, and 64 (90%), including all the single females, received no visits at all. Of the 7 who received visits from someone, 5 were in severity of disease category 3, and one each were in categories 0 and 1.

Table B.16

First population: contact with community health services, subdivided by sex and marital status.

Regular visits from:	Single females	Married females	Males	Total
General practitioner, district nurse, health visitor and social worker	-	-	1	1
General practitioner and district nurse	-	1	-	1
General practitioner	-	3	2	5
None	14	26	24	64
Total	14	30	27	71

Patients were also asked whether they had a home help; none did.

B.13 TAKING OUT

Patients were asked whom they relied on to take them out. For many this was not a problem, since they were well able to get about themselves. Four females relied on their husbands to take them out; two females relied on other relatives (but one of these was a young single girl). None relied on friends or neighbours.

B.14 LIFE INSURANCE

The difficulties for those with an incurable and potentially life threatening illness of getting life insurance has recently been highlighted by those working with patients with HIV. Indeed the inability to get life assurance is frequently stated to be a serious disadvantage for an individual who is found to be HIV positive as well as for those with illnesses such as haemophilia (Wilkie 1987). It is unlikely that applicants for life assurance and known to suffer from APKD could be accepted on ordinary premium rates for ordinary life assurance (Brackenridge 1977). Some form of life assurance is frequently used for mortgage protection and for a prospective house purchaser with dependents some form of mortgage protection is clearly desirable (Wilkie 1987). While it is possible to obtain a loan for house purchase without a life policy the disadvantage is that if the borrower dies prematurely the house may need to be sold to repay the debt. The problem for those with APKD regarding life assurance are, therefore, quite acute. The nature of the illness is that those affected may die prematurely and if they are diagnosed as having APKD or know of their own increased risk of APKD they are likely to have difficulty in obtaining life insurance.

Patients were asked whether they had had difficulties in obtaining life assurance, and, if so, why. The results are shown in Table B.17.

Table B.17

First population: life assurance, subdivided by sex and marital status.

	Single females	Married females	Males	Total
Life assurance and no difficulties	5	20	24	49
Life assurance, but with difficulties	2	5	2	9
No life assurance	7	5	1	13
Total	14	30	27	71

58 out of the 71 (82%) had acquired some form of life assurance and 13 (18%) had not. Nine (13%) stated that they had had difficulties in getting life insurance; 6 were charged a higher premium, 2 got less cover and 1 found that because she knew about APKD she could not now be accepted, though she had been accepted earlier. The nine who had had difficulties included a larger number of females (7) than of males (2).

Only one of those who was most severely affected (category 3) had had difficulty in obtaining life insurance. All of those who were unaffected had life assurance, and none had had difficulty.

APPENDIX C. SUPPLEMENTARY MEDICAL INFORMATION

C.1 MEDICAL POPULATION

Medical information was gathered from the hospital records for 64 out of the 71 in the first population. These 64 (40 females and 24 males) are described as the 'medical population'. The records were not available for 7 patients, as noted in Section 6.7, of whom 6 were found to be unaffected with APKD, and one died before the second interview. The records were incomplete in some respects for a few of the other patients. In this Appendix the medical information is summarised and discussed.

In most cases the data are analysed by sex and marital status, as defined in Section 9.3. The medical population included the following:

Single females	14
Married females (including 2 widows)	26
Males (all but 20 married)	24
Total	64

C.2 REASON FOR REFERRAL AND AGE AT REFERRAL

APKD is an illness of variable age of onset and of variability both in when patients present with symptoms and in the severity of the symptoms, and the population reflects this.

There were several reasons given to explain why the patient was referred to the renal unit by their general practitioner. The reasons for referral subdivided by sex and marital status are shown in Table C.1 and the age at referral subdivided by sex and marital status in Table C.2. The year of referral was also recorded and is shown in Table

C.3. The most common reason for referral was hypertension with 28 out of 64 (44%) referred for hypertension. Curiously, for most of the male patients (10 out of 11) this was described in the hospital records as 'blood pressure', and for most of the female patients (15 out of 17) as 'hypertension', but these have been put together in Table C.1. Loin pain was the second most common reason for referral with 12 out of 64 (19%) referred for loin pain.

The age at referral ranged from 14 to 56 for females (average 29.2, standard deviation 13.2) and from 18 to 60 for males (average 38.6, standard deviation also 13.2).

Table C.1

Medical population: reason for referral, subdivided by sex and marital status.

	Single females	Married females	Males	Total
Hypertension	8	9	11	28
Loin Pain	1	7	4	12
Haematuria	1	2	3	6
End stage renal failure	1	2	1	4
Urinary tract infection	1	1	1	3
'APKD'	2	1	-	3
Chest pain	-	-	2	2
'Pain'	-	2	-	2
Cerebrovascular accident	-	-	1	1
Pregnant	-	1	-	1
Reason missing	-	1	1	2
Total	14	26	24	64

Table C.2

Medical population: age at referral, subdivided by sex and marital status.

	Single females	Married females	Males	Total
10-14	1	-	-	1
15-19	5	-	1	6
20-24	5	6	1	12
25-29	-	3	2	5
30-34	-	4	4	8
35-39	2	2	2	6
40-44	-	4	5	9
45-49	1	2	4	7
50-54	-	2	2	4
55-59	-	1	1	2
60-64	-	-	1	1
Not recorded	-	2	1	3
Total	14	26	24	64

Table C.3

Medical population: year of referral, subdivided by sex and marital status.

	Single females	Married females	Males	Total
1960-64	-	2	-	2
1965-69	2	1	1	4
1970-74	2	4	3	9
1975-79	5	7	12	24
1980-83	5	11	7	23
Not recorded	-	1	1	2
Total	14	26	24	64

C.3 CREATININE

The creatinine level at referral and the latest creatinine level were recorded. These are shown in Tables C.4 and C.5.

Table C.4

Medical population: creatinine at referral, subdivided by sex and marital status.

	Single females	Married females	Males	Total
Up to 99	9	12	3	26
100-199	3	4	12	19
200-299	-	2	4	6
300-499	-	2	2	4
500 & over	2	3	2	7
Not recorded	-	1	1	2
Total	14	26	24	64

Table C.5

Medical population: latest creatinine level, subdivided by sex and marital status.

	Single females	Married females	Males	Total
Up to 99	9	7	-	16
100-199	2	4	8	14
200-299	-	-	3	3
300-499	-	2	1	3
500 & over	1	3	2	6
End stage renal failure	2	9	9	20
Not recorded	-	1	1	2
Total	14	26	24	64

The creatinine level had in almost every case increased between referral and the latest recording, in some cases very substantially. The few cases where there had been a small reduction were all ones with low creatinine, less than 100, levels on both occasions.

The changes were very small a difference between 65 at referral and 60 on the latest reading.

C.4 BLOOD PRESSURE

The blood pressure of the patient on referral and the latest blood pressure reading were recorded. On each occasion two readings are taken, systolic and diastolic. The blood pressures have been grouped in accordance with the following schedule, which also shows the numbers in each category at referral and at the latest date.

Table C.6

Medical population: blood pressure category at referral and at latest date.

Category	Systolic	Diastolic	At referral	At latest date
Low/low	up to 119	up to 89	6	6
Medium/low	120 to 159	up to 89	15	19
Medium/medium	120 to 159	90 to 109	15	29
Medium/high	120 to 159	110 & over	-	2
High/medium	160 & over	90 to 109	15	3
High/high	160 & over	110 & over	12	4
Not recorded	-	-	1	1
Total			64	64

Tables C.7 and C.8 show this subdivided by sex and marital status.

Table C.7

Medical population: blood pressure category at referral, subdivided by sex and marital status.

Category	Single females	Married females	Males	Total
Low/low	3	2	1	6
Medium/low	8	6	1	15
Medium/medium	2	9	4	15
Medium/high	-	-	-	-
High/medium	1	6	8	15
High/high	-	3	9	12
Not recorded	-	-	1	1
Total	14	26	24	64

Table C.8

Medical population: blood pressure category at present, subdivided by sex and marital status.

Category	Single females	Married females	Males	Total
Low/low	2	1	3	6
Medium/low	8	8	3	19
Medium/medium	3	14	12	29
Medium/high	-	1	1	2
High/medium	1	1	1	3
High/high	-	1	3	4
Not recorded	-	-	1	1
Total	14	26	24	64

Table C.9 compares the blood pressure readings at referral and at present (excluding one patient whose blood pressures were not recorded). Many of those with high blood pressure at referral had lower current readings, which possibly reflects successful treatment. In total 20 out of 63 (32%) were currently in the same category as at referral,

28 (44%) were in a lower blood pressure category and 15 (24%) were in a higher blood pressure category. The worsening could be because of deterioration with time. There were patients whose hypertension was extremely difficult to control.

Table C.9

Medical population:
blood pressure categories at referral (across) and at present (down).

Category	L/l	M/l	M/m	H/m	H/h	Total
Low/low	2	-	2	1	1	6
Med/low	3	8	5	-	3	19
Med/med	1	7	6	9	6	29
Med/high	-	-	1	-	1	2
High/med	-	-	-	3	-	3
High/high	-	-	1	2	1	4
Total	6	15	15	15	12	63

C.5 Symptoms

The presence of certain symptoms of APKD was recorded. These are discussed below.

C.5.1 Loin pain

Loin pain is a frequent symptom of APKD. The medical records classified loin pain according to severity: none, mild, moderate, severe and very severe. The degree of loin pain is shown in Table C.10 by sex and marital status and in Figure C.1 by sex and marital status and by age group. 49 out of 64 (76%) suffered from some degree of loin pain and this finding supports studies discussed in Chapter 3. Only one male had no pain, but 12 females, all but one under age 35, had no pain.

Table C.10

Medical population: degree of loin pain, subdivided by sex and marital status.

Degree of pain	Single females	Married females	Males	Total
None	7	5	1	13
Mild	5	9	8	22
Moderate	1	8	6	15
Severe	1	2	5	8
Very severe	-	1	3	4
Not recorded	-	1	1	2
Total	14	26	24	64

Figure C.1

Medical population: degree of loin pain,
by sex and marital status and by age group.

Age group	Single females	Married females	Males
up to 24	0001113	-	-
25-34	0000	0000111223X	4
35-44	1	112234	011123344
45-54	1	0222	1122222333X
55-64	2	1111	111
65 & over	-	2	-

Note: the digit (0-5) shows the degree of loin pain: 0=none; 1=mild;
2=moderate; 3=severe; 4=very severe; X=not recorded

C.5.2 Haematuria

The number of patients with recorded episodes of haematuria and the number of those episodes are shown in Table C.11 by sex and marital status and in Figure C.2 by sex and marital status and by age group. 27 out of 64 (42%) had had no reported episodes of haematuria. 17 out of 24 (70%) of the males had had haematuria compared with only 18 out of 40 (45%) of the females.

Table C.11

Medical population: Number of episodes of haematuria, subdivided by sex and marital status.

Number of episodes	Single females	Married females	Males	Total
None	10	12	6	28
1	1	2	1	4
2	1	4	1	6
3	2	1	2	5
4	-	3	6	9
5	-	2	3	5
6	-	2	3	5
7	-	-	1	1
Not recorded	-	-	1	1
Total	14	26	24	64

Figure C.2

Medical population: number of episodes of haematuria, by sex and marital status and by age group.

Age group	Single females	Married females	Males
up to 24	0000123	-	-
25-34	0000	00000001244	6
35-44	0	000226	013445567
45-54	0	1246	0000344456X
55-64	3	0035	024
65 & over	-	5	-

Note: the digit (0-7) shows the number of episodes of haematuria; X=not recorded

C.5.3 Urinary tract infection

The number of incidents of urinary tract infections for each patient was recorded, and is shown in Table C.12 by sex and marital status and in Figure C.3 by sex and marital status and by age group. 33 out of 64 (51%) had had no reported incidents of

urinary tract infections. Urinary tract infection were more common among female patients and this finding is in keeping with other studies (Chapter 3). A majority of the female patients had reported an infection (21 out of 40 or 52%), whereas only about one third of male patients had done so (9 out of 24 or 38%).

Table C.12

Medical population: Number of urinary tract infections, subdivided by sex and marital status.

Number	Single females	Married females	Males	Total
None	9	10	14	33
1	1	1	-	2
2	1	4	1	6
3	1	6	1	8
4	-	2	2	4
5	2	1	3	6
6	-	2	2	4
Not recorded	-	-	1	1
Total	14	26	24	64

Figure C.3

Medical population: number of urinary tract infections, by sex and marital status and by age group.

Age group	Single females	Married females	Males
up to 24	0000125	-	-
25-34	0000	00000222336	4
35-44	0	012335	000000556
45-54	3	0446	0000023456X
55-64	5	0003	000
65 & over	-	3	-

Note: the digit (0-6) shows the number of urinary tract infections: X=not recorded.

C.5.4 Cerebral haemorrhage

Dalgaard (1957) found that there was an increased incidence of cerebral haemorrhage amongst APKD patients. In this study 5 patients (3 females and 2 males) had had a cerebral haemorrhage. Cerebral haemorrhage was also the reported cause of death of 11 of affected parents.

C.5.5 Headache

Whether the patient suffered from headaches was recorded, and was classified by degree of headache: none, mild, moderate, severe and very severe, as shown in Table C.13 by sex and marital status, and in Figure C.4 by sex and marital status and by age group. Headache was a symptom for 25 out of the 64 (39%). By comparison with other symptoms, headache seems almost to be a problem of younger patients, or at least it was as common among them as among older ones.

Table C.13

Medical population: degree of headache, subdivided by sex and marital status.

Degree of headache	Single females	Married females	Males	Total
None	10	19	8	37
Mild	2	3	1	6
Moderate	1	3	8	12
Severe	1	-	5	6
Very severe	-	-	1	1
Not recorded	-	1	1	2
Total	14	26	24	64

Figure C.4

Medical population: degree of headache, by sex and marital status and by age group.

Age group	Single females	Married females	Males
up to 24	0000023	-	-
25-34	0000	0000000122X	3
35-44	0	000012	000122334
45-54	1	0001	0000222233X
55-64	1	0000	022
65 & over	-	0	-

Note: the digit (0-5) shows the degree of headache: 0=none; 1=mild; 2=moderate; 3=severe; 4=very severe; X=not recorded

C.5.6 Gastro-intestinal complaints

The extent of gastro-intestinal complaints for each patient was recorded. These complaints included indigestion, acidity, and diarrhoea, and were classified as: none, mild, moderate, severe and very severe. In addition one patient had had a duodenal ulcer and 12 had had a hiatus hernia. These are all shown in Table C.14 by sex and marital status and in Figure C.5 by sex and marital status and by age group. 27 out of the 64 (42%) had had some degree of gastro-intestinal trouble, including those with duodenal ulcer and hiatus hernia. Apart from hiatus hernia, gastro-intestinal trouble seemed to be rather more common among males than among females, but many more females than males suffered from hiatus hernia; 9 females and 3 males out of the 12 had this complaint, all above the age of 35.

Table C.14

Medical population: gastro-intestinal complaints, subdivided by sex and marital status.

Degree of complaint	Single females	Married females	Males	Total
None	10	14	11	35
Mild	-	2	-	2
Moderate	2	2	7	11
Severe	-	-	-	-
Very severe	-	-	1	1
Duodenal ulcer	-	-	1	1
Hiatus hernia	2	7	3	12
Not recorded	-	1	1	2
Total	14	26	24	64

Figure C.5

Medical population: degree of gastro-intestinal complaints, by sex and marital status and by age group.

Age group	Single females	Married females	Males
up to 24	0000002	-	-
25-34	0000	0000000001X	0
35-44	H	0122HH	000022DHH
45-54	2	00HH	000002224HX
55-64	H	0HHH	022
65 & over	-	0	-

Note: the digit (0-5) shows the degree of gastro-intestinal trouble: 0=none; 1=mild; 2=moderate; 3=severe; 4=very severe; D=duodenal ulcer; H=hiatus hernia; X=not recorded

C.5.7 Chest pain

The extent of chest pain for each patient was recorded. One patient had had a coronary. Others who suffered from chest pain were classified as having: none, mild, moderate, severe or very severe. These are all shown in Table C.15 by sex and marital

status and in Figure C.6 by sex and marital status and by age group. Only 7 (in addition to the one with a coronary) out of 64 (11%) had had some degree of chest pain, and none had very severe chest pain. Those who suffered from chest pain were mostly males and were generally older males.

Table C.15

Medical population: degree of chest pain, subdivided by sex and marital status.

Degree of chest pain	Single females	Married females	Males	Total
None	13	23	17	53
Mild	1	1	-	2
Moderate	-	-	2	2
Severe	-	-	3	3
Coronary	-	-	1	1
Not recorded	-	2	1	3
Total	14	26	24	64

Figure C.6

Medical population: degree of chest pain, by sex and marital status and by age group.

Age group	Single females	Married females	Males
up to 24	0000000	-	-
25-34	0000	0000000000X	0
35-44	0	00001X	00000002C
45-54	0	0000	0000000003X
55-64	1	0000	233
65 & over	-	0	-

Note: the digit (0-4) shows the degree of chest pain: 0=none; 1=mild; 2=moderate; 3=severe; C=coronary; X=not recorded

C.6 TREATMENT OF SYMPTOMS

There is no specific treatment for APKD. Treatment is of the symptoms as they appear: control of hypertension with antihypertensive drugs; antibiotics for urinary tract infection and haematuria; analgesics, cyst aspiration and in some cases Rovsings operation for loin pain; and renal replacement therapy for ESRF.

C.6.1 End stage renal failure

When patients approach end stage renal failure there are only two life-saving treatments available: dialysis and transplant. 20 patients were noted as having reached end stage renal failure. Each of these either was on dialysis or had had a transplant. A further one whose creatinine levels were not recorded had presumably also done so, since he was on dialysis. These treatments are discussed in turn.

C.6.2 Dialysis

At the time of data collection 11 out of the 71 patients (15%) were receiving some form of dialysis (none of those excluded from the 'medical population' were receiving dialysis or had had a transplant). The type of dialysis being received is shown in Table C.16 by sex and marital status and in Figure C.7 by sex and marital status and by age group. In addition two patients had had unsuccessful transplants and were again receiving dialysis.

Table C.16

Medical population: treatment for renal failure, dialysis, transplant and other, subdivided by sex and marital status.

	Single females	Married females	Males	Total
No treatment	11	14	13	38
Dialysis				
CAPD	-	4	2	6
Hospital dialysis	-	1	-	1
Home dialysis	-	1	3	4
Successful transplant	2	3	5	10
Other Treatment (Diet)	1	3	1	5
Total	14	26	24	64

Figure C.7

Medical population: dialysis and transplant, by sex and marital status and by age group.

Age group	Single females	Married females	Males
up to 24	OOOOOSS	-	-
25-34	OOOO	OOOOOOOOOD	O
35-44	O	OOOCCD	OOOOCHDSS
45-54	O	CHSS	OOOOOCHHSSS
55-64	D	CGDS	OOO
65 & over	-	O	-

Note: C=CAPD; G=hospital dialysis; H=home dialysis; S=successful transplant; D=diet; O=none

Theoretically at Glasgow Royal Infirmary patients approaching end stage renal failure and requiring dialysis were given a choice between home haemodialysis and CAPD. For some patients there may be medical reasons why one form of dialysis is more or less appropriate. The numbers of patients on dialysis in this study are too small for any conclusion to be drawn about whether the sex of the patient has any bearing on the type of dialysis. Only 4 patients (3 male and 1 female) were on home haemodialysis which

requires someone to train with the patient to assist in dialysing. It is not known whether female partners are more willing to take on this role than their male counterparts.

C.6.3 Transplant

10 out of 71 patients (14%) had had a successful transplant. For two further patients (one female one male) the transplant had been unsuccessful and the patient was once again being dialysed. The numbers of those who had had transplants are shown in Table C.16 and Figure C.7.

C.6.4 Other renal failure treatment (diet)

Besides those who were on dialysis or had had a transplant there were a further 5 patients (4 females and one male) who were receiving other treatment for renal failure, principally diet. These are also noted in Table C.16 and Figure C.7. For each of these the latest creatinine level exceeded 500. It was routine to start patients on a special diet restricting the intake of protein and other foods and fluid to ease the task of the failing kidneys prior to dialysis. It was anticipated that the need for dialysis would be within 12 months, once patients with APKD had a creatinine level of 500. There were no other patients with a creatinine level exceeding 500 (though one with creatinine exactly 500 was not receiving treatment).

C.6.5 Hypertension

44 out of the 64 patients in the medical population (69%) were receiving treatment for hypertension. Hypertension affected male and female patients equally and was not related to age. The longest time that a patient was recorded to have been treated for hypertension was 16 years.

C.6.6 Treatment of loin pain

At the time the study began the treatment of loin pain was mainly drug treatment with analgesics. The type of treatment is compared with the degree of loin pain in Table C.17. Only those with no loin pain or only mild loin pain were not receiving some form of treatment.

Table C.17

Medical population: treatment of loin pain by degree of loin pain.

Degree of pain	None	Analgesics	Other	Not recorded	Total
None	13	-	-	-	13
Mild	12	9	-	1	22
Moderate	-	15	-	-	15
Severe	-	7	1	-	8
Very severe	-	1	3	-	4
Not recorded	-	-	-	2	2
Total	25	32	4	3	64

C.6.7 Other treatments

The treatments classified as 'other' are now discussed. Three patients (1 female and 2 male) had previously undergone Rovsings operation for the puncture of cysts (Rovsing 1912). Two out of these 3 were recorded as having very severe loin pain and one as severe. Rovsings is a major operation and although pain may be temporarily relieved post-operatively there is no guarantee that the existence of cysts will not continue to cause pain.

During the period of this study alternative methods of the treatment of loin pain were being examined. This included a detailed discussion about the episodes of pain, the

type of pain and when the pain occurred. Patients were asked to keep a diary. There did not appear to be any consistency as to when the pain occurred. For the two of the four patients with severe episodes of loin pain, analgesics such as Temgesic and DF118 were ineffective. Discussions were held with Dr Yogi, consultant radiologist, who suggested that where patients with APKD had severe loin pain and were not responding to analgesics and for whom the episodes of pain required admission to hospital, he would consider aspiration of cysts with local anaesthetic and under ultrasound guidance. During the course of the study 2 patients (one of whom had also had Rovsings operation) underwent cyst aspiration (Bennett et al 1987) on 3 separate occasions each. One other patient (who also had also had Rovsings operation) used a TEMS stimulator.

Loin pain in APKD can be very disabling (*Lancet* 1987). The two patients who underwent cyst aspiration had long spells off work and eventually became unemployed. There had been no particular policy regarding the management of loin pain in patients with APKD. Interest had been shown by the medical staff and by the patients to try to establish whether any factors triggered or caused the pain.

Both these patients underwent cyst aspiration which gave some relief from pain for a short period of time. In addition the method of administering analgesics to APKD patients admitted to the renal unit for treatment of loin pain was examined. The practise was to prescribe a particular drug for the patient to be administered at regular given intervals. If the pain becomes severe the patient can ask for additional relief. This system does not allow the patient any control over when they receive the analgesic. It was decided to try with one patient with very severe loin pain the use of a self administered intravenous morphine pump. In this system the patient can self-administer analgesic when it is required. The patient liked this system because it gave the patient control over the pain relief and reduced the anxiety felt by the patient of being dependent on the nurse

bringing the injection. Furthermore there is evidence (Sofaer 1985, Autton 1986) that with this system patients have less drugs than they would do if they were given on a regular basis. However, there is also some evidence that this system is not popular with nursing staff (Sofaer 1985) who feel they loose control and express concern about possible addiction of the patient. Loin pain is certainly one of the most problematic symptoms of APKD. In this study loin pain was not a problem amongst patients approaching or in ESRF. The need for pain relief is likely to be on a finite basis.

APPENDIX D: RESULTS: EXPERIENCE OF GENETIC COUNSELLING

D.1 INTRODUCTION: THE SECOND INTERVIEW

In this Appendix the responses to questions about the patients' experience of genetic counselling, which was explored during the second interview, are reported. The second population consisted of the 65 patients who attended the second interview, of whom 14 were single females, 26 were married females (including one widowed) and 25 were males. The results are presented in the first place by the sex and marital status of the respondents. Since severity of disease proved to be the most important explanatory variable in the first part of the analysis, the results are displayed also subdivided by this factor. Family history proved to be the most important single explanatory variable in the second half of the analysis and relevant results are displayed subdivided by family history. The factor combining age and number of children was also of relevance, and some results are also displayed subdivided by this factor.

D.2 THE ENVIRONMENT OF GENETIC COUNSELLING

The first set of questions related to the environment within which patients had had genetic counselling, and the circumstances of it. Respondents were asked whether they had had any genetic counselling, which was described as 'a discussion about the inheritance of polycystic kidney disease, the effect on the patient and their children and what they could do about it'. Table D.1 shows the responses subdivided by sex and marital status. Although a higher proportion of males (8 out of 25 or 32%) than of females (8 out of 40 or 20%) had not received genetic counselling the difference is not statistically significant, Fisher's exact test showing that the probability of 8 or fewer females is 0.084.

Table D.1

Second population: 'have you received information about inheritance?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Yes	10	22	17	49
No	4	4	8	16
Total	14	26	25	65

Table D.2 shows the same responses, subdivided by severity of disease.

Table D.2

Second population: 'have you received information about inheritance?', subdivided by severity of disease.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
Yes	4	13	13	19	49
No	1	0	7	8	16
Total	5	13	20	27	65

A 2 by 2 table was formed by grouping Grades 0 and 1 and Grades 2 and 3 together; Fisher's exact test shows that the probability that as few or fewer than 1 in the bottom left hand corner of this table is 0.023, ie 2.3%. All but one of those in Grades 0 and 1 had received genetic counselling; 15 out of 47 in Grades 2 and 3 had not.

Tables D.3 and D.4 show the same responses subdivided by family history and by age and number of children.

Table D.3

Second population: 'have you received information about inheritance?', subdivided by family history.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
Yes	4	4	24	17	49
No	1	6	4	5	16
Total	5	10	28	22	65

Table D.4

Second population: 'have you received information about inheritance?', subdivided by age and number of children.

	≤44, ≤2	≤44, ≥3	≥45, ≤2	≥45, ≥3	Total
Yes	30	4	9	6	49
No	5	2	5	4	16
Total	35	63	14	10	65

Patients were asked from whom they had received information about the inheritance of APKD. Tables D.5 and D.6 show the responses, subdivided by sex and marital status and by severity of disease respectively. Most had received information from doctors in the Renal Unit at GRI.

Table D.5

Second population: 'who gave you information about inheritance?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Doctors at GRI	5	19	16	40
Other doctors	1	2	-	3
GP	1	-	-	1
Genetic counsellor	2	-	1	3
Parents	1	-	-	1
Self	-	1	-	1
No one	4	4	8	16
Total	14	26	25	65

Table D.6

Second population: 'who gave you information about inheritance?', subdivided by severity of disease.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
Doctors at GRI	3	10	10	17	40
Someone else	1	3	3	2	9
No one	1	-	7	8	16
Total	5	13	20	27	65

Respondents were also asked whether they had had any information about other problems associated with APKD. Tables D.7, D.8, D.9 and D.10 show that only 6 of them (9%) said that they had.

Table D.7

Second population: 'did you receive any information about other aspects of APKD?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Yes	2	-	4	6
No	12	26	21	59
Total	14	26	25	65

Table D.8

Second population: 'did you receive any information about other aspects of APKD?', subdivided by severity of disease.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
Yes	-	2	1	3	6
No	5	11	19	24	59
Total	5	13	20	27	65

Table D.9

Second population: 'did you receive any information about other aspects of APKD?', subdivided by family history.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
Yes	-	-	5	1	6
No	5	10	23	21	59
Total	5	10	28	22	65

Table D.10

Second population: 'did you receive any information about other aspects of APKD?', subdivided by age and number of children.

	≤44, ≤2	≤44, ≥3	≥45, ≤2	≥45, ≥3	Total
Yes	5	1	-	-	6
No	30	5	14	10	59
Total	35	6	14	10	65

Patients were asked how they had come to have genetic counselling. Tables D.11 and D.12 show the replies. For the majority it had occurred during a routine medical consultation.

Table D.11

Second population: 'how did you come to have genetic counselling?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Routine medical	2	16	13	31
Other doctor's suggestion	4	2	-	7
Spouse/family suggestion	2	2	1	5
Own idea	1	2	-	3
Not asked	-	1	3	4
None	5	3	8	16
Total	14	26	25	65

Table D.12

Second population: 'how did you come to have genetic counselling?', subdivided by severity of disease.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
Routine medical	4	4	8	15	31
Other doctor's suggestion	-	3	2	1	6
Spouse/family suggestion	-	3	1	1	5
Own idea	-	2	-	1	3
Not asked	1	-	1	2	4
None	-	1	8	7	16
Total	5	13	20	27	65

Respondents were asked whether anyone else was present when the patient received genetic counselling, and, if so, who. The replies are shown in Tables D.13 and D.14. Among those who had had genetic counselling a much lower proportion of males (6 out of 17 or 35%) than of females (20 out of 32 or 62%) had had someone else present, but Fisher's exact test shows that this is not statistically significant ($p=0.0646$).

Table D.13

Second population: 'was anyone else present when you received genetic counselling, and if so who?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Spouse or partner	2	2	3	7
Spouse and child(ren)	-	1	-	1
Spouse and mother	-	1	-	1
Sib	5	6	1	12
Child(ren)	-	2	1	3
Other family member	-	1	1	2
No one	3	9	11	23
No genetic counselling	4	4	8	16
Total	14	26	25	65

Table D.14

Second population: 'was anyone else present when you received genetic counselling, and if so who?', subdivided by severity of disease.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
Spouse or partner	-	2	1	4	7
Spouse and child(ren)	-	-	-	1	1
Spouse and mother	-	-	-	1	1
Sib	-	8	2	2	12
Child(ren)	1	-	-	2	3
Other family member	-	-	1	1	2
No one	3	3	9	8	23
No genetic counselling	1	-	7	8	16
Total	5	13	20	27	65

The majority of respondents who did not have their spouse or partner present, or who had not had any genetic counselling, would have liked them to have been present (if they had had any counselling) (see Tables D.15 and D.16).

Table D.15

Second population: 'would you have liked your spouse or partner to have been present when you received genetic counselling?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Spouse or partner was present	2	4	3	9
Would have liked spouse or partner to be present	2	16	11	29
Would not have liked	10	6	11	27
Total	14	26	25	65

Table D.16

Second population: 'would you have liked your spouse or partner to have been present when you received genetic counselling?', subdivided by severity of disease.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
Spouse or partner was present	-	2	1	6	9
Would have liked spouse or partner to be present	4	8	8	9	29
Would not have liked	1	3	11	12	27
Total	5	13	20	27	65

Most patients who had received information about the inheritance of APKD had received it on only one occasion (see Tables D.17 and D.18).

Table D.17

Second population: 'how often have you received genetic counselling?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
None	4	4	8	16
Once	8	20	13	41
Twice	2	2	3	7
Several times	-	-	1	1
Total	14	26	25	65

Table D.18

Second population: 'how often have you received genetic counselling?', subdivided by severity of disease.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
None	1	-	7	8	16
Once	3	11	11	16	41
Twice	1	2	2	2	7
Several times	-	-	-	1	1
Total	5	13	20	27	65

D.3 SCORE FOR ENVIRONMENT OF GENETIC COUNSELLING

A numeric score was constructed, denoted EGCS1, based on the answers to some of the questions relating to the environment of genetic counselling:

> For the answer 'yes' to the question: 'have you received information about the inheritance of APKD?' 1 point was given. 48 respondents scored a point.
>
> For the answer 'yes' to the question: 'have you received information about other problems associated with APKD?' 1 point was given. 5 respondents scored a point.
>
> For the answers 'own idea', 'spouse's idea', or 'family suggested it' to the question: 'whose idea was it that you should have genetic counselling?', 2 points. For the answers 'specialist sent me', or 'GP suggested it', to the same question, 1 point. 8 respondents got 2 points, and 6 got 1 point.
>
> For the answer 'yes' to the question: 'was your spouse present when you received genetic counselling?' 1 point. 9 respondents scored a point.
>
> For the answer 'yes' to the question: 'were other family members present when you received genetic counselling?' 1 point. 19 respondents scored a point.
>
> For any answer of once or more often to the question: 'how often have you received genetic counselling?' 1 point. 47 respondents scored a point.

This gave a maximum of 7 points. The distributions of respondents by number of points, subdivided by sex and marital status and by severity of disease, are shown in Tables D.19 and D.20.

The score so constructed was then analysed using the GLIM system using as factors all the explanatory variables defined in Section 9.13, namely: sex, age, marital status, number of children, education level, occupation, housing, religious affiliation, severity of disease, family history, and the factor combining age and number of children. Severity of disease was the best explanatory variable, the only one significant at a 1% probability level. However, this only accounted for 21.3% of the original variance of the score ($R^2 = 0.213$), which is not a particularly high proportion. The statistic R^2 in a GLIM analysis has the same meaning as in a multiple regression analysis; if it were a simple linear regression the value of 0.213 would correspond to a correlation coefficient

of 0.46. The mean scores for the different grades of severity of disease are shown in Table D.20.

Table D.19

Second population: scores for questions on environment of genetic counselling (EGCS1), subdivided by sex and marital status.

	Single females	Married females	Males	Total
0 points	4	4	7	15
1 point	-	2	2	4
2 points	1	5	7	13
3 points	3	7	5	15
4 points	1	4	2	7
5 points	4	4	1	9
6 points	1	-	-	1
7 points	-	-	1	1
Total	14	26	25	65
Mean score	2.9	2.7	2.0	2.5

Table D.20

Second population: scores for questions on environment of genetic counselling (EGCS1), subdivided by severity of disease.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
0 points	1	-	7	7	15
1 point	-	-	2	2	4
2 points	2	2	7	2	13
3 points	2	3	1	9	15
4 points	-	1	1	5	7
5 points	-	6	1	2	9
6 points	-	1	-	-	1
7 points	-	-	1	-	1
Total	5	13	20	27	65
Mean score	2.0	4.1	1.8	2.3	2.5

D.4 INFORMATION GIVEN IN GENETIC COUNSELLING

Respondents were asked whether they had received information about 19 different topics that might have been included in a genetic counselling session. The results are shown by sex and marital status in Table D.21, by severity of disease in Table D.22, by family history in Table D.23 and by age and number of children in Table D.24.

Table D.21

Second population: 'was discussion about the specified topic included in your genetic counselling?', subdivided by sex and marital status; the number responding 'yes' is shown.

		Single females	Married females	Males	Total
	Maximum number	14	26	25	65
1	Risk of inheriting	5	12	3	20
2	Risk to children	10	21	14	45
3	Advantages of testing	2	2	4	8
4	Disadvantages of testing	2	-	1	3
5	How to tell	2	-	-	2
6	Screening of at risk	2	6	5	13
7	Adoption	2	1	-	3
8	Fostering	2	-	-	2
9	Voluntary childlessness	3	2	-	5
10	Having no more children	2	12	2	16
11	Family planning	2	1	-	3
12	Sterilisation	3	10	1	14
13	Vasectomy	2	3	-	5
14	A. I. D.	2	-	-	2
15	Prevention of APKD	6	4	2	10
16	Telling boy/girlfriend	2	-	-	2
17	Telling in-laws	2	-	-	2
18	Symptoms of APKD	2	-	2	4
19	Treatment available	2	1	2	5

Table D.22

Second population: 'was discussion about the specified topic included in your genetic counselling?', subdivided by severity of disease; the number responding 'yes' is shown.

		Grade 0	Grade 1	Grade 2	Grade 3	Total
	Maximum number	5	13	20	27	65
1	Risk of inheriting	3	9	3	5	20
2	Risk to children	3	13	11	18	45
3	Advantages of testing	-	2	3	3	8
4	Disadvantages of testing	-	2	-	1	3
5	How to tell	-	2	-	-	2
6	Screening of at risk	-	4	1	8	13
7	Adoption	-	2	-	1	3
8	Fostering	-	2	-	-	2
9	Voluntary childlessness	-	3	1	1	5
10	Having no more children	2	4	4	6	16
11	Family planning	-	2	1	-	3
12	Sterilisation	1	4	4	5	14
13	Vasectomy	2	2	-	1	5
14	A. I. D.	-	2	-	-	2
15	Prevention of APKD	1	3	3	3	10
16	Telling boy/girlfriend	-	2	-	-	2
17	Telling in-laws	-	2	-	-	2
18	Symptoms of APKD	-	2	-	2	4
19	Treatment available	-	2	1	2	5

Table D.23

Second population: 'was discussion about the specified topic included in your genetic counselling?', subdivided by family history; the number responding 'yes' is shown.

		Grade 0	Grade 1	Grade 2	Grade 3	Total
	Maximum number	5	10	28	22	65
1	Risk of inheriting	3	1	8	8	20
2	Risk to children	3	4	23	15	45
3	Advantages of testing	-	1	5	2	8
4	Disadvantages of testing	-	-	3	-	3
5	How to tell	-	-	2	-	2
6	Screening of at risk	-	1	9	3	13
7	Adoption	-	-	3	-	3
8	Fostering	-	-	2	-	2
9	Voluntary childlessness	-	-	2	3	5
10	Having no more children	2	-	13	1	16
11	Family planning	-	-	3	-	3
12	Sterilisation	1	-	10	3	14
13	Vasectomy	2	-	3	-	5
14	A. I. D.	-	-	2	-	2
15	Prevention of APKD	1	1	5	3	10
16	Telling boy/girlfriend	-	-	2	-	2
17	Telling in-laws	-	-	2	-	2
18	Symptoms of APKD	-	-	4	-	4
19	Treatment available	-	1	3	1	5

Table D.24

Second population: 'was discussion about the specified topic included in your genetic counselling?', subdivided by age and number of children; the number responding 'yes' is shown.

		≤44, ≤2	≤44, ≥3	≥45, ≤2	≥45, ≥3	Total
	Maximum number	35	6	14	10	65
1	Risk of inheriting	15	1	2	2	20
2	Risk to children	29	4	6	6	45
3	Advantages of testing	3	2	1	2	8
4	Disadvantages of testing	2	-	1	-	3
5	How to tell	2	-	-	-	2
6	Screening of at risk	7	1	1	4	13
7	Adoption	3	-	-	-	3
8	Fostering	2	-	-	-	2
9	Voluntary childlessness	5	-	-	-	5
10	Having no more children	14	1	-	1	16
11	Family planning	3	-	-	-	3
12	Sterilisation	12	1	-	1	14
13	Vasectomy	5	-	-	-	5
14	A. I. D.	2	-	-	-	2
15	Prevention of APKD	8	-	1	1	10
16	Telling boy/girlfriend	2	-	-	-	2
17	Telling in-laws	2	-	-	-	2
18	Symptoms of APKD	2	1	1	-	4
19	Treatment available	3	-	2	-	5

The number of topics that each patient had had discussion about were counted, and the results are shown in Tables D.25, D.26, D.27 and D.28.

Table D.25

Second population: number of topics about which patients responded 'yes', subdivided by sex and marital status.

	Single females	Married females	Males	Total
No genetic counselling	4	4	8	16
No topics	-	1	2	3
1	3	2	5	10
2	4	2	4	10
3	-	3	3	6
4	1	11	1	13
5	-	2	2	4
6	-	1	-	1
19	2	-	-	2
Total	14	26	25	65

Table D.26

Second population: number of topics about which patients responded 'yes', subdivided by severity of disease.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
No genetic counselling	1	-	7	8	16
No topics	-	-	2	1	3
1	2	3	2	3	10
2	-	4	3	3	10
3	-	1	1	4	6
4	1	3	4	5	13
5	-	-	1	3	4
6	1	-	-	-	1
19	-	2	-	-	2
Total	5	13	20	27	65

Table D.27

Second population: number of topics about which patients responded 'yes', subdivided by family history.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
No genetic counselling	1	6	4	5	16
No topics	-	-	1	2	3
1	2	2	4	2	10
2	-	-	3	7	10
3	-	1	3	2	6
4	1	1	8	3	13
5	-	-	3	1	4
6	1	-	-	-	1
19	-	-	2	-	2
Total	5	10	28	22	65

Table D.28

Second population: number of topics about which patients responded 'yes', subdivided by age and number of children.

	≤44, ≤2	≤44, ≥3	≥45, ≤2	≥45, ≥3	Total
No genetic counselling	5	2	5	4	16
No topics	1	-	2	-	3
1	4	1	4	1	10
2	7	1	1	1	10
3	3	-	-	3	6
4	10	2	1	-	13
5	2	-	1	1	4
6	1	-	-	-	1
19	2	-	-	-	2
Total	35	6	14	10	65

D.5 Score for content of genetic counselling

A numeric score was constructed based on the answers to the questions relating to the content in genetic counselling, denoted EGCS2. The first two points were the same as used for the environment score, EGCS1:

> For the answer 'yes' to the question: 'have you received information about the inheritance of APKD?' 1 point was given. 48 respondents scored a point.
>
> For the answer 'yes' to the question: 'have you received information about other problems associated with APKD?' 1 point was given. 5 respondents scored a point.
>
> For the answer 'yes' to each of the 19 questions: 'did you have discussion about ... ?', 1 point, with a maximum of 8 points.

This gave a maximum of 10 points. The distributions of respondents by number of points, subdivided by sex and marital status, by severity of disease, by family history and by age and number of children are shown in Tables D.29, D.30, D.31 and D.32.

Table D.29

Second population: scores for questions on content of genetic counselling (EGCS2), subdivided by sex and marital status.

	Single females	Married females	Males	Total
0 points	4	4	8	16
1 point	-	1	2	3
2 points	3	2	4	9
3 points	4	2	5	11
4 points	-	3	2	5
5 points	1	11	1	13
6 points	-	2	2	4
7 points	-	1	1	2
8 or 9 points	-	-	-	0
10 points	2	-	-	2
Total	14	26	25	65
Mean score	3.1	3.7	2.3	3.0

Table D.30

Second population: scores for questions on content of genetic counselling (EGCS2), subdivided by severity of disease.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
0 points	1	-	7	8	16
1 point	-	-	2	1	3
2 points	2	3	1	3	9
3 points	-	4	4	3	11
4 points	-	1	1	3	5
5 points	1	3	4	5	13
6 points	-	-	1	3	4
7 points	1	-	-	1	2
8 or 9 points	-	-	-	-	0
10 points	-	2	-	-	2
Total	5	13	20	27	65
Mean score	3.2	4.4	2.3	2.9	3.0

Table D.31

Second population: scores for questions on content of genetic counselling (EGCS2), subdivided by family history.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
0 points	1	6	4	5	16
1 point	-	-	1	2	3
2 points	2	2	4	1	9
3 points	-	-	3	8	11
4 points	-	1	2	3	5
5 points	1	1	8	3	13
6 points	-	-	3	1	4
7 points	1	-	1	-	2
8 or 9 points	-	-	-	-	0
10 points	-	-	2	-	2
Total	5	10	28	22	65
Mean score	3.2	1.3	4.0	2.6	3.0

Table D.32

Second population: scores for questions on content of genetic counselling (EGCS2), subdivided by age and number of children.

	≤44, ≤2	≤44, ≥3	≥45, ≤2	≥45, ≥3	Total
0 points	5	2	5	4	16
1 point	1	-	2	-	3
2 points	3	1	4	1	9
3 points	8	1	1	1	11
4 points	2	-	-	3	5
5 points	11	1	1	-	13
6 points	1	1	1	1	4
7 points	2	-	-	-	2
8 or 9 points	-	-	-	-	0
10 points	2	-	-	-	2
Total	35	6	14	10	65
Mean score	3.8	2.7	1.7	2.3	3.0

Analysis using the GLIM system showed that of all the possible explanatory variables listed in Section 9.13, family history was the most useful factor, though significant only at a 5% probability level and not at a 1% level; the factor combining age and number of children was second, also significant at a 5% probability level. There was no significant interaction term. Family history by itself accounted for 15.1% of the original variance and the combined factor for age and number of children explained a further 13.4%, making 28.5% in all ($R^2 = 0.285$). The components for the two explanatory variables, relative to the mean score of 3.00, are shown in Table D.33, and the expected scores for the two variables are shown in Table D.34.

Table D.33

Second population: components of mean scores for experience of genetic counselling (EGCS2).

Element	Component
Overall mean	3.03
Family history:	
Grade 0	+0.77
Grade 1	−1.47
Grade 2	+0.94
Grade 3	−0.70
Age and number of children:	
≤44, ≤2	+0.78
≤44, ≥3	−0.35
≥45, ≤2	−1.52
≥45, ≥3	−0.38

Table D.34

Second population: mean scores for experience of genetic counselling (EGCS2), classified by family history and by age and number of children.

	Age and number of children			
Family history	≤44, ≤2	≤44, ≥3	≥45, ≤2	≥45, ≥3
Grade 0	4.58	3.45*	2.28	3.43*
Grade 1	2.33	1.20	0.03	1.18
Grade 2	4.74	3.61	2.44	3.59
Grade 3	3.11	1.98	0.81	1.96

There were no observations in the cells marked * and the mean scores are those implied by the model.

D.6 Correlation between scores

The (Pearson product-moment) correlation coefficient between the two scores, EGCS1 and EGCS2, was calculated, as shown in Table D.35. Its value was 0.73, very significantly different from zero.

Table D.35

Second population: correlation coefficients for scores for experience of genetic counselling (EGCS1 and EGCS2).

	EGCS1	EGCS2
EGCS2	1.00	
EGCS2	0.73*	1.00

Note: * indicates that the coefficient is significantly different from zero at a 1% probability level.

APPENDIX E: RESULTS:

KNOWLEDGE OF SYMPTOMS AND TREATMENT OF APKD

E.1 Introduction

The patients' knowledge of the symptoms and treatment of APKD was elucidated by questions in the first and the third interviews. Their responses are reported in this Appendix and discussed in Chapter 11. The first part of the analysis relates to the first questionnaire with 71 respondents, and the later parts to the third questionnaire, with 47 respondents. The answers are analysed in the first place by the sex and marital status of the respondents and then by their severity of illness and by their education level, since these proved to be the most significant factors in the first part of the analysis. In the later parts the results are subdivided by the combined factor for age and number of children, since this was found to be a relevant factor for the later parts of the analysis.

E.2 Knowledge of disorder and treatment: questionnaire 1, section 6

E.2.1 Questions on knowledge of treatment

Respondents were asked in an open ended question in the first interview what they had been told about APKD. Respondents reported that they had been given information about the following topics: prognosis of APKD, inheritance of APKD, the risks to children and the association of cysts with APKD. The numbers of respondents mentioning each of these points are shown in Tables E.1, E.2 and E.3, subdivided respectively by sex and marital status, severity of disease and education level respectively.

Table E.1

First population: numbers of patients who reported having been told about point noted, subdivided by sex and marital status.

	Single females	Married females	Males	Total
Cysts	8	18	21	47
Prognosis	1	4	7	12
Inheritance of APKD	2	5	5	12
Risks to children	1	2	-	3
'Not to worry'	-	-	1	1
Maximum	14	30	27	71

Table E.2

First population: numbers of patients who reported having been told about point noted, subdivided by severity of disease.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
Cysts	4	9	11	23	47
Prognosis	-	2	3	7	12
Inheritance of APKD	-	1	5	6	12
Risks to children	-	1	1	1	3
'Not to worry'	-	-	1	-	1
Maximum	6	14	21	30	71

Table E.3

First population: numbers of patients who reported having been told about point noted, subdivided by education level.

	Level 1	Level 2	Level 3	Level 4	Total
Cysts	30	6	7	4	47
Prognosis	6	1	2	3	12
Inheritance of APKD	8	1	1	2	12
Risks to children	-	1	1	1	3
'Not to worry'	-	-	-	1	1
Maximum	42	8	14	7	71

Respondents were asked whether or not they were having treatment for APKD. The results are shown in Tables E.4, E.5 and E.6 by sex and marital status, severity of disease, and education level respectively.

Table E.4

First population: 'are you having treatment?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
No	7	7	-	14
Have had a transplant	1	1	1	3
Yes	6	22	26	54
Total	14	30	27	71

Table E.5

First population: 'are you having treatment?', subdivided by severity of disease.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
No	2	10	1	1	14
Transplant	-	-	-	3	3
Yes	4	4	20	26	54
Total	6	14	21	30	71

Table E.6

First population: 'are you having treatment?', subdivided by education level.

	Level 1	Level 2	Level 3	Level 4	Total
No	6	3	4	1	14
Transplant	3	-	-	-	3
Yes	33	5	10	6	54
Total	42	8	14	7	71

The types of treatment that patients said that they were currently receiving are shown in Tables E.7, E.8 and E.9 by sex and marital status, severity of disease, and education level respectively.

Table E.7

First population: 'what treatment are you getting?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Antihypertensive drugs	3	17	19	39
Dialysis	1	3	4	8
CAPD	-	1	2	3
Other	2	1	1	4
None	8	8	1	17
Total	14	30	27	71

Table E.8

First population: 'what treatment are you getting?', subdivided by severity of disease.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
Antihypertensive drugs	4	4	17	14	39
Dialysis	-	-	-	8	8
CAPD	-	-	-	3	3
Other	-	-	3	1	4
None	2	10	1	4	17
Total	6	14	21	30	71

Table E.9

First population: 'what treatment are you getting?', subdivided by education level.

	Level 1	Level 2	Level 3	Level 4	Total
Antihypertensive drugs	21	3	9	6	39
Dialysis	7	1	-	-	8
CAPD	2	-	1	-	3
Other	3	1	-	-	4
None	9	3	4	1	17
Total	42	8	14	7	71

Those who were having no treatment or 'other' treatment were asked whether they knew about what treatments were available. The results are shown in Tables E.10, E.11 and E.12 by sex and marital status, severity of disease, and education level respectively.

Table E.10

First population: 'do you know what treatments are available?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Knows some or all	8	21	16	45
Knows none	5	7	10	22
Not asked	1	2	1	4
Total	14	30	27	71

Table E.11

First population: 'do you know what treatments are available?', subdivided by severity of disease.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
Knows some or all	2	9	8	26	45
Knows none	4	3	11	4	22
Not asked	-	2	2	-	4
Total	6	14	21	30	71

Table E.12

First population: 'do you know what treatments are available?', subdivided by education level.

	Level 1	Level 2	Level 3	Level 4	Total
Knows some or all	26	5	12	1	45
Knows none	14	3	2	3	22
Not asked	2	-	-	2	4
Total	42	8	14	7	71

All patients were asked what forms of treatment they knew about. The forms included: anti-hypertensive treatment, transplant, haemodialysis, CAPD and diet. The numbers of patients who knew about each of these treatments are shown in Tables E.13, E.14 and E.15 by sex and marital status, severity of disease, and education level respectively.

Table E.13

First population: numbers of patients who reported knowing about form of treatment noted, subdivided by sex and marital status.

	Single females	Married females	Males	Total
Hypertension	8	16	13	37
Transplant	11	16	11	38
Haemodialysis	8	18	13	39
CAPD	2	9	8	19
Diet	5	11	11	27
Maximum	14	30	27	71

Table E.14

First population: numbers of patients who reported knowing about form of treatment noted, subdivided by severity of disease.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
Hypertension	2	6	5	24	37
Transplant	1	10	5	22	38
Haemodialysis	1	8	5	25	39
CAPD	-	2	3	14	19
Diet	-	4	2	21	27
Maximum	6	14	21	30	71

Table E.15

First population: numbers of patients who reported knowing about form of treatment noted, subdivided by education level.

	Level 1	Level 2	Level 3	Level 4	Total
Hypertension	21	6	9	1	37
Transplant	20	5	11	2	38
Haemodialysis	22	4	12	1	39
CAPD	12	1	5	1	19
Diet	16	2	8	1	27
Maximum	42	8	14	7	71

E.2.2 Score for knowledge of treatment in questionnaire 1

The questions in questionnaire 1 about the treatment of APKD were scored and a composite score of knowledge of treatment, denoted KDS1, was formed as follows.

> For the answer 'yes' or 'have had a transplant' to the question: 'Are you having treatment for APKD?', or any answer except 'none' to the question: 'what treatment are you having?', or the answer 'yes' to the question: 'do you know what treatment is available?' 1 point. 63 respondents scored a point.
>
> For a respondent who knew some or all of the other forms of treatment when asked: 'What other forms of treatment are available?' 1 point. 41 respondents scored a point.
>
> For a respondent who knew something about each of the items listed below, 1 point:
>
> | | |
> |---|---|
> | Hypertension: | 37 respondents scored a point. |
> | Transplant: | 38 respondents scored a point. |
> | Haemodialysis: | 39 respondents scored a point. |
> | CAPD: | 19 respondents scored a point. |
> | Diet: | 27 respondents scored a point. |

This gives a maximum of 7 points. The distribution of respondents by number of points is shown in Tables E.16, E.17 and E.18, subdivided by sex and marital status, severity of disease, and education level respectively.

This score had both a possible and an observed range of 0 to 7, but inspection of Tables E.16, E.17 or E.18 shows that the distribution is distinctly bimodal, with 23 respondents getting only 1 point and 17 getting 7 points.

Analysis with the GLIM system showed that both severity of disease and education level were significant explanatory variables, both at a 1% level, and that the interaction between them was also significant at a 1% level.

The mean scores for each combination of severity grade and education level are shown in Table E.19. The important point is best brought out by grouping the severity grades into 0-2 and 3, and the education levels into 1 and 2-4, and the mean scores are shown again in Table E.20 with this grouping.

Table E.16

First population: scores for questions on knowledge of treatment of APKD (KDS1), subdivided by sex and marital status.

	Single females	Married females	Males	Total
0 points	2	2	-	4
1 point	3	9	11	23
2 points	-	1	2	3
3 points	1	-	2	3
4 points	1	4	1	6
5 points	2	4	1	7
6 points	3	2	3	8
7 points	2	8	7	17
Total	14	30	27	71
Mean score	3.7	3.8	3.6	3.7

Table E.17

First population: scores for questions on knowledge of treatment of APKD (KDS1), subdivided by severity of disease.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
0 points	1	3	-	-	4
1 point	3	3	13	4	23
2 points	1	-	1	1	3
3 points	-	-	3	-	3
4 points	-	2	1	3	6
5 points	1	2	1	3	7
6 points	-	3	-	5	8
7 points	-	1	2	14	17
Total	6	14	21	30	71
Mean score	1.7	3.3	2.2	5.4	3.7

Table E.18

First population: scores for questions on knowledge of treatment of APKD (KDS1), subdivided by education level.

	Level 1	Level 2	Level 3	Level 4	Total
0 points	3	1	-	-	4
1 point	15	1	2	5	23
2 points	1	1	-	1	3
3 points	3	-	-	-	3
4 points	2	1	3	-	6
5 points	3	2	2	-	7
6 points	4	1	3	-	8
7 points	11	1	4	1	17
Total	42	8	14	7	71
Mean score	3.6	3.8	5.0	2.0	3.7

Table E.19

First population: mean scores for knowledge of treatment of APKD (KDS1), classified by severity of disease and education level.

	Education level			
Severity of disease	1	2	3	4
Grade 0	1.75	2.00	1.00	*
Grade 1	0.50	3.33	5.50	1.00
Grade 2	1.54	2.50	4.33	3.00
Grade 3	5.76	6.50	5.75	1.33

There were no observations in the cell marked *.

Table E.20

First population: mean scores for knowledge of treatment of APKD (KDS1), classified by severity of disease and education level.

	Education level	
Severity of disease	1	2-4
Grade 0-2	1.38	3.70
Grade 3	5.76	4.44

The percentages of the original variance explained by the different factors are shown in Table E.21. The full 4 by 4 table explains 63.1% of the original variance, with the interaction term explaining more than education by itself; when the 2 by 2 table is used, the percentage of the original variance explained by the model falls to 43.9%.

Table E.21

First population: percentages of original variance of score for knowledge of treatment of APKD (KDS1) explained by successive factors.

Table:	4 by 4	2 by 2
	%	%
Original variance	100.0	100.0
Severity of disease	32.9	29.9
Education level	12.9	3.1
Interaction	17.3	10.9
Total explained	63.1	43.9
Residual	36.9	56.1

A similar GLIM analysis was carried out including only the data for the second population, and including also the scores for the experience of genetic counselling, EGCS1 and EGCS2, as possible explanatory variables. Neither proved significant. The correlation coefficients between score KDS1 and these scores are 0.21 and 0.02 respectively (see Table E.43).

E.3 KNOWLEDGE OF SYMPTOMS OF APKD: QUESTIONNAIRE 3, SECTION 6

E.3.1 Questions for knowledge of symptoms of APKD

In Section 6 of questionnaire 3, respondents were asked to state which, if any, of a given list of symptoms might be associated with APKD. The answers, for all respondents combined, are shown in Table E.22. The total in each row is 47. A few respondents did not reply to any of this section, and others omitted certain items.

Table E.22

Third population: 'are the symptoms listed associated with APKD?'.

Symptom	Yes	No	Sometimes	Don't know or no reply
Obesity	7	30	-	10
Headache	21	16	-	10
Kidney stones	11	25	-	11
Infection in urine	33	5	-	9
High blood pressure	37	2	-	11
Heartburn	15	22	-	10
Cloudy urine	32	6	-	9
Tiredness	33	4	-	10
Digestive problems	19	19	-	9
Pain	33	5	1	9
Itchy skin	23	14	-	10
Swollen ankles	23	13	-	11

The numbers of respondents answering correctly in respect of each symptom, subdivided by sex and marital status, severity of disease, and age and number of children in Tables E.23, E.24 and E.25, respectively.

Table E.23

Third population: numbers replying correctly for each symptom associated with APKD ('yes' except where noted), subdivided by sex and marital status.

Symptom	Single females	Married females	Males	Total
Obesity (No)	10	13	7	30
Headache	6	9	6	21
Kidney stones (No)	10	9	6	25
Infection in urine	10	14	9	33
High blood pressure	10	18	9	37
Heartburn	2	9	4	15
Cloudy urine	11	13	8	32
Tiredness	9	14	10	33
Digestive problems	4	10	5	19
Pain	9	15	9	33
Itchy skin	7	10	6	23
Swollen ankles	6	12	5	23
Maximum	12	20	15	47

Table E.24

Third population: numbers replying correctly for each symptom associated with APKD ('yes' except where noted), subdivided by severity of disease.

Symptom	Grade 1	Grade 2	Grade 3	Total
Obesity (No)	10	9	11	30
Headache	4	7	10	21
Kidney stones (No)	10	9	6	25
Infection in urine	11	12	10	33
High blood pressure	11	14	12	37
Heartburn	2	6	7	15
Cloudy urine	12	11	9	32
Tiredness	9	12	12	33
Digestive problems	6	6	7	19
Pain	9	12	12	33
Itchy skin	6	7	10	23
Swollen ankles	8	5	10	23
Maximum	12	15	20	47

Table E.25

Third population: numbers replying correctly for each symptom associated with APKD ('yes' except where noted), subdivided by age and number of children.

Symptom	≤44, ≤3	≤44, ≥3	≥45, ≤2	≥45, ≥3	Total
Obesity (No)	22	3	3	2	30
Headache	16	3	1	1	21
Kidney stones (No)	20	2	1	2	25
Infection in urine	23	3	5	2	33
High blood pressure	26	4	4	3	37
Heartburn	9	3	1	2	15
Cloudy urine	23	4	3	2	32
Tiredness	22	4	4	3	33
Digestive problems	13	2	2	2	19
Pain	24	3	2	4	33
Itchy skin	18	2	2	1	23
Swollen ankles	17	2	3	1	23
Maximum	28	5	7	7	47

E.3.2 Score for knowledge of symptoms of APKD

A composite score for each respondent's knowledge of the symptoms of APKD was formed, denoted KDS2. For each correct answer to the question: 'Which of the following are symptoms of APKD?' 1 point was given. The symptoms, and the number of respondents answering correctly are shown in Table E.22. The maximum score was 12 points. The distribution of respondents by number of points, subdivided by sex and marital status is shown in Table E.26, by severity of disease in Table E.27, and by age and number of children in Table E.28.

Although there were peaks at 0 points and 10 points, the bimodality was less marked than the score for knowledge of treatment in questionnaire 1.

Analysis using the GLIM system, and including all the possible explanatory variables described in Section 9.13 and also the scores for experience of genetic counselling, EGCS1 and EGCS2, showed that the most useful explanatory variable was the combined factor for age and number of children, which was significant at a 1% probability level. It explained 26.7% of the original variance.

Table E.26

Third population: score for questions on symptoms of APKD (KDS2), subdivided by sex and marital status.

	Single females	Married females	Males	Total
0 points	1	1	5	7
1 point	-	1	-	1
2 points	-	1	-	1
3 points	-	-	-	0
4 points	-	2	-	2
5 points	1	2	1	4
6 points	1	-	-	1
7 points	2	1	2	5
8 points	2	2	2	6
9 points	-	1	3	4
10 points	2	6	1	9
11 points	3	3	-	6
12 points	-	-	1	1
Total	12	20	15	47
Mean score	7.8	7.3	5.6	6.9

Table E.27

Third population: score for questions on symptoms of APKD (KDS2), subdivided by severity of disease.

	Grade 1	Grade 2	Grade 3	Total
0 points	-	1	6	7
1 point	-	-	1	1
2 points	-	1	-	1
3 points	-	-	-	0
4 points	-	1	1	2
5 points	2	1	1	4
6 points	-	1	-	1
7 points	3	2	-	5
8 points	2	2	2	6
9 points	-	1	3	4
10 points	4	3	2	9
11 points	1	2	3	6
12 points	-	-	1	1
Total	12	15	20	47
Mean score	8.2	7.2	5.9	6.9

Table E.28

Third population: score for questions on symptoms of APKD (KDS2), subdivided by age and number of children.

	≤44, ≤3	≤44, ≥3	≥45, ≤2	≥45, ≥3	Total
0 points	1	1	2	3	7
1 point	-	-	-	1	1
2 points	-	-	1	-	1
3 points	-	-	-	-	0
4 points	1	-	1	-	2
5 points	2	1	1	-	4
6 points	1	-	-	-	1
7 points	4	-	-	1	5
8 points	5	-	-	1	6
9 points	1	1	1	1	4
10 points	8	1	-	-	9
11 points	4	1	1	-	6
12 points	1	-	-	-	1
Total	28	5	7	7	47
Mean score	8.3	7.0	4.4	3.6	6.9

The mean scores for each combination of age and number of children are shown in Table E.29. The mean scores for those aged 45 and over are lower than those for younger ages, and the mean scores for those with 3 or more children are lower than the scores of those with fewer children.

Table E.29

Third population: mean scores for knowledge of symptoms of APKD (KDS2), classified by age group and number of children.

	Number of children	
Age group	0-2	3 or more
up to 44	8.32	7.00
45 and over	4.43	3.57

E.4 KNOWLEDGE OF TREATMENT OF APKD: QUESTIONNAIRE 3, SECTION 6

E.4.1 Questions for knowledge of treatment of APKD

In the same Section 6 of questionnaire 3, respondents were also asked to state which, if any, of a given list of treatments might be used to treat APKD. The answers, for all respondents combined, are shown in Table E.30. The total in each row is 47. A few respondents did not reply to any of this section, and others omitted certain items.

Table E.30

Third population: 'are the treatments listed used for treating APKD?'.

Treatment	Yes	No	Sometimes	Don't know or no reply
Water tablets	26	10	-	11
Blood pressure tablets	35	1	-	11
Kidney machine	35	3	-	9
Exercise	18	16	1	12
Diet	34	4	-	9
Kidney transplant	35	2	-	10
Rest	25	6	1	15

The numbers answering 'yes' in respect of each treatment are shown in Table E.31, subdivided by sex and marital status, in Table E.32 subdivided by severity of disease, and in Table E.33 subdivided by age and number of children.

Table E.31

Third population: numbers replying 'yes' for each treatment used for APKD, subdivided by sex and marital status.

Treatment	Single females	Married females	Males	Total
Water tablets	7	12	7	26
Blood pressure tablets	11	16	8	35
Kidney machine	10	17	8	35
Exercise	5	8	5	18
Diet	10	16	8	34
Kidney transplant	11	16	8	35
Rest	6	12	7	25
Maximum	12	20	15	47

Table E.32

Third population: numbers replying 'yes' for each treatment used for APKD, subdivided by severity of disease.

Treatment	Grade 1	Grade 2	Grade 3	Total
Water tablets	8	8	10	26
Blood pressure tablets	11	13	11	35
Kidney machine	11	11	13	35
Exercise	7	5	6	18
Diet	12	10	12	34
Kidney transplant	12	11	12	35
Rest	8	9	8	25
Maximum	12	15	20	47

Table E.33

Third population: numbers replying 'yes' for each treatment used for APKD, subdivided by age and number of children.

Treatment	≤44, ≤3	≤44, ≥3	≥45, ≤2	≥45, ≥3	Total
Water tablets	20	18	3	2	26
Blood pressure tablets	26	3	3	3	35
Kidney machine	25	3	4	3	35
Exercise	14	1	2	1	18
Diet	24	3	4	3	34
Kidney transplant	26	3	3	3	35
Rest	17	3	2	3	25
Maximum	28	5	7	7	47

E.4.2 Score for knowledge of treatment of APKD

A composite score for each respondent's knowledge of the treatments for APKD was formed, denoted KDS3. For each correct answer to the question: 'which of the following are ways of treating APKD?' 1 point was given; two treatments, Exercise and Rest, which are not usually prescribed, were ignored. The treatments, and the number of respondents answering correctly are shown in Table E.30. The maximum score was 5 points. The distribution of respondents by number of points, subdivided by sex and marital status, is shown in Tables E.34, E.35 and E.36, subdivided by sex and marital status, severity of disease, and age and number of children respectively.

Table E.34

Third population: scores for questions on knowledge of treatments for APKD, subdivided by sex and marital status.

	Single females	Married females	Males	Total
0 points	1	2	6	9
1 point	-	-	-	0
2 points	-	-	-	0
3 points	1	4	2	7
4 points	4	5	2	11
5 points	6	9	5	20
Total	12	20	15	47
Mean score	4.1	3.8	2.6	3.5

Table E.35

Third population: scores for questions on knowledge of treatments for APKD, subdivided by severity of disease.

	Grade 1	Grade 2	Grade 3	Total
0 points	-	2	7	9
1 point	-	-	-	0
2 points	-	-	-	0
3 points	-	5	2	7
4 points	6	2	3	11
5 points	6	6	8	20
Total	12	15	20	47
Mean score	4.5	3.5	2.9	3.5

Table E.36

Third population: scores for questions on knowledge of treatments for APKD, subdivided by age and number of children

	≤44, ≤3	≤44, ≥3	≥45, ≤2	≥45, ≥3	Total
0 points	1	2	3	3	9
1 point	-	-	-	-	0
2 points	-	-	-	-	0
3 points	3	-	1	3	7
4 points	8	2	1	-	11
5 points	16	1	2	1	20
Total	28	5	7	7	47
Mean score	4.3	2.6	2.4	2.0	3.5

This score too showed some evidence of bimodality, with 9 respondents getting 0 points, and the rest getting 3 or more, with the largest number (20) getting the full 5 points.

Analysis using the GLIM system, and including all the possible explanatory variables described in Section 9.13 and also the scores for experience of genetic counselling, EGCS1 and EGCS2, again showed that the most useful explanatory variable was the combined factor for age and number of children, which was again significant at a 1% probability level. This time it explained 29.2% of the original variance.

The mean scores for each combination of age and number of children are shown in Table E.37. Again, the mean scores for those aged 45 and over are lower than those for younger ages, and the mean scores for those with 3 or more children are lower than the scores of those with fewer children.

Table E.37

Third population: mean scores for knowledge of treatment of APKD (KDS3), classified by age group and number of children.

	Number of children	
Age group	0-2	3 or more
up to 44	4.32	2.60
45 and over	2.43	2.00

E.5 TOTAL SCORES FOR KNOWLEDGE OF SYMPTOMS AND TREATMENT OF APKD

Two further scores were formed as totals: the first, denoted KDS4, was formed as the total score in questionnaire 3, the sum of the number of points for knowledge of symptoms (KDS2) and knowledge of treatments (KDS3). The maximum score was 17 points. The second totals score, denoted KDS5, was formed as the total score for knowledge of the symptoms and treatment for APKD in the two questionnaires combined, KDS1 plus KDS4. The maximum score was 24 points.

The average scores for the questionnaire 3 total, KDS4, and for the grand total, KDS5, are shown in Tables E.38, E.39 and E.40, subdivided by sex and marital status, severity of disease, and age and number of children respectively.

Table E.38

Third population: average scores for questions on knowledge of treatment and symptoms of APKD (KDS4 and KDS5), subdivided by sex and marital status.

	Single females	Married females	Males	Total
Total score in questionnaire 3, KDS4	11.9	11.2	8.2	10.4
Total score in questionnaires 1 and 3, KDS5	15.3	15.0	11.9	14.1

Table E.39

Third population: average scores for questions on knowledge of treatment and symptoms of APKD (KDS4 and KDS5), subdivided by severity of disease.

	Grade 1	Grade 2	Grade 3	Total
Total score in questionnaire 3, KDS4	12.7	10.7	8.8	10.4
Total score in questionnaires 1 and 3, KDS5	15.9	13.1	13.8	14.1

Table E.40

Third population: average scores for questions on knowledge of treatment and symptoms of APKD (KDS4 and KDS5), subdivided by age and number of children.

	≤44,≤3	≤44,≥3	≥45,≤2	≥45,≥3	Total
Total score in questionnaire 3, KDS4	12.6	9.6	6.9	5.6	10.4
Total score in questionnaires 1 and 3, KDS5	16.1	14.4	11.3	8.9	14.1

Both these scores were analysed using the GLIM system. The first totals score, KDS4, like its component parts, showed evidence of bimodality, with a small peak at 0 points (7) and the main peak at 15 points, though the peak is very flat.

This score is the sum of two scores which were highly correlated (correlation coefficient 0.82), and for both of which the combined factor for age and number of children was the most useful explanatory one. It is not surprising that this factor provided the best explanation for this score, again significant at a 1% probability level, and explaining again 29.2% of the original variance.

The mean scores for each combination of age and number of children are shown in Table E.41; they are simply the sum of the mean scores for the component parts, KDS2 and KDS3. The scores conform to the same pattern as before.

Table E.41

Third population: mean scores for total score for knowledge of symptoms and treatment of APKD (KDS4), classified by age group and number of children.

	Number of children	
Age group	0-2	3 or more
up to 44	12.64	9.60
45 and over	6.86	5.57

The final score in this section, KDS5, is the grand total of the scores for knowledge of the symptoms and treatment of APKD, both in questionnaire 1 and questionnaire 3. The distribution has rather little evidence of bimodality, but is fairly flat.

This component parts of this score, KDS1 and KDS4, are not closely correlated (correlation coefficient 0.01), and these two scores were best explained by different factors, severity of disease and education for KDS1 and age and number of children for KDS4. This latter factor explained more of the variance in this case, this time significant at a 5% but not at a 1% probability level, and explaining only 21.9% of the original variance.

The mean scores for each combination of age and number of children are shown in Table E.42. The scores conform to the same pattern as before.

Table E.42

Third population: mean scores for grand total score for knowledge of symptoms and treatment of APKD (KDS5), classified by age group and number of children.

	Number of children	
Age group	0-2	3 or more
up to 44	16.11	14.40
45 and over	11.29	8.86

E.6 CORRELATIONS BETWEEN SCORES

The (Pearson product-moment) correlation coefficients were calculated for each of the scores for knowledge of symptoms and treatment of APKD with each other, and with the scores for experience of genetic counselling (EGCS1 and EGCS2). The results are shown in Table E.43. Necessarily these calculations are based on the third population, except where indicated.

Table E.43

Third population: correlation coefficients for scores for knowledge of symptoms and treatment of APKD (KDS1 to KDS5).

	KDS1	KDS2	KDS3	KDS4	KDS5
KDS1	1.00				
KDS2	0.06	1.00			
KDS3	−0.11	0.82*	1.00		
KDS4	0.01	0.98#	0.92#	1.00	
KDS5	0.42#	0.92#	0.78#	0.91#	1.00
EGCS1	0.21[2]	0.27	0.31	0.30	0.33
EGCS2	0.02[2]	0.24	0.28	0.27	0.24

Notes: [2] indicates that the correlation coefficients have been calculated using the second population.
# indicates that one score forms part of another, so a correlation is likely to be found.
* indicates that the coefficient is significantly different from zero at a 1% probability level.

E.7 FURTHER QUESTIONS ON KNOWLEDGE OF APKD:

QUESTIONNAIRE 3, SECTION 1

Respondents to questionnaire 3 were asked what could be done to help someone suffering from APKD. Their responses are shown in Table E.44, E.45 and E.46,

subdivided by sex and marital status, severity of disease, and age and number of children respectively.

Table E.44

Third population: 'what can be done to help?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Monitoring	5	3	3	11
Dialysis	3	3	5	11
Control blood pressure	2	5	3	10
Combination of treatments	-	1	1	2
Research or understanding	1	1	-	2
Nothing	1	7	3	11
Total	12	20	15	47

Table E.45

Third population: 'what can be done to help?', subdivided by severity of disease.

	Grade 1	Grade 2	Grade 3	Total
Monitoring	5	4	2	11
Dialysis	3	-	8	11
Control blood pressure	-	7	3	10
Combination of treatments	-	-	2	2
Research or understanding	-	1	1	2
Nothing	4	3	4	11
Total	12	15	20	47

Table E.46

Third population: 'what can be done to help?', subdivided by age and number of children.

	≤44, ≤3	≤44, ≥3	≥45, ≤2	≥45, ≥3	Total
Monitoring	8	-	2	1	11
Dialysis	6	1	2	2	11
Control blood pressure	5	2	2	1	10
Combination of treatments	1	-	1	-	2
Research or understanding	1	-	-	1	2
Nothing	7	2	-	2	11
Total	28	5	7	7	47

Patients were asked 'what are the medical problems associated with APKD?'. The results are shown in Tables E.47(a), E.48(a) and E.49(a).

Patients were asked whether they knew of other problems that might be associated with APKD. The list of problems given is shown in Table E.47(b), E.48(b) and E.49(b).

Table E.47

Third population: subdivided by sex and marital status.

	Single females	Married females	Males	Total
(a) 'what are the medical problems of APKD?'				
Blood pressure	5	10	8	23
Kidneys don't work	4	2	4	10
Other suggestions	1	2	-	3
Combinations of suggestions	2	4	2	8
Don't know or nothing	-	2	1	3
(b) 'what other problems of APKD are there?'				
Pain	2	2	3	7
'Not a whole person'	-	2	4	6
Restricts children	1	3	-	4
Blood pressure	1	1	1	3
Infection	1	1	1	3
Assorted others	1	3	2	6
Don't know or none	6	8	4	18
Total	12	20	15	47

Table E.48

Third population: subdivided by severity of disease.

	Grade 1	Grade 2	Grade 3	Total
(a) 'what are the medical problems of APKD?'				
Blood pressure	8	7	8	23
Kidneys don't work	2	3	5	10
Other suggestions	1	-	2	3
Combinations of suggestions	1	3	4	8
Don't know or nothing	-	2	1	3
(b) 'what other problems of APKD are there?'				
Pain	1	4	2	7
'Not a whole person'	-	2	4	6
Restricts children	1	1	2	4
Blood pressure	1	1	1	3
Infection	2	-	1	3
Assorted others	1	3	2	6
Don't know or none	6	4	8	18
Total	12	15	20	47

Table E.49

Third population: subdivided by age and number of children.

	≤44, ≤3	≤44, ≥3	≥45, ≤2	≥45, ≥3	Total
(a) 'what are the medical problems of APKD?'					
Blood pressure	14	3	4	2	23
Kidneys don't work	6	2	-	2	10
Other suggestions	1	-	1	1	3
Combinations of suggestions	5	-	1	2	8
Don't know or nothing	2	-	-	1	3
(b) 'what other problems of APKD are there?'					
Pain	3	1	1	2	7
'Not a whole person'	4	-	2	-	6
Restricts children	4	-	-	-	4
Blood pressure	2	1	-	-	3
Infection	3	-	-	-	3
Assorted others	4	-	-	2	6
Don't know or none	8	3	4	3	18
Total	28	5	7	7	47

Respondents were asked whether APKD could be described as serious. The results are shown in Tables E.50, E.51 and E.52, subdivided by sex and marital status, severity of disease, and age and number of children respectively.

Table E.50

Third population: 'is APKD serious?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Yes	4	9	10	23
Moderately or 'could be'	5	6	1	12
No	3	5	3	11
Don't know	-	-	1	1
Total	12	20	15	47

Table E.51

Third population: 'is APKD serious?', subdivided by severity of disease.

	Grade 1	Grade 2	Grade 3	Total
Yes	4	6	13	23
Moderately or 'could be'	6	4	2	12
No	2	5	4	11
Don't know	-	-	1	1
Total	12	15	20	47

Table E.52

Third population: 'is APKD serious?', subdivided by age and number of children.

	≤44, ≤3	≤44, ≥3	≥45, ≤2	≥45, ≥3	Total
Yes	10	4	6	3	23
Moderately or 'could be'	10	-	1	1	12
No	8	1	-	2	11
Don't know	-	-	-	1	1
Total	28	5	7	7	47

These rather diverse questions were not consolidated into a single score.

APPENDIX F: RESULTS:

KNOWLEDGE OF GENETIC INHERITANCE AND TRANSMISSION OF APKD

F.1 Introduction

In Chapter 11 and Appendix E, the patients' knowledge of the symptoms and treatment of APKD was discussed. Their knowledge of the genetic inheritance and transmission of the disease is now considered. The patients' understanding and knowledge of the genetic inheritance and transmission of APKD was elucidated by questions in the first and the third interviews. The results are presented in this Appendix, and are discussed in Chapter 12.

The answers are analysed below in the first place by the sex and marital status of the respondent, and then in the first part of the analysis by family history and by housing tenure, which the subsequent analysis showed to be the most useful explanatory variables; in the later parts of the analysis the combined factor for age and number of children is relevant, and results are presented according to this factor.

F.2 Knowledge of inheritance in the first questionnaire

F.2.1 Questions and basic results

Section 5 of the first questionnaire was concerned with what the patients knew about the inheritance and transmission of APKD. Ten questions were asked, but some of these questions were not relevant for some respondents.

Respondents were first asked whether they could describe how they got the condition. The word 'got' was used in order to allow respondents describe how they thought the illness was acquired, whether genetically or otherwise. The answers,

subdivided by sex and marital status, by family history and by housing tenure, are shown in Tables F.1, F.2 and F.3.

Table F.1

First population: 'how did you get the condition?', subdivided by sex and marital status

	Single females	Married females	Males	Total
Correct answer	8	19	11	38
Incorrect answer	1	7	8	16
Don't know	5	4	8	17
Total	14	30	27	71

Table F.2

First population: 'how did you get the condition?', subdivided by family history.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
Correct answer	3	4	15	16	38
Incorrect answer	3	4	7	2	16
Don't know	1	3	7	6	17
Total	7	11	29	24	71

Table F.3

First population: 'how did you get the condition?', subdivided by housing tenure.

	Owner-occupier	Tenant	Total
Correct answer	17	21	38
Incorrect answer	5	11	16
Don't know	5	12	17
Total	27	44	71

Respondents were next asked whether the disorder 'ran in the family'. The answers are shown in Tables F.4, F.5 and F.6.

Table F.4

First population: 'does it run in the family?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Yes	12	20	16	48
No	1	7	4	12
Doesn't seem to	1	3	4	8
Not sure	-	-	2	2
Don't know	-	-	1	1
Total	14	30	27	71

Table F.5

First population: 'does it run in the family?', subdivided by family history.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
Yes	1	2	21	24	48
No	2	6	4	-	12
Doesn't seem to	3	3	2	-	8
Not sure	-	-	2	-	2
Don't know	1	-	-	-	1
Total	7	11	29	24	71

Table F.6

First population: 'does it run in the family?', subdivided by housing tenure.

	Owner-occupier	Tenant	Total
Yes	19	29	48
No	4	8	12
Doesn't seem to	3	5	8
Not sure	1	1	2
Don't know	-	1	1
Total	27	44	71

Every one of those with a strong family history (Grade 3) said that APKD did run in the family, as did most of those in family history Grade 2.

Respondents were next asked which of their relatives were affected. The answers are shown in Tables F.7, F.8 and F.9.

Table F.7

First population: 'which relatives are affected?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
None	2	7	10	19
Parent only	3	6	4	13
Parent and sibs	7	6	6	19
Sibs only	1	6	1	8
Parent and other relatives	1	2	1	4
Sibs and other relatives	-	1	-	1
Parent, sibs and others	-	-	2	2
Children only	-	2	1	3
Not asked	-	-	2	2
Total	14	30	27	71

Table F.8

First population: 'which relatives are affected?', subdivided by family history.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
None	5	8	5	1	19
Parent only	-	2	6	5	13
Parent and sibs	-	-	8	11	19
Sibs only	1	-	5	2	8
Parent and other relatives	1	-	1	2	4
Sibs and other relatives	-	-	-	1	1
Parent, sibs and others	-	-	1	1	2
Children only	-	-	2	1	3
Not asked	-	1	1	-	2
Total	7	11	29	24	71

Table F.9

First population: 'which relatives are affected?', subdivided by housing tenure.

	Owner-occupier	Tenant	Total
None	7	12	19
Parent only	6	7	13
Parent and sibs	9	10	19
Sibs only	3	5	8
Parent and other relatives	2	2	4
Sibs and other relatives	-	1	1
Parent, sibs and others	-	2	2
Children only	-	3	-
Not asked	-	2	2
Total	27	44	71

The respondents were next asked whether APKD was inherited. The answers are shown in Tables F.10, F.11 and F.12.

Table F.10

First population: 'is APKD inherited?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Yes	11	22	18	51
It seems to be	-	1	7	8
No	2	3	1	6
Don't know	1	3	1	5
Not asked	-	1	-	1
Total	14	30	27	71

Table F.11

First population: 'is APKD inherited?', subdivided by family history.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
Yes	4	4	23	20	51
It seems to be	-	1	5	2	8
No	1	5	-	-	6
Don't know	1	1	1	2	5
Not asked	1	-	-	-	1
Total	7	11	29	24	71

Table F.12

First population: 'is APKD inherited?', subdivided by housing tenure.

	Owner-occupier	Tenant	Total
Yes	21	30	51
It seems to be	4	4	8
No	2	4	6
Don't know	-	5	5
Not asked	-	1	1
Total	27	44	71

The patients were then asked how the condition was inherited. The answers are shown in Tables F.13, F.14 and F.15. The 7 who were not asked included those who thought that APKD was not inherited (in the previous question).

Table F.13

First population: 'how is APKD inherited?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Almost correct	-	2	4	6
Skips generations	1	1	-	2
Inherited by same sex	2	2	1	5
Not sure	4	11	7	22
Don't know	5	9	15	29
Not asked	2	5	-	7
Total	14	30	27	71

Table F.14

First population: 'how is APKD inherited?', subdivided by family history.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
Almost correct	-	2	2	2	6
Skips generations	-	-	-	2	2
Inherited by same sex	-	-	-	5	5
Not sure	-	1	11	10	22
Don't know	5	3	16	5	29
Not asked	2	5	-	-	7
Total	7	11	29	24	71

Table F.15

First population: 'how is APKD inherited?', subdivided by housing tenure.

	Owner-occupier	Tenant	Total
Almost correct	1	5	6
Skips generations	1	1	2
Inherited by same sex	3	2	5
Not sure	13	8	22
Don't know	7	22	29
Not asked	2	5	7
Total	27	44	71

Patients were then asked whether APKD could be passed on. The answers are shown in Tables F.16, F.17 and F.18.

Table F.16

First population: 'can APKD be passed on?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Yes	11	23	23	57
Don't know	3	4	4	11
Not asked	-	3	-	3
Total	14	30	27	71

Table F.17

First population: 'can APKD be passed on?', subdivided by family history.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
Yes	2	5	29	21	57
Don't know	3	5	-	3	11
Not asked	2	1	-	-	3
Total	7	11	29	24	71

Table F.18

First population: 'can APKD be passed on?', subdivided by housing tenure.

	Owner-occupier	Tenant	Total
Yes	23	33	57
Don't know	2	9	11
Not asked	2	1	3
Total	27	44	71

Patients were then asked how APKD was passed on. The results are shown in Tables F.19, F.20 and F.21.

Table F.19

First population: 'how is APKD passed on?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Almost correct	2	6	2	10
Skips generations	-	1	1	2
Mother to daughter	1	1	-	2
Germ	-	-	1	1
Not sure	5	12	8	25
Don't know	5	4	13	22
Not asked	1	6	2	9
Total	14	30	27	71

Table F.20

First population: 'how is APKD passed on?', subdivided by family history.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
Almost correct	2	-	3	5	10
Skips generations	-	-	1	1	2
Mother to daughter	-	-	1	1	2
Germ	-	-	-	1	1
Not sure	1	2	13	9	25
Don't know	1	3	11	7	22
Not asked	3	6	-	-	9
Total	7	11	29	24	71

Table F.21

First population: 'how is APKD passed on?', subdivided by housing tenure.

	Owner-occupier	Tenant	Total
Almost correct	4	6	10
Skips generations	-	2	2
Mother to daughter	1	1	2
Germ	-	1	1
Not sure	9	16	25
Don't know	10	12	22
Not asked	3	6	9
Total	27	44	71

Different words including inherited, genetic and familial have been used to describe APKD (See Chapter 3). The next question for respondents was: 'is APKD a genetic disorder?'. The answers are shown in Tables F.22, F.23 and F.24.

Table F.22

First population: 'is APKD a genetic disorder?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Yes	10	20	12	42
Not sure	-	2	1	3
No	-	1	-	1
Don't know	4	5	14	23
Not asked	-	2	-	2
Total	14	30	27	71

Table F.23

First population: 'is APKD a genetic disorder?', subdivided by family history.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
Yes	2	3	20	17	42
Not sure	-	1	2	-	3
No	-	1	-	-	1
Don't know	4	5	7	7	23
Not asked	1	1	-	-	2
Total	7	11	29	24	71

Table F.24

First population: 'is APKD a genetic disorder?', subdivided by housing tenure.

	Owner-occupier	Tenant	Total
Yes	23	19	42
Not sure	-	3	3
No	-	1	1
Don't know	4	19	23
Not asked	-	2	2
Total	27	44	71

The question about whether the patient knew his or her own risk of inheriting APKD was not asked of 56 out of the 71 patients (79%) at this stage as it was clear to the researcher that the method of transmission was problematic and potentially distressing to

these patients. The answers, such as they are, are given in Tables F.25, F.26 and F.27. Only 3 patients knew the correct risk for them to inherit APKD.

Table F.25

First population: 'do you know the risk for you to inherit APKD?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Yes	-	2	1	3
No	7	5	-	12
Not asked	7	23	26	56
Total	14	30	27	71

Table F.26

First population: 'do you know the risk for you to inherit APKD?', subdivided by family history.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
Yes	-	-	2	1	3
No	-	-	6	6	12
Not asked	7	11	21	17	56
Total	7	11	29	24	71

Table F.27

First population: 'do you know the risk for you to inherit APKD?', subdivided by housing tenure.

	Owner-occupier	Tenant	Total
Yes	2	1	3
No	4	7	12
Not asked	21	35	56
Total	27	44	71

For the same reasons 18 patients were not asked about whether they knew the risk to their children. The results of this question are shown in Tables F.28, F.29 and F.30.

Table F.28

First population: 'do you know the risk for your children to inherit APKD?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Yes	2	8	7	17
No	9	15	12	36
Not asked	3	7	8	18
Total	14	30	27	71

Table F.29

First population: 'do you know the risk for your children to inherit APKD?', subdivided by family history.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
Yes	2	-	7	8	17
No	-	5	17	14	36
Not asked	5	6	5	2	18
Total	7	11	29	24	71

Table F.30

First population: 'do you know the risk for your children to inherit APKD?', subdivided by housing tenure.

	Owner-occupier	Tenant	Total
Yes	7	10	17
No	16	20	36
Not asked	4	14	18
Total	30	27	71

F.2.2 Score for knowledge of inheritance in questionnaire 1

Certain of the questions in questionnaire 1 about the genetic inheritance of APKD were used to form a composite score for the respondents' knowledge of genetics, formed as follows.

> For the answer 'yes' or 'it seems to be' to the question: 'Is APKD inherited?' 1 point was given. 59 respondents scored a point.
>
> For the answer 'yes' to: 'can APKD be passed on?' 1 point. 57 respondents scored a point.
>
> For the answer 'yes' to: 'is APKD genetic?' 1 point. 42 respondents scored a point.
>
> If the respondent now had all 3 points scored so far one extra point was given. 40 respondents scored an extra point.
>
> For the answer 'yes' to: 'does APKD run in the family?' 1 point. 48 respondents scored a point.

This gives a maximum of 5 points. A score of 4 or 5 represents good knowledge of the inheritable nature of APKD, even if the precise details were not known; a lower score indicates some uncertainty at least about the terminology used. The distributions of respondents by number of points, subdivided by sex and marital status, by family history and by housing tenure, are shown in Tables F.31, F.32 and F.33.

Table F.31

First population: scores for questions on knowledge of genetic inheritance of APKD in questionnaire 1 (KIS1), subdivided by sex and marital status.

	Single females	Married females	Males	Total
0 points	2	6	-	8
1 point	-	-	4	4
2 points	1	1	4	6
3 points	2	4	7	13
4 points	-	4	5	9
5 points	9	15	7	31
Total	14	30	27	71
Mean score	3.9	3.5	3.3	3.5

Table F.32

First population: scores for questions on knowledge of genetic inheritance of APKD in questionnaire 1 (KIS1), subdivided by family history.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
0 points	3	5	-	-	8
1 point	2	1	-	1	4
2 points	-	1	2	3	6
3 points	-	2	8	3	13
4 points	1	2	6	-	9
5 points	1	-	13	17	31
Total	7	11	29	24	71
Mean score	1.6	1.5	4.0	4.2	3.5

Table F.33

First population: scores for questions on knowledge of genetic inheritance of APKD in questionnaire 1 (KIS1), subdivided by housing tenure.

	Owner-occupier	Tenant	Total
0 points	2	6	8
1 point	-	4	4
2 points	2	4	6
3 points	1	12	13
4 points	5	4	9
5 points	17	14	31
Total	27	44	71
Mean score	4.1	3.0	3.5

Analysis using the GLIM system of the score for knowledge of inheritance, KIS1, using all the possible explanatory variables described in Section 9.13 and including the first population showed that only the factors to produce effects significant at a 1% probability level were family history and housing. Family history accounted for 41.7%

of the original variance, and housing an extra 7.4%, making 49.1% in all, quite a substantial reduction in the original variance.

Further analysis using also the scores for experience of genetic counselling (EGCS1 and EGCS2) and including the second population showed that again family history and housing were significant at a 1% level, explaining 48,3% of the original variance, but score EGCS1 explained a further 4,5%, significant at a 5%, though not at a 1%, probability level. The correlation coefficient between scores KIS1 and EGCS1 was 0.44, so by itself it would explain 19.1% of the variance, but this is reduced when this factor is brought in after other and more powerful factors.

The components of the mean score for the two populations are shown in Table F.34, and the mean scores for those with each combination of family history and housing, using the results from the first population, are shown in Table F.35. Those with higher grades of family history (2 and 3) have better knowledge of the genetic inheritance of APKD than those with lower grades, to the extent of about 2.5 points more, and those who are owner occupiers have better knowledge than tenants to the extent of about one point.

Table F.34

First and second populations: components of mean scores for knowledge of inheritance in questionnaire 1 (KIS1).

	Population	
Element	First	Second
Overall mean	3.46	3.55
Family history:		
Grade 0	−1.80	−1.28
Grade 1	−1.90	−1.78
Grade 2	+0.60	+0.54
Grade 3	+0.67	+0.41
Housing:		
Owner-occupier	+0.60	+0.62
Tenant	−0.37	−0.36
Per unit of EGCS1	-	+0.21

Table F.35

First population: mean scores for knowledge of inheritance in questionnaire 1 (KIS1), classified by family history and housing.

Family history	Owner occupier	Tenant
Grade 0	2.27	1.29
Grade 1	2.16	1.19
Grade 2	4.67	3.70
Grade 3	4.74	3.76

F.3 KNOWLEDGE OF INHERITANCE OF APKD: QUESTIONNAIRE 3, SECTION 5

F.3.1 Questions and basic results

In Section 5 of questionnaire 3 five questions were asked about the patients' understanding of the genetics of APKD. These were of 'multiple choice' form, in that the respondent was asked to select one from a specified set of answers. In the tables below, the correct answer is given first, though it was not always in this position in the questionnaire.

The first question was: 'how is APKD passed on?'. The replies, subdivided by sex and marital status and by age and number of children, are shown in Tables F.36 and F.37.

Table F.36

Third population: 'how is APKD passed on?', subdivided by sex and marital status

	Single females	Married females	Males	Total
From generation to generation	11	14	9	34
From female to female	-	2	-	2
From female to male	-	-	2	2
From male to female	-	-	1	1
From male to male	-	-	-	0
Some combination of the four above	1	1	-	2
Don't know or no reply	-	3	3	6
Total	12	20	15	47

Table F.37

Third population: 'how is APKD passed on?', subdivided by age and number of children.

	≤44, ≤2	≤44, ≥3	≥45, ≤2	≥45, ≥3	Total
From generation to generation	24	2	5	3	34
From female to female	-	1	-	1	2
From female to male	-	-	-	2	2
From male to female	1	-	-	-	1
From male to male	-	-	-	-	0
Some combination of the four above	2	-	-	-	2
Don't know or no reply	1	2	2	1	6
Total	28	5	7	7	47

The next two questions were: 'what is the risk of inheriting APKD?' and 'what is the risk of passing on APKD?'. The replies to both questions are shown in Tables F.38 and F.39.

Table F.38

Third population: subdivided by sex and marital status

	Single females	Married females	Males	Total
(a) 'what is the risk of inheriting APKD?'				
A big risk	4	4	4	12
A medium risk	8	13	4	25
A small risk	-	1	3	4
Don't know or no reply	-	2	4	6
(b) 'what is the risk of passing on APKD?'				
A big risk	5	4	4	13
A medium risk	5	11	6	22
A small risk	1	3	1	5
Don't know or no reply	1	2	4	7
Total	12	20	15	47

Table F.39

Third population: subdivided by age and number of children.

	≤44, ≤2	≤44, ≥3	≥45, ≤2	≥45, ≥3	Total
(a) 'what is the risk of inheriting APKD?'					
A big risk	7	2	1	1	12
A medium risk	19	1	3	2	25
A small risk	1	-	1	2	4
Don't know or no reply	1	2	1	2	6
(b) 'what is the risk of passing on APKD?'					
A big risk	10	-	2	1	13
A medium risk	15	1	2	4	22
A small risk	2	1	1	1	5
Don't know or no reply	1	3	2	1	7
Total	28	5	7	7	47

Respondents were then asked: 'when does APKD skip generations?'. The replies are shown in Tables F.40 and F.41.

Table F.40

Third population: 'when does APKD skip generations?', subdivided by sex and marital status

	Single females	Married females	Males	Total
Never	5	10	6	21
Sometimes	7	7	5	19
Always	-	-	-	0
Don't know or no reply	-	3	4	7
Total	12	20	15	47

Table F.41

Third population: 'when does APKD skip generations?', subdivided by age and number of children.

	≤44, ≤2	≤44, ≥3	≥45, ≤2	≥45, ≥3	Total
Never	18	-	1	2	21
Sometimes	9	3	4	3	19
Always	-	-	-	-	0
Don't know or no reply	1	2	2	2	7
Total	28	5	7	7	47

The final question in this section was: 'the risk of inheriting APKD is .. ?', and the possible replies were specific numbers. The replies are shown in Tables F.42 and F.43.

Table F.42

Third population: 'the chance of inheriting APKD is..?', subdivided by sex and marital status

	Single females	Married females	Males	Total
50-50	12	13	4	29
1 in 2	-	1	-	1
Both the above	-	2	2	4
1 in 20	-	1	3	4
1 in 4	-	1	-	1
Two inconsistent replies	-	-	1	1
Don't know or no reply	-	2	5	7
Total	12	20	15	47

Table F.43

Third population: 'the chance of inheriting APKD is..?', subdivided by age and number of children.

	≤44, ≤2	≤44, ≥3	≥45, ≤2	≥45, ≥3	Total
50-50	21	3	4	1	29
1 in 2	1	-	-	-	1
Both the above	3	-	-	1	4
1 in 20	-	-	2	2	4
1 in 4	1	-	-	-	1
Two inconsistent replies	-	-	-	1	1
Don't know or no reply	2	2	1	2	7
Total	28	5	7	7	47

F.3.2 Score for knowledge of inheritance in questionnaire 3, section 5

A composite score, denoted KIS2, was constructed from the answers to the questions in this Section of Questionnaire 3, as follows.

> For the answer 'from generation to generation' to: 'how is APKD passed on?' 2 points. 34 respondents scored 2 points.
>
> For the answers 'from female to female', 'from female to male' or 'from male to female', or some combination of these, to the same question: 'how is APKD passed on?' 1 point. 7 respondents scored 1 point. (This might have been the situation for their own family, although it is not true in general.)
>
> For the answer 'never' to: 'does APKD skip generations?' 1 point. 21 respondents scored 1 point.
>
> For the answers '50-50' or '1 in 2' or both to: 'what is the risk of inheriting APKD?' 1 point. 34 respondents scored 1 point.

This gave a maximum of four points. The distributions of respondents by number of points, subdivided by sex and marital status and by age and number of children, are shown in Tables F.44 and F.45.

Table F.44

Third population: scores for questions on knowledge of genetic inheritance of APKD (KIS2), subdivided by sex and marital status.

	Single females	Married females	Males	Total
0 points	-	2	3	5
1 point	-	1	1	2
2 points	1	2	5	8
3 points	6	8	2	16
4 points	5	7	4	16
Total	12	20	15	47
Mean score	3.3	2.8	2.2	2.8

Table F.45

Third population: scores for questions on knowledge of genetic inheritance of APKD (KIS2), subdivided by age and number of children.

	≤44, ≤2	≤44, ≥3	≥45, ≤2	≥45, ≥3	Total
0 points	1	2	1	1	5
1 point	-	-	1	1	2
2 points	2	1	1	4	8
3 points	10	2	4	-	16
4 points	15	-	-	1	16
Total	28	5	7	7	47
Mean score	3.4	1.6	2.1	1.9	2.8

Analysis of this score using the GLIM system and considering all the possible explanatory variables described in Section 9.13 showed that the variable that had the most significant effect was the factor combining age and number of children, which was significant at a 1% probability level. Once this was taken into account no other factor had a significant effect. This factor accounted for 33.7% of the original variance of the score.

The mean scores for those in each combination of age and number of children are shown in Table F.46. Those who have a larger number of children (3 or more) had less

knowledge of this aspect of genetic inheritance than those with fewer children or none. Those under 45 with fewer than 3 children had the best knowledge.

Table F.46

Third population: mean scores for knowledge of inheritance of APKD (KIS2), classified by age group and number of children.

	Number of children	
Age group	0-2	3 or more
up to 44	3.36	1.60
45 and over	2.14	1.86

F.4 KNOWLEDGE OF TRANSMISSION OF APKD: QUESTIONNAIRE 3, SECTION 5

F.4.1 Questions and basic results

In Section 5 of questionnaire 3 six further questions were asked about the patients' understanding of the risks of transmitting APKD, with one question about the presence of symptoms. These too were of multiple choice form, in this case asking whether the given statements were true or false.

The first two statements were: 'all children of a person with APKD will develop the condition' and 'on average half the children of a person with APKD will develop the condition'. The replies, subdivided by sex and marital status and by age and number of children, are shown in tables F.47 and F.48.

Table F.47

Third population: subdivided by sex and marital status

	Single females	Married females	Males	Total
(a) 'all children of a person with APKD will develop the condition'				
False	2	7	2	11
True	2	1	1	4
Don't know or no reply	8	12	12	32
(b) 'on average half the children of a person with APKD will develop the condition'				
True	8	12	7	27
False	1	2	2	5
Don't know or no reply	3	6	6	15
Total	12	20	15	47

Table F.48

Third population: subdivided by age and number of children.

	≤44, ≤2	≤44, ≥3	≥45, ≤2	≥45, ≥3	Total
(a) 'all children of a person with APKD will develop the condition'					
False	8	-	1	2	11
True	2	-	2	-	4
Don't know or no reply	18	5	4	5	32
(b) 'on average half the children of a person with APKD will develop the condition'					
True	21	3	-	3	27
False	2	-	2	1	5
Don't know or no reply	5	2	5	3	15
Total	28	5	7	7	47

The next two statements were: 'on average half the children of a person with APKD are at risk of developing the condition' and 'all children of a person with APKD are at risk of developing the condition'. The replies are shown in Tables F.49 and F.50.

Table F.49

Third population: subdivided by sex and marital status

	Single females	Married females	Males	Total
(a) 'on average half the children of a person with APKD are at risk of developing the condition'				
False	2	4	3	9
True	3	7	4	14
Don't know or no reply	7	9	8	24
(b) 'all children of a person with APKD are at risk of developing the condition'				
True	8	7	5	20
False	-	3	2	5
Don't know or no reply	4	10	8	22
Total	12	20	15	47

Table F.50

Third population: subdivided by age and number of children.

	≤44, ≤2	≤44, ≥3	≥45, ≤2	≥45, ≥3	Total
(a) 'on average half the children of a person with APKD are at risk of developing the condition'					
False	6	-	2	1	9
True	9	1	-	4	14
Don't know or no reply	13	4	5	2	24
(b) 'all children of a person with APKD are at risk of developing the condition'					
True	16	2	2	-	20
False	2	-	1	2	5
Don't know or no reply	10	3	4	5	22
Total	28	5	7	7	47

The next statement gave a choice of three mutually contradictory statements: 'a person with APKD sometimes (always/never) has a parent with APKD.'. Some respondents gave two replies as true. The replies are shown in Tables F.51 and F.52.

Table F.51

Third population: 'a person with APKD ... has a parent with APKD', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Always	10	13	6	29
Sometimes	1	1	4	6
Never	-	1	-	1
Contradictory	-	2	1	3
Don't know or no reply	1	3	4	8
Total	12	20	15	47

Table F.52

Third population: 'a person with APKD ... has a parent with APKD', subdivided by age and number of children.

	≤44, ≤2	≤44, ≥3	≥45, ≤2	≥45, ≥3	Total
Always	25	2	1	1	29
Sometimes	1	1	2	2	6
Never	-	1	-	-	1
Contradictory	1	-	-	2	3
Don't know or no reply	1	1	4	2	8
Total	28	5	7	7	47

The final statement in this section gave a choice of two mutually contradictory statements: 'APKD always (sometimes) has symptoms'. Some respondents gave both replies as true. The replies are shown in Tables F.53 and F.54.

Table F.53

Third population: 'APKD ... has symptoms', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Always	4	5	9	18
Sometimes	7	10	1	18
Contradictory	-	1	1	2
Don't know or no reply	1	4	4	9
Total	12	20	15	47

Table F.54

Third population: 'APKD ... has symptoms', subdivided by age and number of children.

	≤44, ≤2	≤44, ≥3	≥45, ≤2	≥45, ≥3	Total
Always	10	2	3	3	18
Sometimes	16	2	-	-	18
Contradictory	-	-	-	2	2
Don't know or no reply	2	1	4	2	9
Total	28	5	7	7	47

F.4.2 Score for knowledge of transmission in questionnaire 3, section 5

A composite score, denoted KIS3, was constructed from the answers to the questions on transmission in Section 5 of Questionnaire 3, as follows.

> For the answer 'false' to: 'is it true that all children of an affected parent will develop APKD?' 1 point. 35 respondents scored 1 point.
>
> For the answer 'true' to: 'is it true that half of the children of an affected parent will develop APKD?' 1 point. 27 respondents scored 1 point.
>
> For the answer 'false' to: 'is it true that half of the children of an affected parent are at risk of developing APKD?' 1 point. 25 respondents scored 1 point.
>
> For the answer 'true' to: 'is it true that all of the children of an affected parent are at risk of developing APKD?' 1 point. 20 respondents scored 1 point.
>
> For the answer 'true' to: 'is it true that a person with APKD always has a parent with APKD', 1 point. 32 respondents scored 1 point.

This gave a maximum of five points. The distributions of respondents by number of points, subdivided by sex and marital status and by age and number of children, are shown in Tables F.55 and F.56.

Table F.55

Third population: scores for questions on knowledge of transmission of APKD in questionnaire 3, section 5, (KIS3), subdivided by sex and marital status.

	Single females	Married females	Males	Total
0 points	1	3	4	8
1 point	-	-	-	0
2 points	1	2	5	8
3 points	4	7	2	13
4 points	1	5	-	6
5 points	5	3	4	12
Total	12	20	15	47
Mean score	3.6	3.0	2.4	3.0

Table F.56

Third population: scores for questions on knowledge of transmission of APKD in questionnaire 3, section 5, (KIS3), subdivided by age and number of children.

	≤44, ≤2	≤44, ≥3	≥45, ≤2	≥45, ≥3	Total
0 points	1	1	4	2	8
1 point	-	-	-	-	0
2 points	1	2	2	3	8
3 points	10	-	1	2	13
4 points	6	-	-	-	6
5 points	10	2	-	-	12
Total	28	5	7	7	47
Mean score	3.8	2.8	1.0	1.7	3.0

Analysis of this score using the GLIM system showed that the factor that had the most significant effect was again that combining age and number of children, which was significant at a 1% probability level. After this was taken into account the only other factor that had a significant effect was one of the scores for experience of genetic

counselling, EGCS2; the correlation coefficient between KIS3 and EGCS2 was 0.44, significantly different from zero at a 1% level. The combined factor accounted for 42.5% of the original variance, and EGCS2 for a further 6.0%, making a total of 48.5%.

The components of the mean score for those in each combination of age and number of children and for each unit of EGCS2 are shown in Table F.57. Those who were 45 or over had less knowledge of this aspect of genetic inheritance than those who were younger. Those under 45 with fewer than 3 children had the best knowledge.

Table F.57

Third population: components of mean scores for knowledge of transmission of APKD (KIS3).

Element	Component
Age and number of children:	
≤44, ≤2	3.28
≤44, ≥3	2.41
≥45, ≤2	0.72
≥45, ≥3	1.43
Per unit of EGCS2	+0.16

F.5 KNOWLEDGE OF INHERITANCE OF APKD: QUESTIONNAIRE 3, SECTION 1

F.5.1 Questions and basic results

Respondents were asked a variety of questions to ascertain their knowledge of APKD, the first of which was: 'how did you get APKD?'. The results, subdivided by sex and marital status and by age and number of children, are shown in Tables F.58 and F.59.

Table F.58

Third population: 'how did you get APKD?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Because it is hereditary	10	18	13	41
During pregnancy	-	1	-	1
Because of pain	-	-	1	1
Don't know	2	1	1	4
Total	12	20	15	47

Table F.59

Third population: 'how did you get APKD?', subdivided by age and number of children.

	≤44, ≤2	≤44, ≥3	≥45, ≤2	≥45, ≥3	Total
Because it is hereditary	25	4	7	5	41
During pregnancy	1	-	-	-	1
Because of pain	-	-	-	1	1
Don't know	2	1	-	1	4
Total	28	5	7	7	47

Respondents were asked how APKD is discovered in patients. The answers shown in Tables F.60 and F.61 reflect the different ways in which APKD came to be diagnosed.

Table F.60

Third population: 'how is APKD discovered?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
It is hereditary	9	6	1	16
Blood pressure	1	4	5	10
During pregnancy	-	6	-	6
By chance	1	1	3	5
Because of pain	-	-	3	3
From infection	-	1	2	3
Because of bleeding	1	1	-	2
Don't know	-	1	1	2
Total	12	20	15	47

Table F.61

Third population: 'how is APKD discovered?', subdivided by age and number of children.

	≤44, ≤2	≤44, ≥3	≥45, ≤2	≥45, ≥3	Total
It is hereditary	13	-	2	1	16
Blood pressure	4	3	1	2	10
During pregnancy	6	-	-	-	6
By chance	3	-	2	-	5
Because of pain	1	-	-	2	3
From infection	-	1	1	1	3
Because of bleeding	1	-	1	-	2
Don't know	-	1	-	1	2
Total	28	5	7	7	47

Respondents were asked whether APKD could be caught. Almost all the patients (43 out of 47 or 91%) knew that APKD could not be caught. Tables F.62 and F.63 show the results.

Table F.62

Third population: 'can you catch APKD?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
No	12	18	13	43
Yes	-	2	1	3
Don't know	-	-	1	1
Total	12	20	15	47

Table F.63

Third population: 'can you catch APKD?', subdivided by age and number of children.

	≤44, ≤2	≤44, ≥3	≥45, ≤2	≥45, ≥3	Total
No	25	5	7	6	43
Yes	3	-	-	-	3
Don't know	-	-	-	1	1
Total	28	5	7	7	47

Patients were asked whether APKD could be prevented. A majority (35 out of 47 or 74%) thought that APKD could not be prevented. The results are shown in Table F.64 and F.65.

Table F.64

Third population: 'can APKD be prevented?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
No	8	17	10	35
Yes	1	-	1	2
By not having children	3	3	3	9
Don't know	-	-	1	1
Total	12	20	15	47

Table F.65

Third population: 'can APKD be prevented?', subdivided by age and number of children.

	≤44, ≤2	≤44, ≥3	≥45, ≤2	≥45, ≥3	Total
No	20	4	5	6	35
Yes	1	-	1	-	2
By not having children	7	1	1	-	9
Don't know	-	-	-	1	1
Total	28	5	7	7	47

F.5.2 Score for knowledge of inheritance in questionnaire 3, section 1

These questions also formed the basis of a score, denoted KIS4, formed as follows.

For the answer 'hereditary' to: 'how did you get APKD?' 1 point. 41 respondents scored a point.

For the answer 'no' to: 'can you catch it?' 1 point. 43 respondents scored a point.

For the answers 'yes' or 'by having no children' to: 'can it be prevented?' 1 point. 11 respondents scored a point.

This gave a maximum of three points. The distributions of respondents by number of points, subdivided by sex and marital status and by age and number of children, are shown in Tables F.66 and F.67.

Table F.66

Third population: scores for questions in section 1 on knowledge of genetic inheritance of APKD (KIS4), subdivided by sex and marital status.

	Single females	Married females	Males	Total
0 points	-	-	-	0
1 point	2	4	3	9
2 points	6	13	9	28
3 points	4	3	3	10
Total	12	20	15	47
Mean score	2.2	2.0	2.0	2.0

Table F.67

Third population: scores for questions in section 1 on knowledge of genetic inheritance of APKD (KIS4), subdivided by age and number of children.

	≤44, ≤2	≤44, ≥3	≥45, ≤2	≥45, ≥3	Total
0 points	-	-	-	-	0
1 point	5	1	-	3	9
2 points	16	3	5	4	28
3 points	7	1	2	-	10
Total	28	5	7	7	47
Mean score	2.1	2.0	2.3	1.6	2.0

Analysis of this score using the GLIM system showed not one explanatory variable that had any significant effect.

F.6 TOTAL SCORES FOR KNOWLEDGE OF GENETIC INHERITANCE

Two further scores were formed as totals: the first, denoted KIS5, was formed as the total score in questionnaire 3, the sum of the number of points for the three sections described above, combined, i.e KIS2 plus KIS3 plus KIS4. The maximum score was 12 points. The final total score, denoted KIS6, was formed as the total score in the two questionnaires combined, the sum of the score for questionnaire 1 and the total score for questionnaire 3, i.e. KIS1 plus KIS5. The maximum score was 17 points.

The average scores for the questionnaire 3 total, KIS5, and for the grand total, KIS6, subdivided by sex and marital status, by family history and by age and number of children, are shown in Tables F.68, F.69 and F.70.

Table F.68

Third population: average scores for questions on knowledge of inheritance of APKD (KIS5 and KIS6), subdivided by sex and marital status.

	Single females	Married females	Males	Total
Total score in questionnaire 3, KIS5	9.1	7.8	6.6	7.7
Total score in questionnaires 1 and 3, KIS6	12.7	12.1	10.1	11.6

Table F.69

Third population: average scores for questions on knowledge of inheritance of APKD (KIS5 and KIS6), subdivided by family history.

	Grade 1	Grade 2	Grade 3	Total
Total score in questionnaire 3, KIS5	5.8	7.3	9.0	7.7
Total score in questionnaires 1 and 3, KIS6	7.0	11.3	13.5	11.6

Table F.70

Third population: average scores for questions on knowledge of inheritance of APKD (KIS5 and KIS6), subdivided by age and number of children.

	$\leq 44, \leq 2$	$\leq 44, \geq 3$	$\geq 45, \leq 2$	$\geq 45, \geq 3$	Total
Total score in questionnaire 3, KIS5	9.2	6.4	5.4	5.1	7.7
Total score in questionnaires 1 and 3, KIS6	13.1	10.4	9.4	8.7	11.6

It would be reasonable to expect that a factor that had been relevant in explaining the variation in a part of any total score might contribute also to the explanation of the total. Analysis of the first total score, KIS5, showed that only one factor had a significant influence, again that combining age and number of children, which explained 61.5% of the original variance. No other variable made any significant contribution.

The mean scores, classified by age and number of children, are shown in Table F.71. Again those under age 45 with fewer than 3 children have the highest score.

Table F.71

Third population: mean scores for knowledge of inheritance of APKD in questionnaire 3 (KIS5), classified by age and number of children.

	Number of children	
Age group	0-2	3 or more
up to 44	9.21	6.40
45 and over	5.43	5.14

Again it would be reasonable to expect that a factor that had been relevant in explaining the variation in a part of the total might contribute also to the total. Analysis of the final total score, KIS6, showed that the first two candidates were family history and the combined factor for age and number of children, both significant at a 1% probability level. No other factor had any significant effect; the experience score EGCS2 was next, but not at a 5% level.

The components for the two explanatory variables, relative to the mean score of 11.62, are shown in Table F.72, and the expected scores for the two variables are shown in Table F.73.

Table F.72

Third population: components of mean scores for total knowledge of inheritance of APKD (KIS6).

Element	Component
Overall mean	11.62
Family history:	
Grade 1	−4.00
Grade 2	−0.11
Grade 3	+1.34
Age and number of children:	
≤44, ≤2	+1.20
≤44, ≥3	−1.69
≥45, ≤2	−1.73
≥45, ≥3	−1.89

Table F.73

Third population: mean scores for total knowledge of inheritance of APKD (KIS6), classified by family history and by age and number of children.

	Age and number of children			
Family history	≤44, ≤2	≤44, ≥3	≥45, ≤2	≥45, ≥3
Grade 1	8.83	5.94	5.89	5.73
Grade 2	12.71	9.82	9.78	9.62
Grade 3	14.17	11.28	11.24	11.08

This analysis shows that family history is the strongest single factor overall and particularly of the larger population in questionnaire 1 in explaining variation in knowledge of genetic inheritance. Those with a stronger family history have a better appreciation of the genetics. Within the population in questionnaire 3 age and number of children were relevant. Those who ere aged less than 45 and had fewer then 3 children had the best knowledge.

F.7 CORRELATIONS BETWEEN SCORES

The (Pearson product-moment) correlation coefficients were calculated for each of the scores for knowledge of inheritance with each other, and with the scores for experience of genetic counselling (EGCS1 and EGCS2) and the scores for knowledge of the symptoms and treatment of APKD (KIS1 to KIS6). The results are shown in Table F.74. Necessarily these calculations are based on the third population, except where indicated.

Table F.74

Third population: correlation coefficients for scores for knowledge of genetic inheritance (KIS1 to KIS6).

	KIS1	KIS2	KIS3	KIS4	KIS5	KIS6
KIS1	1.00					
KIS2	0.03	1.00				
KIS3	0.15	0.72*	1.00			
KIS4	0.10	0.17	0.18	1.00		
KIS5	0.12	0.88#	0.93#	0.39#	1.00	
KIS6	0.52#	0.77#	0.86#	0.38#	0.91#	1.00
EGCS1	0.44[2]	0.24	0.38	0.03	0.33	0.41
EGCS2	0.21[2]	0.26	0.45*	0.09	0.39	0.40
KDS1	0.33[1]	−0.10	−0.03	0.21	−0.02	0.07
KDS2	0.06	0.59*	0.67*	0.13	0.67*	0.60*
KDS3	0.20	0.52*	0.62*	0.01	0.59*	0.59*
KDS4	0.11	0.59*	0.68*	0.09	0.67*	0.62*
KDS5	0.18	0.49*	0.61*	0.17	0.60*	0.59*

Notes:
[1] and [2] indicate that the correlation coefficients have been calculated using the first and second populations respectively.
# indicates that one score forms part of another, so a correlation is likely to be found.
* indicates that the coefficient is significantly different from zero at a 1% probability level.

Within the third population there was substantial correlation between the scores for knowledge of the disease (KDS2 and KDS3) and the factual knowledge of inheritance (KIS2 and KIS3), and the analysis in this Appendix and in Appendix E has shown that these scores are in each case mainly dependent on the combined variable for age and number of children. Family history has an effect on the score in questionnaire 1 (KIS1), and through that on the grand total score (KIS6). The score for the content of genetic counselling (EGCS2) is correlated with one of the scores for knowledge of inheritance (KIS3) and it was found to have some effect in the GLIM analysis.

APPENDIX G: RESULTS: RESULTS OF GENETIC COUNSELLING

G.1 Introduction

In this Appendix the results of the topics noted below of interest in genetic counselling are reported; the results are discussed in Chapter 14:

attitudes to having children;

attitudes to screening and testing of at risk relatives;

attitudes to testing of children;

outcomes of genetic counselling;

who should do genetic counselling.

G.2 Respondents' attitudes to having children

G.2.1 Numbers of children and influence of APKD thereon

Respondents were asked how many children they had had, and then how many children would like or would have liked to have had; the two answers were compared. The responses to these two questions, subdivided by sex and marital status, are shown in Tables G.1 and G.2; then the two answers are compared with each other in Table G.3.

There were 20 respondents who had two children, 14 who had more than two, and 31 who had fewer than two. Nevertheless 36 respondents would like to have two children; these included 10 single females and 13 married females.

Table G.3 shows that only one person would have liked fewer children than she (a married woman) already had. 29 respondents (out of 65 or 44%) were happy with the number of children they had, and 33 (out of 65 or 51%) would have liked more. In a few cases it was not appropriate to ask the respondent this question.

Table G.1

Second population: 'how many children have you had?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
None	13	2	2	17
1	1	10	3	14
2	-	9	11	20
3	-	2	5	7
4	-	2	4	6
5	-	1	-	1
Total	14	26	25	65

Table G.2

Second population: 'how many children would you like or have liked?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
None	-	1	-	1
1	-	-	1	1
2	10	13	13	36
3	2	5	4	11
4	-	3	4	7
5	-	1	-	1
More	-	1	1	2
Some	1	1	1	3
Not asked	1	1	1	3
Total	14	26	25	65

Table G.3

Second population: 'how many children would you like or have liked?', subdivided by 'how many children have you had?'.

	None	1	2	3	4	5	Total
None	-	1	-	-	-	-	1
1	-	1	-	-	-	-	1
2	11	10	15	-	-	-	36
3	2	1	2	6	-	-	11
4	-	-	1	-	6	-	7
5	-	-	-	-	-	1	1
More	-	1	1	-	-	-	2
Some	3	-	-	-	-	-	3
Not asked	1	-	1	1	-	-	3
Total	17	14	20	7	6	1	65

The answer to these two questions are shown in Tables G.4 and G.5 subdivided by severity of disease.

Table G.4

Second population: 'how many children have you had?', subdivided by severity of disease.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
None	-	6	6	5	17
1	2	6	-	6	14
2	3	1	8	8	20
3	-	-	5	2	7
4	-	-	1	5	6
5	-	-	-	1	1
Total	5	13	20	27	65

Table G.5

Second population: 'how many children would you like or have liked?', subdivided by severity of disease.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
None	-	-	-	1	1
1	-	-	-	1	1
2	3	12	6	15	26
3	1	1	7	2	11
4	-	-	2	5	7
5	-	-	-	1	1
More	1	-	1	-	2
Some	-	-	1	2	3
Not asked	-	-	3	-	3
Total	5	13	20	27	65

Among those with more than two children, all but one had obtained no school qualifications (education level 1), as shown in Table G.6.

Table G.6

Second population: 'how many children have you had?', subdivided by educational level.

	Level 1	Level 2	Level 3	Level 4	Total
None	6	4	5	2	17
1	6	1	6	1	14
2	13	2	2	3	20
3	6	-	1	-	7
4	6	-	-	-	6
5	1	-	-	-	1
Total	38	7	14	6	65

Respondents were asked whether their knowledge of APKD had affected their views about how many children they would like, as shown in Table G.7. 17 out of the 40

female respondents (42%) said that the decision about the number of children they would like had been affected by the knowledge of APKD were women, as opposed to only 6 out of 24 (25%) among the men. However the contrast, although suggestive, is not statistically significant.

Table G.7

Second population: 'has your knowledge of APKD affected your decision about how many children you would like?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Yes	6	11	6	23
No	8	15	19	42
Total	14	26	25	65

Respondents were asked to describe the effect that their knowledge of APKD had had on the number of children they had had or would have liked. The responses are shown in Table G.8.

Table G.8

Second population: 'how has your knowledge of APKD affected your decision about how many children you have had?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Sterilised or vasectomy*	1	5	2	8
To have fewer children	1	1	1	3
Have not yet made plans about children	-	3	1	4
Not to have children	1	-	-	1
Did not marry	1	-	-	1
Altered spacing of children	-	2	-	2
No decision made yet	2	-	2	4
Not affected decision	8	15	19	42
Total	14	26	25	65

Note *: one male had had a vasectomy; the wife of another had been sterilised.

G.2.2 Attitudes to voluntary childlessness, sterilisation, vasectomy and A.I.D.

Respondents were asked whether they would be prepared to consider various forms of action to limit their families: 'having no children', sterilisation or vasectomy (of themselves or their partner as appropriate), or A. I. D. (strictly appropriate only if the male partner is affected). The results are shown subdivided by sex and marital status in Tables G.9(a) to G.9(d) respectively and subdivided by severity of disease in Tables G.10(a) to G.10(d) respectively.

Table G.9

Second population: subdivided by sex and marital status.

	Single females	Married females	Males	Total
(a) 'would you consider having no children?'				
Yes	4	5	9	18
Perhaps	-	-	2	2
No	9	20	11	40
Not asked	1	1	3	5
(b) 'would you consider sterilisation?'				
Yes	6	16	5	27
After having a child	1	-	-	1
Perhaps	1	1	-	2
No	6	8	8	22
Not asked	-	1	12	13
(c) 'would you consider vasectomy?'				
Yes	1	7	11	19
Perhaps	-	1	2	3
No	7	14	11	32
Not asked	6	4	1	11
(d) 'would you consider A. I. D.?'				
Yes	-	1	2	3
Perhaps	-	1	5	6
No	8	17	16	41
Not asked	6	7	2	15
Total	14	26	25	65

Table G.10

Second population: subdivided by severity of disease.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
(a) 'would you consider having no children?'					
Yes	2	1	6	9	18
Perhaps	-	-	1	1	2
No	3	12	11	14	40
Not asked	-	-	2	3	5
(b) 'would you consider sterilisation?'					
Yes	2	4	8	13	27
Perhaps	-	1	-	1	2
After having had a child	-	1	-	-	1
No	1	7	9	5	22
Not asked	2	-	3	8	13
(c) 'would you consider vasectomy?'					
Yes	3	2	7	7	19
Perhaps	-	-	1	2	3
No	2	8	9	13	32
Not asked	-	3	3	5	11
(d) 'would you consider A. I. D.?'					
Yes	-	1	-	2	3
Perhaps	-	-	3	3	6
No	4	9	12	16	41
Not asked	1	3	5	6	15
Total	5	13	20	27	65

In each case there was a tendency for the more severely affected patients to say that they would consider limiting their family in the way suggested, though in no single case is the result statistically significant.

G.2.3 Score for attitude to having no more children.

A composite score, denoted ATCS, was formed to represent each respondent's attitude to having no more children as follows.

For the answer 'yes' to each of the questions listed below 2 points, and for the answer 'perhaps' 1 point.

	Yes	Perhaps
Would you consider having no children?	18	2
Would you consider sterilisation?	27	3
Would you consider vasectomy?	19	3
Would you consider A.I.D.?	3	6

The maximum score was 8 points, and the distribution of scores, subdivided by sex and marital status, is shown in Table G.11.

Table G.11

Second population: scores for questions on attitude to having no more children, subdivided by sex and marital status.

	Single females	Married females	Males	Total
0 points	5	9	11	25
1 point	1	-	-	1
2 points	4	7	2	13
3 points	1	1	2	4
4 points	3	5	4	12
5 points	-	-	2	2
6 points	-	4	2	6
7 points	-	-	1	1
8 points	-	-	1	1
Total	14	26	25	65
Mean score	1.7	2.3	2.5	2.3

Analysis of this score using the GLIM system, and considering to all the possible explanatory variables described in Section 9.13, and also all the constructed scores for

experience of genetic counselling described in Chapter 10, and for knowledge of all aspects of APKD described in Chapters 11 and 12, showed that not one explanatory variable had any significant bearing on the score representing the respondent's attitude to having no children.

G.3 Attitudes to screening and testing at risk relatives

G.3.1 Attitudes to screening at risk

Respondents were asked a number of questions that related to their attitude to the screening and testing of relatives who were at risk of APKD. The first question was whether those at risk should be told of their situation. The responses are shown in Table G.12. Almost all (59 out of 65 or 91%) thought that those at risk should be told, or were not sure (5 out of 65 or 8%), making a total of 64 out of 65 or 98%. Only one thought that they should not.

Table G.12

Second population: 'should those at risk be told of their risk?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Yes	14	25	19	59
Not sure	-	1	4	5
No	-	-	1	1
Total	14	26	25	65

Respondents were asked why they thought that those at risk should be told of their risk. There was a variety of answers, as shown in Table G.13.

Table G.13

Second population: 'why should those at risk be told of their risk?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
'So that they can be screened'	5	10	9	24
'So that they can have choices'	4	8	5	17
'So that they can plan their lives'	1	3	2	6
'So that they are prepared'	1	2	1	4
'They have to know'	2	-	1	3
'Because it is sensible'	-	-	2	2
'Because it relieves anxiety'	-	-	1	1
'Because it is appropriate'	-	1	-	1
Not sure	-	-	1	-
Not asked	1	2	3	6
Total	14	26	25	65

G.3.2 Attitudes to testing at risk

Respondents were asked whether they thought that those at risk should be tested. The responses are shown in Table G.14. Almost all (62 out of the 65 or 95%) thought that they should be tested. One male and one married female were unsure and one married female felt strongly that testing was the choice of the individual concerned and she could herself give no opinion. Not one was against.

Table G.14

Second population: 'should those at risk be tested?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Yes	14	24	24	62
Not sure	-	1	1	2
It is the choice of the individual	-	1	-	1
Total	14	26	25	65

The reasons that respondents gave to explain why they thought that those at risk should be tested are shown in Table G.15.

Table G.15

Second population: 'why should those at risk be tested', subdivided by sex and marital status.

	Single females	Married females	Males	Total
'So that they will know'	5	7	6	18
'So that they can plan their lives'	2	7	4	13
'So that they can be treated'	3	3	2	8
'So that they are aware'	1	2	3	6
'It depends on the individual'	1	-	4	5
'So that they can be watched'	1	3	-	4
'They need to know, even though it could upset them'	-	1	1	2
'Because it relieves anxiety'	-	-	1	1
'Because it is sensible'	-	-	1	1
Not asked	1	3	3	7
Total	14	26	25	65

G.4 Attitudes to testing children

G.4.1 Testing children at risk

Most respondents were asked whether they thought that their children should be tested; 10 respondents were not asked because they were either younger or older and the question was inappropriate. The answers are shown in Table G.16. Of those that were asked 43 out of 51 (84%) said 'yes' unconditionally, and a further 9 (18%) said 'yes' with qualifications. Two felt that it was the choice of the individual. Not one respondent was against.

Table G.16

Second population: 'should your children be tested?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Yes	2	21	20	43
Yes, but not currently applicable	6	1	1	8
Yes, but not too young	-	1	-	1
It is the choice of the individual	-	1	1	2
Not asked	6	2	2	10
Total	14	26	25	65

The reasons given by respondents for having their own children tested are shown in Table G.17.

Table G.17

Second population: 'why should your children be tested?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
'So that they can be looked after'	1	5	6	12
'So that they can plan their lives'	1	5	4	10
'So that they may know'	1	3	1	5
'So that I can know and they can be treated'	-	5	-	5
'It depends on the individual'	-	2	1	3
'To prevent APKD'	-	-	3	3
'So that they can be free of trouble'	-	1	2	3
'Better earlier than in their teens'	-	1	-	1
'When they are mature enough'	-	-	1	1
Not asked	11	4	7	22
Total	14	26	25	65

Respondents were asked whether or not to have their child tested was a difficult decision. 11 respondents (17%) said that it was, but 35 (54%) said that it was not, as shown in Table G.18.

Table G.18

Second population: 'was it a difficult decision to have your children tested?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Yes	1	4	6	11
No	1	19	15	35
Not asked	12	3	4	19
Total	14	26	25	65

G.4.2 Informing children at risk

Respondents were asked at what age they thought that children of an affected parent should be told of their risk. The results are shown in Table G.19.

Table G.19

Second population: 'at what age should children be told of their risk?', subdivided by sex and marital status.

Age	Single females	Married females	Males	Total
0-5	-	7	6	13
6-10	-	-	1	1
11-15	3	3	1	7
16	5	8	4	17
17	2	2	1	5
18	3	3	8	14
19-28	1	3	4	8
Total	14	26	25	65

Most respondents (54 out of 65 or 83%) thought that the at risk child should be tested between the ages of 16 and 20, as shown in Table G.20.

Table G.20

Second population: 'at what age should children at risk be tested for APKD?', subdivided by sex and marital status.

Age	Single females	Married females	Males	Total
0-5	-	5	4	9
6-10	-	-	-	0
11-15	-	-	1	1
16	6	5	7	18
17	2	3	3	8
18	5	7	7	19
20	1	6	2	9
28	-	-	1	1
Total	14	26	25	65

G.4.3 Dependency on other factors

It would have been interesting to investigate whether the attitudes of respondents to the screening and testing of those at risk and of children differed by any of the possible factors that could have been considered. However, when there is almost complete unanimity in the responses, almost all being in favour of informing, testing and screening, no further analysis is possible.

G.5 Outcomes of genetic counselling

G.5.1 Consequences of genetic counselling

Respondents were asked five questions about whether genetic counselling had helped them in specific ways. The numbers of respondents answering 'yes' to each of these questions are shown in Table G.21.

Table G.21

Second population: numbers of respondents answering 'yes' to questions commencing 'did genetic counselling ...' , subdivided by sex and marital status.

	Single females	Married females	Males	Total
... inform you about risk?	9	20	11	40
... help you to decide on your family?	3	7	4	14
... inform you about APKD?	2	3	4	9
... relieve stress?	2	3	-	5
... inform you about treatment?	2	2	1	5

G.5.2 Score for outcomes of genetic counselling

A score was formed from the questions relating to the outcomes of genetic counselling, denoted OGCS, and constructed as follows:

For the answer 'yes' to the question: 'Did genetic counselling relieve stress?' 1 point. 5 respondents scored a point.

For the answer 'yes' to the question: 'Did genetic counselling inform you about the risk?' 1 point. 40 respondents scored a point.

For the answer 'yes' to the question: 'Did genetic counselling help you to decide about your family?' 1 point. 14 respondents scored a point.

For the answer 'yes' to the question: 'Did genetic counselling inform you about APKD?' 1 point. 9 respondents scored a point.

For the answer 'yes' to the question: 'Did genetic counselling inform you about treatment?' 1 point. 5 respondents scored a point.

This gave a maximum of 5 points. The distribution of respondents by number of points, subdivided by sex and marital status, is shown in Table G.22.

Table G.22

Second population: scores for questions on outcome of genetic counselling (OGCS), subdivided by sex and marital status.

	Single females	Married females	Males	Total
0 points	5	6	14	25
1 point	6	11	5	22
2 points	1	6	3	10
3 points	-	1	3	4
4 points	-	1	-	1
5 points	2	1	-	3
Total	14	26	25	65
Mean score	1.3	1.3	0.8	1.1

Analysis of this score using the GLIM system showed that, of the possible explanatory variables described in Section 9.13, the only factor with any significance was that combining age and number of children, which was significant only at a 5% probability level.

However, when the scores for the experience of genetic counselling, EGCS1 and EGCS2, are introduced as possible factors, each is very significant by itself, but adding the other provides little improvement. The correlation coefficients of OGCS with these two scores are 0.60 and 0.65 respectively (see Table G.24). The higher of the two is that with EGCS2, which therefore gives the better fit. The model then takes the form of a simple linear regression, accounting for 42.5% of the original variance. The parameters are shown in Table G.23.

Table G.23

Parameters for score on outcome of genetic counselling (OGCS).

Element	Score
Constant	0.09
Per unit of EGCS1	+0.34

G.5.3 Correlation coefficients between scores

The (Pearson product-moment) correlation coefficients were calculated for each of the scores discussed in this Appendix with each other, with the scores for experience of genetic counselling (EGCS1 and EGCS2) and with all the scores for knowledge of APKD (KDS1 to KDS5 and KIS1 to KIS6). The results are shown in Table G.24. The calculations are based on the second or third populations as indicated.

Table G.24

Third population: correlation coefficients for scores for results of genetic counselling (ATCS and OGCS).

	ATCS	OGCS
ATCS[2]	1.00	
OGCS[2]	0.04	1.0
EGCS1[2]	0.02	0.60*
EGCS2[2]	0.17	0.65*
KDS1[2]	0.10	−0.01
KDS2[3]	-0.18	0.25
KDS3[3]	-0.18	0.27
KDS4[3]	-0.18	0.27
KDS5[3]	-0.16	0.20
KIS1[2]	0.07	0.16
KIS2[3]	-0.17	0.22
KIS3[3]	-0.13	0.42*
KIS4[3]	-0.03	0.02
KIS5[3]	-0.16	0.34
KIS6[3]	-0.14	0.34

Notes: [2] and [3] indicate that the correlation coefficients have been calculated using the second and third populations respectively.
* indicates that the coefficient is significantly different from zero at a 1% probability level.

The score for attitude to having children, ATCS, had no significant correlations with any other score. The score for the outcomes of genetic counselling, OGCS, had fairly strong correlations with the scores for the experience of genetic counselling (0.60 and 0.65), as noted in Section G.5.2, and also a correlation of 0.42 with knowledge of treatment, KIS3.

G.5.4 Decisions taken as a result of genetic counselling

Patients were asked whether they had taken decisions as a result of the information they had received in genetic counselling. Out of the 49 patients who had received genetic counselling, only 16 or 33% had taken decisions as a result of genetic counselling information compared with 33 (67%) who had not, as shown in Table G.25. Most of those who had taken decisions were married women (13 out of 16), or a majority of the married women counselled (13 out of 22 or 59%). By contrast, only one male (out of 17) and 2 single females (out of 10) had taken decisions. In a 2 by 2 contingency table, setting single females plus males versus married females and 'yes' versus 'no' among those who had taken decisions the difference is strongly significant (Fisher's exact test shows that the probability that 3 or fewer of the former category saying 'yes' is 0.0005).

Table G.25

Second population: 'have you taken any decisions as a result of genetic counselling?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Yes	2	13	1	16
No	8	9	16	33
No genetic counselling	4	4	8	16
Total	14	26	25	65

Of the 16 patients who had taken decisions following genetic counselling 8 were in severity grade 3 (out of 19 who had received genetic counselling), as shown in Table G.26; but 2 out of the 4 unaffected also had taken decisions.

Table G.26

Second population: 'have you taken any decisions as a result of genetic counselling?', subdivided by severity of disease.

	Grade 0	Grade 1	Grade 2	Grade 3	Total
Yes	2	2	4	8	16
No	2	11	9	11	33
No genetic counselling	1	-	7	8	16
Total	5	13	20	27	65

The decisions taken by patients are shown in Table G.27.

Table G.27

Second population: 'what decisions have you made as a result of genetic counselling?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Sterilisation or vasectomy	1	6	1	8
No more children	-	2	-	2
No children	-	1	-	1
Smaller family	1	1	-	2
Told child(ren)	-	1	-	1
Suggested child(ren) screened	-	2	-	2
No decisions	8	9	16	33
No genetic counselling	4	4	8	16
Total	14	26	25	65

G.6 Who should give genetic counselling?

G.6.1 Who should give genetic counselling?

Patients were asked what kind of person they would like to see giving genetic counselling. The results are shown in Table G.28.

Table G.28

Second population: numbers of respondents who thought that genetic counselling should be given by the stated category of person, subdivided by sex and marital status.

	Single females	Married females	Males	Total
Specialist genetic counsellor	10	20	17	47
Doctors in renal unit	10	16	18	44
GP	2	8	7	17
Nurse	1	-	1	2
Social Worker	-	1	-	1
Maximum	14	26	25	65

Respondents were also asked to name any other person or category of person who could give genetic counselling. This question did not yield many suggestions; only 15 out of 65 made a suggestion, as shown in Table G.29. The majority of patients had no suggestions. 1 patient suggested a parent or cousin, 4 patients suggested another affected patient, and 10 patients (8 female and 2 male) suggested 'someone like you', ie like the research worker.

Table G.29

Second population: 'whom else can you name who could give genetic counselling?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
'Someone like you'	3	5	2	10
'Someone who is affected'	1	2	1	4
Parents or cousins	1	-	-	1
No suggestions	9	19	22	50
Total	14	26	25	65

Patients were also asked whether they thought that no genetic counselling should be given. Not one patient out of the 65 believed that no genetic counselling should be given.

Respondents were asked about the sort of information that they would like in a genetic counselling service if it were to become available, or about the features that should characterise it. The results are shown in Table G.30.

Table G.30

Second population: 'if you have not had genetic counselling, what sort of information should be included in it, or what features should characterise it?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Information	3	5	11	19
Quiet	-	3	-	3
GP could be involved	-	1	2	3
Use of questionnaire	-	1	1	2
Use of Counsellor	-	-	2	2
Time	-	1	-	1
Group discussion	-	-	1	1
Not clinical	-	1	-	1
A good listener	-	-	1	1
Not asked	11	14	7	32
Total	14	26	25	65

G.6.2 Who should inform children?

Almost all the patients (63 out of 65 or 97%) thought that the parents should tell their children that they were at risk. One male was not asked this question. The results are shown in Table G.31.

Table G.31

Second population: 'who should tell children of their risk?', subdivided by sex and marital status.

	Single females	Married females	Males	Total
Parents	13	26	24	63
Doctor and parents	1	-	-	1
Not asked	-	-	1	1
Total	14	26	25	65