J Med Genet 1999;**36**:580–582

Genetic counselling: do we recognise and meet the consultands' agenda?

EDITOR—Agreeing the issues that are to be addressed is central to the process of genetic counselling. We undertook a study to document the consultand's agenda at the genetic clinic and whether the geneticist recognises that agenda. We also addressed whether failure to recognise the agenda is the prime factor when consultands think that issues important to them have been dealt with badly.

Ethical approval was given by the Newcastle and North Tyneside Health Authority Joint Ethics Committee to invite new consultands, over 18 years of age, attending a general genetic clinic to take part in this study. Consultands attending follow up appointments or those who had had a home visit before the hospital appointment were not included. Consultands were given an information sheet with details of the study and a verbal explanation on arrival at the clinic. Those wishing to take part were given a questionnaire to complete in the waiting room before seeing the geneticist. The first question was whether they had asked for the appointment or it had been suggested to them, and the second question asked if they knew the name of the medical condition they had come about. These two questions were followed by a list of issues (table 1) that they might want to address in the consultation. The consultands were asked to mark boxes indicating whether a topic was important, not important, or not applicable to them. There was a space at the end of the questionnaire to add further issues. The study design was based on self completion questionnaires because there are no spare rooms available at the clinics and therefore no privacy for interviews. The benefits of the method are that it is simple and does not intrude unduly on the consultation or the consultand's time. The major disadvantage is that the list one gives may influence the consultand's initial agenda.

The questionnaire related to a single counselling situation and so could be completed by a single person or several people. For example, a couple attending the clinic with an affected child were considered as one consultation and asked to complete the questionnaire together. However, when a number of family members at risk of developing the same condition were seen together each was given a questionnaire. When a couple was seen because one of them was at risk of developing a disorder they were asked to complete the questionnaire together.

After the consultation the same 12 statements were given to the geneticist (nurse or doctor) who was asked to mark whether they had felt an issue was important, not important, or not applicable in that consultation. If the geneticist marked a statement important they were asked to mark a box to indicate how well they felt they had addressed the issue, the options being: very well, well, neither badly nor well, badly, or very badly. The geneticist was also given space at the end of the questionnaire to add other information.

Table 1 List of issues on each of the questionnaires

(1)	I would like to know if I am going to get this condition.
(2)	I would like to know how likely my child(ren) are to get this condition.
(3)	I would like to know about treatments for this condition available at the moment.
(4)	I would like to know what is happening in the developments of new treatments for this condition.
(5)	I would like to know what will happen to someone with this condition as time goes by.
(6)	I would like to know if there is a medical or genetic test to see if I or others will get this condition.
(7)	I would like to know if there is a test for this condition in pregnancy.
(8)	I would like to know what causes this condition.
(9)	I would like to know a medical name for this condition.
(10)	I would like to be able to contact other families affected with this condition for support and advice.
(11)	I would like an explanation of the way this condition is passed on from one generation to the next.
(12)	I would like the information discussed today written down so that I can look at it again when I want to.

Four to five weeks after the clinic appointment the consultands were visited at home by AS. They were asked to complete a third questionnaire which had the same list but the statements that the consultands had thought important before their clinic appointment were highlighted. For these highlighted topics the consultands were asked to mark how well they felt the issue had been addressed using the same box system as the geneticist, so that direct comparisons could be made. There was also a recorded interview at this stage. The time delay of four to five weeks was chosen to ensure that they would have received the letter summarising the consultation. Comparison of responses between groups was analysed using the chi-squared test.

One hundred consultands were enrolled after approaching 110 consultands. The questionnaire was completed by 60 male/female couples and 40 individuals, 28 women and 12 men. A corresponding questionnaire was completed by the geneticist for each of the consultand questionnaires. Eighty four of the consultations were with doctors and 16 with nurses. The consultations were undertaken by seven doctors and three nurses.

Sixty three of the consultations were about childhood illness and 34 about adult onset disease. Three consultations did not fit into this simple classification; two were for infertility and one for genetic risk to a consanguineous couple. Thirty one of the respondents said that they had asked to be referred to the genetic clinic. Sixty four said it had been suggested to them and five did not complete the question.

The number of consultands who thought an issue important is shown in fig 1. The difference between consultand and geneticist thinking an issue important was significant ($p<0.001$) for recurrence risk, current treatments, future developments, prognosis, medical/genetic testing, prenatal diagnosis, cause, how the condition is inherited, and written information. Fig 2 shows the discrepancies between consultand and geneticist about whether an issue was important or not. The largest discrepancy (73%) between consultands and geneticists was that consultands rated treatment important more frequently than geneticists. While treatment and development of new treatment were clearly important to the consultands, they might not have expected them to be addressed at the genetic clinic if it had not been itemised in the questionnaire. It is debatable whether discussion of treatment should be part of the geneticists' remit. However, the geneticist should recognise the importance of this information in the consultand's decision making process, if only to clarify whether they will be addressing it or not, and if not perhaps making suggestions about how the consultand can obtain information.

Comparison of the responses of those who had requested the appointment with those to whom it had been suggested identified significant differences in response to two of the statements. Those who had requested the appointment were more interested in personal risk (16/31 *v* 19/64, $p<0.05$) and in support groups (16/31 *v* 6/64, $p<0.001$). When comparing consultations relating to adult onset disorders with childhood illness, personal risk (28/34 *v* 6/63, $p<0.001$), prognosis (32/34 *v* 44/63, $p<0.01$), and

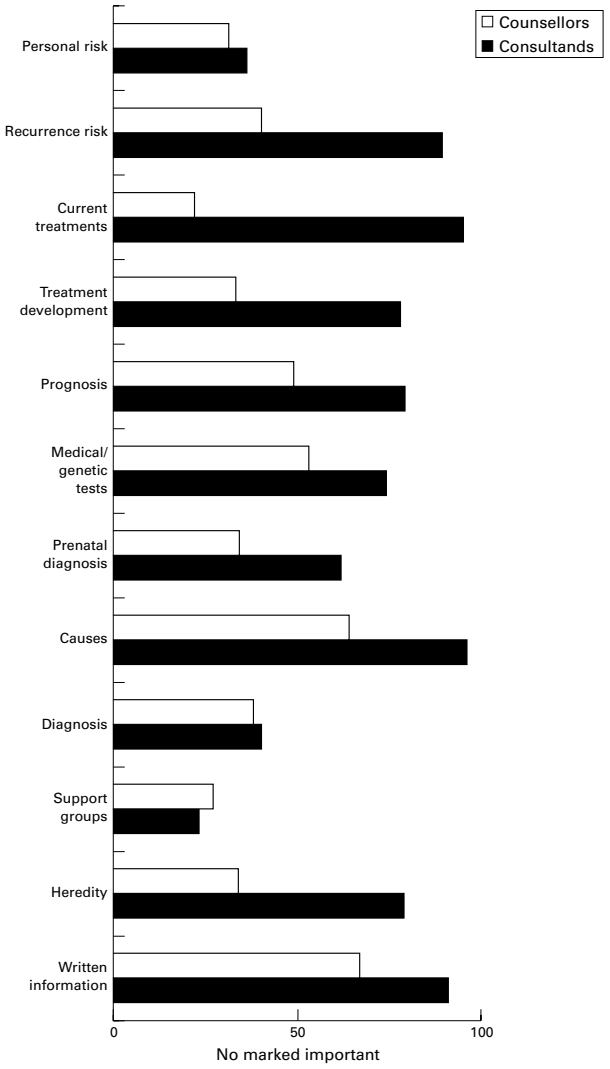

Figure 1 The number of times an issue was marked important by consultands and by geneticists. The number of questionnaires completed by each group was 100.

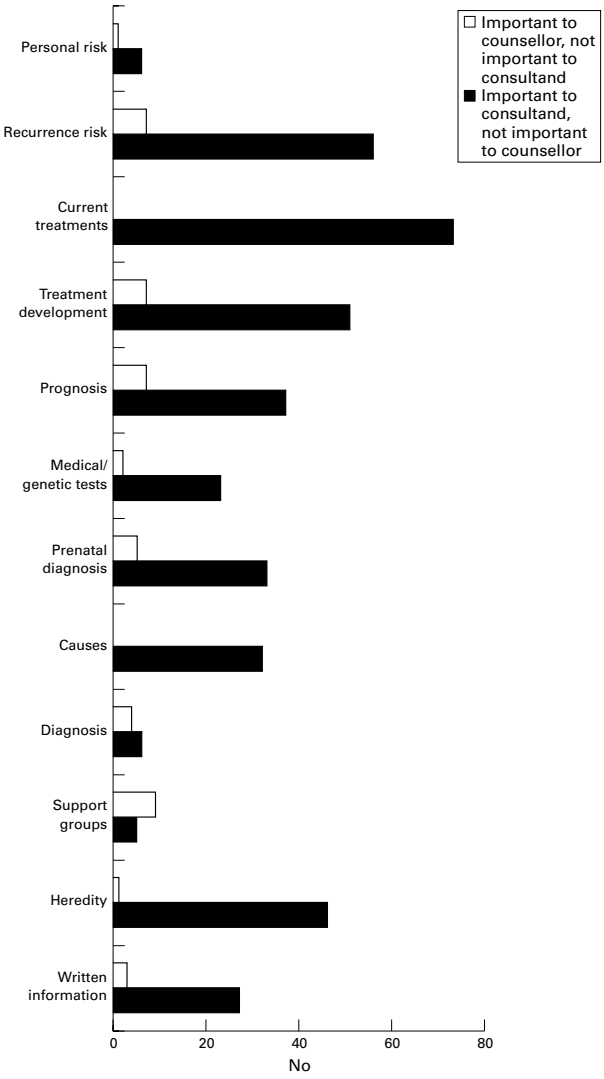

Figure 2 The discrepancies between consultand and geneticist for each issue on the questionnaire.

current treatments (30/34 *v* 45/63, $p<0.05$) were all marked important more often for the adult onset conditions. No issues were marked more important for the childhood onset conditions.

Follow up questionnaires and home interviews were completed for 69 of the 100 consultations. Forty nine of the first questionnaires had been completed by couples and 20 by individuals. Of the 49 initially completed by couples, both partners were present for 35 of the follow up questionnaires. There were no significant differences in responses to the first questionnaire between those who were followed up and those who either declined or could not be contacted. Five consultands (7%) marked that the geneticist had given information about one or more issues very badly and were dissatisfied; these consultations are outlined below.

A couple with a 2 year old child who has developmental delay and dysmorphic features had asked to be referred. No diagnosis was reached. The geneticist (doctor 4) had recognised the issues important to the couple but had marked that the information had been given neither badly nor well for both. The couple were disappointed that no diagnosis was forthcoming and, while they understood the geneticist might not have been able to give a diagnosis, they felt they were "fobbed off". A follow up appointment had been arranged.

Another couple had been referred to the clinic by their GP who thought the child had dysmorphic features. No diagnosis was made when they attended the consultation. The geneticist (doctor 4) recognised the important issues but was unaware of the couple's perception that they had been dealt with badly, marking all of these topics as having been dealt with well. At interview the couple said they did not think there was anything wrong with the child and were upset that it had been suggested. No follow up appointment was made.

A 44 year old lorry driver was referred by his GP. He thought he had been referred because of acne when in fact the diagnosis was hereditary haemorrhagic telangiectasia. His late mother had bled massively from a pulmonary arteriovenous malformation. The geneticist (doctor 7) had recognised the important issues and that their explanation had been poor. This consultand failed to attend a follow up appointment.

A 28 year old woman was referred because of a family history of colon cancer. The geneticist (nurse 2) recognised the important issues and was aware that she had dealt badly with three of them but marked that she had dealt with the fourth well. The consultand said she "was not told anything for sure....they couldn't be sure.... I just felt that if I hadn't gone, nothing would have changed and I

wouldn't have had all this worry". Screening was recommended and follow up arranged.

Another woman referred because of a family history of breast cancer attended with her husband. The geneticist (doctor 6) had not recognised the issues important to the couple. The consultands felt that too much time was spent talking about tests and genetics and not enough devoted to practical measures such as treatment and how the disease would affect someone. She said "(the geneticist) was going on about genes and DNA and what it would mean if I got a positive test, but I just wanted to know if I was going to get cancer and what I could do about it". Screening was recommended and no follow up appointment was made. It was only in this fifth consultation that failure to recognise part of the agenda was the crux of the problem.

In two of the consultations, those relating to the dysmorphic children, the geneticist had been unaware of the consultands' dissatisfaction. Thus one's subjective opinion of how well a consultation has gone cannot be relied upon. In none of these consultations where the consultands were dissatisfied was the geneticist giving bad news when one might have expected a "shoot the messenger" response. In four of the five consultations the consultands were left with an element of uncertainty. Failure to arrive at a diagnosis in the two children with delay meant that clear answers could not be given. Both of the women with a family history of cancer really wanted to know if they would get it or not rather than have a risk estimate, and although correct information was given and screening arranged both felt dissatisfied. In summary, failure to recognise the agenda was not a major contributor to patient dissatisfaction.

The most important finding of this study was that consultands want to know about available and developing treatments. This puts an onus on the geneticist to stay up to date with treatment modalities for the condition under discussion in any consultation and will require close links with colleagues in other specialities. Where the number of consultands with specific disorders is large enough, the most practical way of addressing treatment issues would be to hold joint clinics with the relevant specialist.

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J Med Genet 1999;**36**:582–583

Costello syndrome and rhabdomyosarcoma

EDITOR—Kerr *et al*[1] reported two children with Costello syndrome who also had embryonal rhabdomyosarcomas. I report a 14 year boy with Costello syndrome and an alveolar rhabdomyosarcoma.

The baby was born after 35 weeks gestation but weighed 3544 g. Polyhydramnios was present. At birth the infant appeared to be somewhat dysmorphic, was oedematous, and had low set ears. He required a respirator for five days. His oral intake was poor and at the age of 2 weeks he was admitted to hospital because of failure to thrive. At the age of 6 months, a diagnosis of alveolar rhabdomyosarcoma of the right foot was made. Treatment consisted of below the knee amputation and chemotherapy (doxorubicin, actinomycin D, vincristine, and cyclophosphamide). Continual follow up by oncologists has not indicated any recurrence of the rhabdomyosarcoma.

Subsequently, he was seen by various geneticists because of the following findings: nystagmus, a low set ear, midface hypoplasia, slightly coarse facial features, bitemporal narrowing, anteverted nares, broad nasal bridge, prominent pouting lower lip, marked joint laxity and hyperexten-

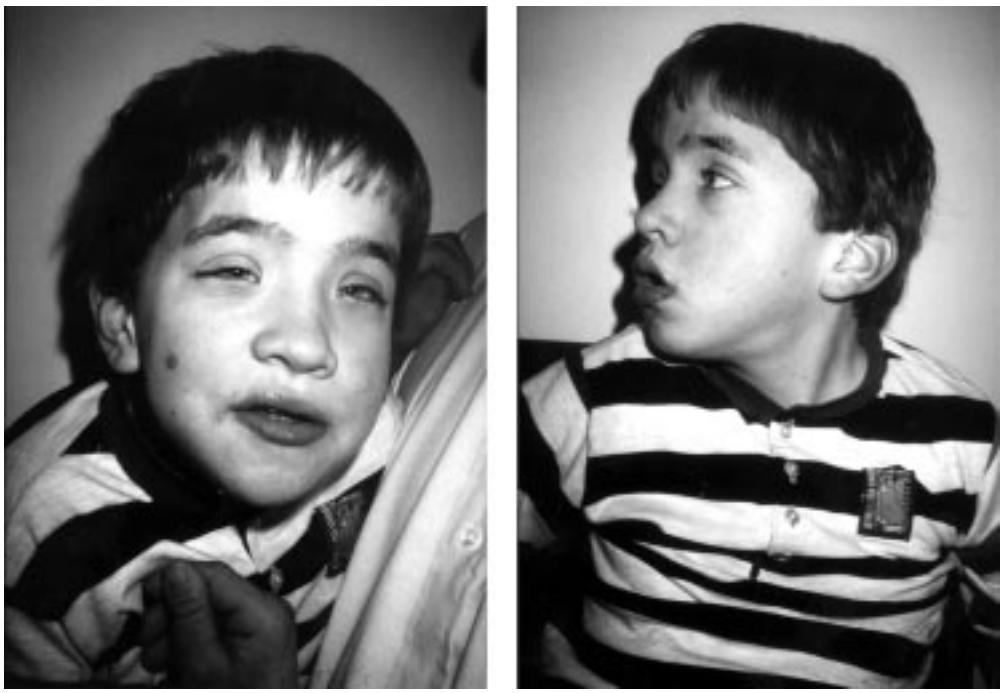

Figure 1 The proband.