considered as part of the original episode of care. For example, should the health authority pay once, twice, or three times for a patient who was originally admitted as an emergency with pleurisy, then readmitted for bronchoscopy, then admitted again for a chest drain procedure—all within the space of one month? In such cases providers and purchasers are likely to have different perspectives.

It seems likely that dealing with extracontractual referrals will always take a disproportionate amount of time because it involves a large number of cases, each handled individually. With the new emphasis on the role of health authorities in promoting the health of

populations, however, such referrals safeguard the rights of the individual against the rigidity of contracts that have been placed in the interest of the public health and provide a fascinating insight into the sociodynamics of referral patterns.

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Dealing with extracontractual referrals

J D Williamson

Abstract

Objective—To describe the mechanism established by Richmond, Twickenham, and Roehampton Health Authority to manage extracontractual referral requests made on behalf of its resident population and to examine its working in the first three months of the new arrangements.

Design—Description of the procedures for managing extracontractual referrals and the decisions made on requests submitted to the district health authority between 1 April and 30 June 1991.

Results—235 requests were submitted, 79 for emergency care. 156 requests were clearly for elective treatments and, of these, 61 were ultimately refused, 20 because the district health authority was not liable to pay. Of the remaining 41, 17 were appealed successfully and three unsuccessfully. More than half of the elective extracontractual referrals correctly submitted were to either orthopaedics, general surgery, oral surgery, gynaecology, or plastic surgery. Overall, the district health authority approved three quarters of the requests for which it would be financially liable; this was the predicted workload for the period.

Conclusions—The management of extracontractual referral requests is complex and time consuming for clinicians and managers alike. Patient choice is clearly being limited to some extent, but this is necessary if the number of requests is not to exceed the levels on which funding is based.

Introduction

The way that district health authorities react to requests for funding care in provider units not covered by a contract will determine whether money follows the patients or patients follow the money.

The government's view is quite clear. The general practitioner should be free to refer patients anywhere, secure in the knowledge that there is finance available. On the other hand, the district health authority is accountable for its expenditure and cannot merely reflect individual practitioners' wishes regardless of their effect on other patient services. The synthesis of these apparently contradictory statements means that patients can go where they like, but the health authority may not pay.

The government has made it clear that purchasing authorities should have firm contracts with all provider units offering a significant service to their resident population. This we have interpreted as a total workload of more than 50 inpatient or day cases (around

£100 000) a year, or fewer if the average cost per case exceeds £20 000. In most districts this would mean tying up about 95% of the health authority's financial resource in contracts, leaving only a small amount for extracontractual referrals.

Financial issues

Extracontractual referrals fall into three main categories: emergencies, elective secondary care, and elective tertiary care. Little research has been done, but a general survey of our last three years' data suggests that roughly a half of all referrals will be emergencies and a quarter will be referrals for tertiary care. The prudent purchasing authority will divide its extracontractual referral contingency (around 5%) to reflect this; thus less than 2% of its financial resource is likely to be available to support non-emergency (or elective) referrals by general practitioners. This marginal amount removes from both the funding authority and the referring clinician the flexibility implicit in "the market." The manoeuvrability that does exist may well be progressively reduced as the larger contractors offer either a lower cost per case or an improved quality of care in return for an increased workload. Financial and managerial pressure will inevitably squeeze extracontractual referrals as time goes by.

Three other issues may effect the availability of funding for extracontractual referrals. The first is the statutory requirement that purchasing authorities pay the excess costs of hospital care arranged by any general practitioner fundholder when these exceed £5000 for any individual patient in any one year. The number of such cases is not yet known. Secondly, there is the need to consider possible spending on rehabilitation or continuing care, with community care being implemented in 1993. Thirdly, there is the whole question of overspending. The existence of an agreement or contract between a purchaser or provider does not mean that all health needs are automatically met. Although many contracts have banded workload targets to take account of fluctuations in demand, it is by no means impossible that there should be unexpectedly high workloads which cannot be diverted to other contractors and will require additional funds. Because these three extras will probably have to be met from district reserves, the district contingency needs to be divided into several categories, none of which will be large (box). The risk of a queue developing for the money not allocated in the elective secondary care category is enormous.

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Categories of funding for extracontractual referrals

Emergency referrals

- Committed
- Unallocated

Referrals for elective tertiary care

- Committed
- Unallocated

Referrals for elective secondary care

- Committed
- Unallocated
- Reserve for overspends, community care, or expensive treatments.

Clearly, funds will be rationed. This raises the question of how requests to be funded are chosen. Is it simply a case of first come, first served, or will requests somehow be screened? The government suggests that the purchasing authority will not challenge the general practitioner's choice of provider unless it can be shown that the proposed referral is wholly unjustified on clinical grounds or that an alternative referral will be equally efficacious for the patient, taking into account the patient's wishes. Ignoring the final rider, which would mean that virtually every request would be honoured (if money was available) the arbiter of legitimacy of an extracontractual referral request lies in its clinical appropriateness. Presumably that is the basis on which purchasing authorities will devise methods for handling extracontractual referrals as sensitively as possible.

Emergency extracontractual referrals

Government policy, clinical need, patients' views, and logic all agree that emergency care must be given

whenever and wherever it is required, and it should not be delayed by a bureaucracy seeking prior funding. Occasionally the purchasing authority will be notified of an emergency referral and will be able to commit funds for it but more often than not its first notification will be receipt of an invoice. This must always be honoured provided that the patient is a district resident. I believe that providers are unlikely to cheat by admitting urgent referrals quickly and calling them emergencies so that payment can be guaranteed.

Last year 2.8% of what we now define as extracontractual inpatient episodes for residents of my district were classified as emergencies (and this year 2.5% of the budget was allocated for emergency referrals). These ranged from road traffic accidents through heart attacks and strokes to such things as pneumonia or ear infections suffered while staying with friends or relatives in distant parts of the country. These are obvious, but sometimes it is not clear whether the case was a true emergency. Where there is blatant misuse of the emergency procedure the purchasing authority is likely to refuse to pay the invoice, but where it is merely a matter of fine definition then it is more likely that the case will be managed as a post hoc request for an elective episode of care (fig 1). Grey areas like this should not be prejudiced by the fact that care has already started and so the shift from emergency to non-emergency status should not invalidate the claim for payment.

Non-emergency extracontractual referral

The first question we, as the purchasing authority, ask is, "Has care started?" This is absolutely critical because we would prefer to consider elective extracontractual referrals proactively, not reactively: we expect to be involved in the decision about referral before the provider unit takes any action.

The crucial issue is to decide when care actually starts. In our view this is when the provider unit first contacts the patient, either by offering an appointment or simply by acknowledging that the referral has been received. Such communication puts pressure on the purchasing authority to allow the release of money for that referral, and where there has been prior communication with the patient we may not fund that episode of care. In the early days we will try to be helpful to provider units who are not used to our policies, but in the long term we may well be more robust in implementing this rule. This resilient approach is essential if we are to develop discipline within the market. We do not, however, intend to make life difficult for those provider units that inadvertently blur the distinction between urgency and emergency, and in any case we would relax this prohibition for such cases. That is not to say that provider units will be allowed to abuse that loophole, for we will monitor very carefully the transfer between the emergency and non-emergency procedures. Where a provider unit seems to have a loose definition of emergency care there will have to be further discussions.

The next stage is to check the patient's address. The purchasing authority's money is reserved for its resident population, and sometimes this can be problematic. One obvious difficulty is when a patient moves into the district from another where funds have been committed for his or her care through a contract, but now this care requires extracontractual referral reserves to be used. National policy is that the postcode used by the patient determines the district health authority responsible for care, even if care is given in a hospital miles away. But what if patients have two homes in different districts—which is the residence? In the three cases we have encountered I have asked the family health services authority to identify the

FIG 1—Management of extracontractual referrals

NHS registered address and accepted this as the residence.

Provider units that cannot show evidence of a patient's address will not be funded for that care. It is therefore important for clinicians and hospital managers alike to recognise that poor administration at local level can deny them money to which they would otherwise be entitled.

The next question we ask, and this also applies to so called emergency cases we deem to be non-emergency, is whether or not the patient's general practitioner is a fundholder. If so, is the procedure to be performed on the approved list for payment? If the answer to both these questions is yes then the provider unit will have to seek post hoc funding from the general practitioner. This is likely to be difficult, and once again it is essential that clinicians and managers ensure that they do not embark on care that will not be funded because of administrative slip ups.

Up to this point there is no real rationing. The answer to the questions posed can only be yes or no, and the basis for each will be objective. Consequently, these procedures can be carried out by administrative or managerial staff. But now we come to clinical issues that need the input of an independent medical adviser like the director of public health medicine.

The first complex issue is whether or not the extracontractual referral is an unapproved tertiary referral. It may come as a surprise to recognise that consultants' referrals may be squeezed just as much as general practitioners'. In our contracts we have insisted that tertiary referrals by hospital consultants should conform whenever practicable to the contract package that we have negotiated.

One example of the difficulty unapproved tertiary referrals can create is in cardiovascular surgery. My district has a contract with several hospitals, but not with one particular centre that in the past dealt with very few patients in this district. Clinical preference in one of our acute provider units has switched from their usual centre, for which we have a contract, to this unit. Consequently, their referrals are deemed to be unapproved tertiary referrals, which could be rejected. The reason is that the government required purchasing authorities to continue previous referral patterns as far as possible and the change in referral pattern was not reflected in our contracts, particularly since this provider did not tell us about it. Clinicians must discuss tertiary referral policies with their managers to ensure that these are covered in future contract discussions. Otherwise consultants are bound by the arrangements made by the purchaser in its contracts portfolio. Once contracts are made it is pointless to demand in year additions because there is no money to support them unless there is another clinical reason for funding individual referrals.

Once the request has passed all the previous screens the only real barrier to approval is clinical need for referral to that particular unit. This is not a simple concept and the criteria of clinical justification have been derived with some difficulties. I use nine such criteria.

(1) *Is care continuing?* An extracontractual referral can be merely for outpatient care, but sometimes admission may become necessary. It could be ludicrous to insist on referring the patient to another hospital. Consequently, the fact that an extracontractual referral is for relatively cheap outpatient investigations should not blind purchasers to the realisation that they are probably accepting the need to fund any subsequent inpatient episode.

(2) *Is care by supradistrict services not available in the contracts portfolio?* In the first week I received a request

to fund an extracontractual referral of a baby to attend a national centre 200 miles distant for leg straightening. I know of no other centre doing this work and so readily agreed. For such referrals medical advisers need a reference file of the special skills and interests of specialists.

(3) *Is care urgent, though not emergency, with further referral likely to prejudice the clinical outcomes?* Such a case was a patient with bronchial carcinoma who required lung biopsy. The clinician had made tentative arrangements with a cardiovascular centre with which we do not have a contract and they could operate within one working week. I could have insisted that the case be referred to a contracted centre but realised that none could act as quickly. Because speed was essential to histopathological diagnosis I approved the referral.

(4) *Is care non-contracted but requested by a consultant from a contracted specialty in the same hospital, whereas referral to another provider would badly interrupt continuity of care?* I was informed of a woman with biliary cirrhosis who was suffering from menorrhagia. It was proposed that the gynaecologist in the same hospital should undertake the work even though this would be an extracontractual referral. Because of the possible association of her conditions or the effect of one on anaesthesia for the other I approved the referral.

(5) *Are the contractors unable to meet the patient's need, either because workload limits have been achieved or because services have been disrupted or withdrawn?* If a patient has clinical need and no contractors can help because they have run out of money or because they have had to close wards down to save money then clearly the purchasing authority has a responsibility to meet that patient's needs. Assuming there is money available in the authority's reserve then the extracontractual referral will be approved (with or without some claw back from any defaulting provider).

(6) *Has the patient been on a waiting list for a long time, with referral to another provider resulting in an unacceptable increase in the total time waited?* Though transitional, this is an important concept. We would not wish to increase waiting times unnecessarily. This can lead to some unfortunate consequences. One case I approved was a man who had been on a waiting list for four years for removal of his wisdom teeth. The hospital concerned was able to admit him shortly after implementing the new invoicing mechanisms; the earliest alternative was nine months after that. I felt that four years was long enough to wait for anything, but this dental operation cost £1400.

(7) *Are the patient's family or chief carers out of district?* Some of our residents are students. One asked her general practitioner to arrange an operation at her "local hospital" on Teeside and I readily agreed as that was where her family lived. Similarly, approval was given for an orthopaedic operation on a pensioner whose only living relative was a sister in the west country. Both cases required post-hospital care that was only reasonably available outside the district.

(8) *Has the patient had a bad experience with a contractor?* A general practitioner sought an extracontractual referral for a lady who had had an operation go wrong. A second operation had been performed in the same hospital by a locum surgeon who had subsequently moved to a different hospital. The patient had no confidence in the first hospital and wished to be transferred to the care of the intervening surgeon, to which I agreed. Negative health outcomes are not the only measures of a bad experience; environmental and attitudinal issues could be important.

(9) *Does the patient's disability require a particular response?* A paraplegic patient required some unrelated surgery that needed the special expertise of a spinal injuries unit. Although the procedure was readily available through our contracts portfolio, the care

necessitated by the disability was not and so the referral was approved.

Two points should be kept in mind. The first is that information is key—it will often be necessary for the medical advisers to discuss the case with the relevant clinicians. The second is that this listing may require extension and further revision as time goes on, particularly as purchasing authorities will wish to decide if, or in which circumstances, they wish to pay for such things as tattoo removal, gamete intrafallopian transfer (GIFT), or dental implants.

Once the referral is medically approved the contracts manager is informed and the provider notified. If the medical adviser decides that the extracontractual referral is really an unapproved tertiary referral or otherwise clinically unacceptable, the provider and the referrer are notified of that and the referral is refused after a short delay to allow appeal.

The appeals procedure

The procedure shown in the flow chart (fig 2) allows appeals by providers or referrers. Those by referrers usually go direct to the independent medical adviser. They can be successful on medical or financial grounds. Most doctors are not irrational; many appeals will succeed once the referrer has discussed the case with the independent medical adviser.

If, however, there seem to be no medical grounds for appeal the independent medical adviser will advise accordingly. Even then the contracts manager has discretion to approve an extracontractual referral. Before this can happen, though, he or she must be certain that the procedure represents value for money. Clearly, there should be a question mark over proposals to remove a wisdom tooth at a cost of £1400 or to treat psoriasis at an estimated cost of £8000—both these proposals have been put to us in the first month. Few clinicians or finance officers would disagree: it would be hard to justify a proposed cost of 150% or more of the local equivalent.

If the proposed extracontractual referral seems to be value for money then the key to approval will simply be the existence of unallocated, unreserved money. It is

important, however, to be clear which element of the fund is being tapped, and it is vital that we are clear on the periods of cash allocation. Should the contracts manager divide the unallocated pool monthly, fortnightly, weekly, or on some other basis? If the period is too long then there is a danger that there will be no funding available in the second half of the period, but if it is too short then referrals may be rejected when only a few days later they could have been approved. The management of cash flow is all important.

Experience of the first three months

I have given some examples of extracontractual referral requests I have received but this does not give the full picture. My district covers a relatively affluent London suburb used to sampling all the resources of the capital. As a consequence we have 44 contracts already signed, together with regular support from five special health authorities with whom we do not need a contract for previous workload levels. Despite this we expect about 700 extracontractual admissions a year.

In the first quarter of the 1991-2 year we were asked to fund 237 extracontractual referrals (table I), a third of the number predicted for the whole year. Of these,

TABLE I—Outcome of 235 extracontractual referrals requested in first three months

	Emergency (n=79)	Elective (n=156)
Accepted immediately	74	73
Accepted on appeal	5	17
Refused immediately		58
Patient defaulted		1
Care started		4
Non-resident		10
GP fundholder		6
Unapproved tertiary referral		2
Care available elsewhere		35
Refused after appeal		3
Funded by Department of Health		2
Poor value for money		1
No money available		0
Total accepted	79	90
Total refused		61
Not yet classified		5

62% were readily approved; some three weeks after the close of the quarter five await further information. This is just about the number we predicted on the assumption that all would culminate in inpatient care. At the end of the quarter 79 emergency referrals were reported, 47% of all approved extracontractual referrals. This figure is likely to rise as it seems to be taking several weeks to notify purchasers of emergency admissions.

An astonishing number of extracontractual referral requests submitted—4.2%—were for residents of other districts. Only one case was possibly deliberate, that of a young lady who, in requesting abortion, gave a temporary address in my district, though she actually lived in the midlands. More disconcerting are the six referrals refused because the referring general practitioner fundholder should be paying. I assume that submission to us was an administrative hiccup on the part of the provider rather than general practitioner fundholders trying to avoid their financial responsibilities.

We have been accused of being hawkish in our approach to care already started. In four such cases I approved the extracontractual referral after hearing of the providers' difficulties. Curiously, the other four cases were not argued.

Leaving aside the 20 extracontractual referrals for which my authority was not responsible, only 42% (63) of elective requests were initially refused. Half of these were appealed and three quarters succeeded. Those refused after appeal, all in the first month, were two

FIG 2—Management of appeals

cases funded directly by the Department of Health and three cases in which the referral represented poor value for money. All told, funding was finally refused for 31% (41) of elective extracontractual referrals for which the district health authority would be liable. It seems that local practitioners are reasonably content with the system, though it is possible that appeals will increase over time. Happily only one conversation with a referrer to date has been in any way acrimonious.

More than half of elective extracontractual referrals were arranged by general practitioners outside the district, mostly before the patient moved here. More than half the elective referrals were to either orthopaedics (31, 20%), general surgery (20, 13%), oral surgery (17, 11%), gynaecology (9, 6%), or plastic surgery (9, 6%).

Issues

My chief role in extracontractual referrals is to judge whether or not the care individual patients need is available within the contracts portfolio. I am not there to rule on the legitimacy of the decision to refer, merely the destination. None of the referrals I refused on clinical grounds were subsequently funded, even though that possibility is built into our procedures. This is perhaps understandable when we do not yet know how many cases we will be able to fund. Inevitably there will be some financial caution to avoid an overspend later in the year when faced with a backlog of bills for emergency work which we cannot and would not refuse to pay. Add to that the desire to discipline the main players in the new order and there is clear disincentive to be overflexible in the first weeks.

Nevertheless, even with a high rate of refusals, we have accepted the number of referrals we anticipated. There are three possible explanations for this. The first is the doubtful possibility that some general practitioners have been testing the new system by referring to unusual centres more frequently than before: to date, only two practices seem to be doing this but they may change during the year.

The second explanation is that provider units have suddenly developed an enthusiasm for dealing with those patients at the end of long waiting lists now that there is extra money in it. More than a dozen patients who have been waiting longer than five years were offered surgery in the first quarter. We shall probably have eliminated the current waiting list for extracontractual referrals by the end of this first financial year.

Thirdly, but most importantly, is the fact that some referrals are for outpatient work whereas all our planning is on the basis of inpatient activity. That did not help those specialties who care mainly for ambulant patients, such as dental care, sports medicine, psychiatry, and dermatology. Because we have no information on the ultimate cost consequences of outpatient care we have in these early days assumed an equivalence between these and inpatient episodes, but there will have to be a mid-year review to ensure that we are not restricting patient choice unreasonably.

One minor complication is the question of the district reserve for overspend against our block contracts. We seem to have a slight increase to date in workload over expectation—to an extent almost mirrored by a drop in general practitioner fundholder referrals. In this first year the general practitioners' fund was based on average rather than actual referral rates, and it looks as though some have been given more money than they needed. Consequently, the district health authority was given less. Caution is necessary if the purchaser is not to overspend.

Pricing extracontractual referrals is a quite distinct

problem. Some providers want to charge for each item of service separately whereas others prefer average specialty costs. Although separate charges are financially attractive, they are extremely difficult to manage because we have so little information to predict the true cost of any particular referral. It is also subject to abuse by retaining patients for financially rewarding outpatient care when they ought to be referred back to their general practitioner. Average specialty costs, on the other hand, disadvantage non-fundholding general practitioners wanting relatively minor work (such as removal of sebaceous cysts or wisdom teeth) in their patients since fundholders can demand the cheaper procedure price.

Technically, two issues have caused some difficulty. The first is the question of urgency versus emergency. I have tended to err on the side of declaring an urgent case an emergency; not everyone takes that view. The second issue is the two home family. I take the view that the patient's registered NHS address, not the patient's choice, is the key to purchaser responsibility; again, not everyone agrees.

Several problems should disappear during the year. The waiting list is one; others are the lack of knowledge of local procedures for approving extracontractual referrals, the tendency (now being stamped on by many purchasers) of starting care to try to encourage approval of referrals, and confusion over postcodes and general practitioner fundholders.

One provider issue seems likely to remain, though. At present I can judge an extracontractual referral only if I have information. But, for reasons of confidentiality, the information I need is not passed to the provider's contract office by the appointments staff and so I am not given it. All I can do is write to the referrer rejecting the referral, asking him or her to appeal if the case conforms to the nine criteria I have outlined above. The appeals that failed were by referring doctors who had not received such a letter, and it seems that referrers respond positively to clear local rules. Allowing time for appeal causes a delay of at least a week.

All of this brings us to the patient. My region has decided that the patient should not be involved in the administrative procedures relating to extracontractual referrals. This in effect means that neither provider or purchaser should contact the patient until a referral is approved. If it is refused then in theory the purchaser could notify the patient, but under our system it is not entirely clear at which point that should be. This issue needs early resolution.

To make sense of extracontractual referrals in future we need a great deal more information than is currently available, in terms of both referral rates and types of care given. We need to discuss with clinicians not only their referral patterns but also the specialisms available and those conditions which ought to be limited (either completely or to block contracts). Such discussion is too subjective without better data, more research into the course of an outpatient referral, a directory of special interests and skills in provider units, and outcome assessments of alternative treatments. None of this will be available quickly.

Meanwhile purchasers' financial advisers have to balance the books somehow. They will have to do that by processes previously inimical to patient care, and experience in my district suggests that we tend to be overcautious in the early days. Even so problems may not be avoided. Whereas in the past patients could go to another centre if their local hospital could not manage their care, the new system effectively cash limits work done for patients. Once the money has gone it is theoretically pointless to look for a provider who can accept the care. Capitation makes no allowance for higher than average referral demands, and it

will be interesting to see how the reformed NHS deals with this question. This is yet another reason for our erring on the side of financial caution.

Conclusion

We have to ask ourselves whether or not we are really enhancing patient choice and using money to the best effect. What is absolutely certain is that we are spending a lot of management and clinical time making

decisions on a relatively small number of patients. This time may not be used to its best advantage. There are benefits from NHS reforms, but billing may not be one of them. There are other methods by which provider units could have been recompensed or sanctioned for better or worse than average performance. True, we gain a lot of information about patient care within the service and we have been stimulated to find out what is available, but patient choice could easily be progressively restricted in future.

Screening for carriers of cystic fibrosis through primary health care services

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Abstract

Objective—To evaluate the uptake of cystic fibrosis carrier testing offered through primary health care services.

Design—Carrier testing for cystic fibrosis was offered to patients of reproductive age through primary health care services.

Setting—Three general practice surgeries and four family planning clinics in South West Hertfordshire District Health Authority.

Subjects—Over 1000 patients aged 16-44 attending two general practices and four family planning clinics and a stratified random sample of patients aged 16-44 from one general practice's age-sex register.

Results—When screening was offered opportunistically the uptake was 66% in general practice and 87% in family planning clinics. Ten per cent of those offered a screening appointment by letter took up the invitation. Of the screened population, 76% had previously heard of cystic fibrosis, 35% realised it is inherited, and 18% realised that carriers need not have any family history. If they found themselves in an "at risk" partnership 39% would consider not having children and 26% would consider terminating an affected pregnancy, but in each case most people were unsure how they would react.

Conclusions—Most people offered a cystic fibrosis test opportunistically wish to be tested, and the responses of those tested indicate that knowledge of carrier state would be considered in future reproductive decisions.

Introduction

Cystic fibrosis is a severe, autosomal recessive disease affecting one in 2500 live births in the British population. The clinical expression of cystic fibrosis is variable, although most patients show progressive pulmonary disease and pancreatic insufficiency with malabsorption. The average life expectancy for people with cystic fibrosis who were listed on the British Paediatric Association's register between 1977 and 1985 was 20 years, but these data relate to patients who were first treated more than 20 years ago and there have been considerable advances in treatment since then.¹ The carrier frequency in the British population is approximately one in 25; as for all autosomal recessive disorders, most infants with cystic fibrosis are born into families with no previous history of the disorder. A sizable reduction in the number of new cases of cystic fibrosis can only come from a programme of early identification of heterozygotes followed by prenatal diagnosis and selective termination if requested.

The gene mutated in cystic fibrosis has been shown to code for the cystic fibrosis transmembrane regulator.^{2,4} A three base pair deletion at position 508 of the protein accounts for about 78% of cystic fibrosis mutations in the British population.⁵ Two point mutations, G551D and R553X,⁶ account for a further 4% of cystic fibrosis mutations (unpublished data from this laboratory). These three mutations can be detected simultaneously by using the polymerase chain reaction, which will identify approximately 82% of carriers in our population. A simple mouthwash test serves as a reliable method of obtaining DNA for such an analysis.

Not all mutations within the cystic fibrosis gene have yet been identified, and many occur in only a few people. It may not be economically possible to achieve a much higher rate of carrier detection on a large scale. About two thirds of "at risk" couples can already be identified by using the simple test described above, so we carried out a pilot study of one approach to population screening for carrier state.

There are several approaches to carrier screening in the community, each with advantages and disadvantages (table 1).⁷ As cystic fibrosis carrier state has no known harmful effect on the personal health of the carrier it could be argued that screening should be offered to couples planning a pregnancy rather than to individuals. We chose to study pre-pregnancy carrier testing offered through community health services (general practitioners and family planning clinics). It is difficult to envisage how testing could be offered either opportunistically or by letter to "couples" through these services and we therefore elected to offer screening to any person of reproductive age who requested testing after they had been provided with an information leaflet and had a short discussion with a counsellor. We thought that by offering screening to people of reproductive age the carriers could be counselled at length, would have time to learn about cystic fibrosis and carrier state, and would retain a full range of reproductive options. The approach we have adopted is one of two recommended for pilot studies by the Cystic Fibrosis Research Trust (United Kingdom). It is important that such pilot studies are carried out in advance of widescale testing, whether by public bodies or commercial bodies, as such testing is not assessed for effectiveness or impact on those tested and counselled. We report the results of tests on 1000 people offered screening in three ways through primary health care facilities in south west Hertfordshire.

Methods

Subjects were recruited for screening from four health service family planning clinics and three general

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