Antenatal screening for haemoglobinopathies in primary care:

a whole system participatory action research project

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ABSTRACT

Background

The usual system for antenatal screening for haemoglobinopathies permits termination only late in the second trimester of pregnancy.

Aim

To evaluate a system where pregnant women are screened in general practice, and to develop a model of care pathway or whole system research able to bring into view unexpected effects of health service innovation.

Design of study

A whole system participatory action research approach was used. Six purposefully chosen general practices screened women who attended with a new pregnancy. Data of gestational age of screening were compared with two control groups. Qualitative data were gathered through workshops, interviews and feedback to the project steering group. At facilitated annual workshops participants from all parts of the care pathway produced a consensus about the meaning of the data as a whole.

Setting

Six general practices in north London.

Method

A whole system participatory action research approach allowed stakeholders from throughout the care pathway to pilot the innovation and reflect on the meaning and significance of quantitative and qualitative data.

Results

The gestational age of screening in general practice was 4.1 weeks earlier (95% confidence interval (CI) = 3.41 to 4.68) than in hospital clinics ($P<0.001$), and 2.9 weeks earlier (95% CI = 2.07 to 3.65) than in community midwife clinics ($P<0.001$). However, only 35% of pregnant women in the study were screened in the practices. Changes required throughout the whole care pathway make wider implementation more difficult than at first realised. The cost within general practice is greater than initially appreciated owing to a perceived need to provide counselling about other issues at the same time. Practitioners considered that other ways of early screening should be explored, including preconceptual screening. The research approach was able to bring into view unexpected effects of the innovation, but health workers were unfamiliar with the participatory processes.

Conclusion

Antenatal screening for haemoglobinopathies in general practice lowers the gestational age at which an at-risk pregnancy can be identified. However, widespread implementation of such screening may be too difficult.

Keywords

antenatal diagnosis; participatory action research; sickle cell anaemia; thalassaemia.

INTRODUCTION

Early antenatal diagnosis of a fetus at risk of a major haemoglobinopathy, such as sickle cell anaemia or β-thalassaemia major, is essential if pregnant women and their partners are to have sufficient time to think through the options available to them. Usually the heterozygous state for these conditions does not give rise to clinical symptoms but the homozygous and compound heterozygous state frequently results in severe, debilitating lifelong-illness. Audit[1] from the UK Confidential Enquiry into Counselling for Genetic Disorders between 1990 and 1994[2] showed that only half of pregnancies (68/138) affected by β-thalassaemia major were identified before delivery.

In 1993 the Department of Health's Standing Medical Advisory Committee recommended that universal antenatal screening for sickle cell and thalassaemia should be provided in areas where the antenatal population contains 15% or more women from minority ethnic groups — all women in such areas should be offered screening irrespective of

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their ethnic origin.[3] This screening usually takes place at first hospital attendance, roughly between 12- and 20-weeks gestation of pregnancy. It takes 7 weeks from the taking of a screening sample from the mother to the earliest date that termination of an affected pregnancy can be offered; this period allows for the maternal test result, partner screening, counselling, and arrangement of the operation. The usual system of screening, therefore, permits termination only late in the second trimester of pregnancy.

In 1995 a randomised controlled trial recruited a quarter of practices in north London (26/93) to pilot haemoglobinopathy screening in the antenatal period.[4] The rationale for this was that most pregnant women present in general practice several weeks before they are seen in a hospital antenatal booking clinic. Screening in general practice, therefore, might avoid late termination of pregnancy. Intervention practices received educational materials and training from an experienced facilitator. The number of requests for screening received by the laboratory from both intervention and control practices was measured. The study showed an increase in screening tests by general practices, but not whether pregnant women overall were actually screened at an earlier stage. Many of the 26 practices that expressed initial interest failed to undertake any screening at all; 56% of the observed increase in GP screening was single-handedly due to three motivated practitioners. There was no evidence of enthusiasm to further progress the new system for screening.

From a pregnant woman's perspective, the acceptability of prenatal screening in general practice can be inferred from a study of screening for cystic fibrosis in eight general practices in northwest England (total list size 42 000). In that study 529 (84.9%) women accepted the test and 97% of these subsequently felt that they had made the right decision.[5]

We designed the antenatal screening for haemoglobinopathies project to further examine the feasibility of antenatal screening for haemoglobinopathies in general practice. Brent and Harrow Health Authority was adjacent to the area of the 1995 study and also falls into the criteria for universal screening. The London boroughs of Brent and Harrow have almost 50% and 23% minority ethnic populations, respectively. Universal antenatal screening was implemented at Central Middlesex Hospital in 1979, and in 1998 Brent and Harrow Health Authority extended its service to Northwick Park Hospital, thereby having a hospital-based universal antenatal screening policy in the two local hospitals.

The aim of our project was, first, to assess whether general practice screening reduces the gestational age of antenatal screening and, second, to identify implications of this new way of screening for different professions throughout the whole care pathway. As a purely quantitative or qualitative study was unlikely to achieve these two aims concurrently, we devised a research approach that used quantitative and qualitative methods and set these inside a framework of whole system participatory action research.[6,7]

How this fits in

There remains a need to identify, at an early stage, those pregnancies that are at risk of a major haemoglobinopathy. Both hospital and general practice systems for antenatal screening have disadvantages, and other approaches need to be considered. Whole system participatory action research shows promise at illuminating unexpected consequences of care pathway innovation.

METHOD

A survey of 123 practitioners in the health authority identified 42 general practices interested in the project. Six general practices were chosen from these to reflect a range of practices (that is, small and large, and those relating to different hospitals and with different previous experience of antenatal screening for haemoglobinopathies). The practices were supported in a similar way to that of the 1995 study — a facilitator, with specially prepared materials, assisted participating practices in screening pregnant women at first presentation in general practice.

Pregnant women presenting in these practices between July 1999 and October 2000 were recruited opportunistically (241 women), and given information about the project. At next appointment they were counselled (usually by a practice nurse or GP) and consent was obtained. A specially designed form for the screening blood tests recorded the date of the last menstrual period and the date of venesection (to calculate gestational age of screening). Excluded from the analysis were outliers where the gestational age was below 4 weeks or above 26 weeks (n = 10), and those whose forms had incomplete data (n = 58). Different practices started and ended recruitment at different times, depending on their readiness to take part.

The first control group comprised 276 women at their first antenatal outpatient appointment at two local hospitals between 18 August 1999 and 27

September 2000. The second control group comprised 131 pregnant women attending community midwife clinics in Harrow between 4 April 2000 and 29 September 2000. From the control groups 28 outliers and those with incomplete data were excluded. Calculation of gestational age was made by subtracting the date of screening from the date of last menstrual period as recorded on the laboratory report. Table 1 shows the range of coverage by the practices.

Random checks were made for errors in data recording on the data collection forms and in data entry onto the computer. Data were triangulated between researcher records, practice records analysed at the time, and retrospective audit from practice computers (2004).

Roughly 20–30 participants from different parts of the care pathway reflected on data and their own experience of the project at annual 'whole system' workshops. Through small group reflections on project data and large group feedback, they derived a shared understanding about the meaning of the whole project. We use the term 'system' here in the sense described by Pratt as the 'people and organisations that connect around a shared purpose';[8] this includes support disciplines not usually considered to be a part of the care pathway, such as phlebotomists. The word 'whole' is meant to imply that all relevant perspectives from the system were considered. The disciplines considered to be part of the 'whole system' included GPs and practice nurses, obstetric and midwifery staff, laboratory technicians, phlebotomists, and specialist nurses from the Brent Sickle Cell and Thalassaemia Centre. Phlebotomists were not involved at the outset and this had an impact on later events, as detailed in the results section. Participation in research of people from the whole system of care means that this study is an example of whole system[8,9] participatory action research.[5,9] Such methods have increasingly been used in the UK health service.[8–11]

Representatives of the disciplines involved attended a steering group that met quarterly. A small project team met monthly. A facilitator undertook the necessary fieldwork.

We recognised that the quantitative data did not have the power to reveal the attitudinal and systemic obstacles to widespread screening within general practice, so the following methods were added:

- At an open meeting for the 42 interested general practices (April 1999) the practical demands of the project were discussed in small and large groups. This revealed that only 15 practices considered that they had adequate resources (such as midwife contact, IT skills) to take part in the project. From these 15, six practices were purposefully chosen;
- At a workshop for the six pilot practices (May 1999), through small group discussion and feedback, the practices identified needs for the development of their practice systems, specially designed blood forms, explanatory literature for staff and pregnant women, and in-practice workshops;
- Through two public meetings and 10 interviews with community leaders, the project facilitator recorded perceptions of the acceptability of the proposed new system (throughout 1999);
- Informal feedback about project literature was given to the facilitator who visited practices when needed — this ranged from almost weekly visits to them being undertaken every few months (1999);
- Annual whole system workshops (described above) generated new knowledge through facilitated reflection on data and health professionals' own experiences (November 1999 and November 2000);
- An independent external evaluator shadowed various project members and interviewed key informants.[12]

Table 1. Women screened in different locations.

Location	Registered practice population in 2000	Total pregnant women in 2000 (per 1000 practice population)[a]	Women screened in the practice (%)
Practice 1	11 637	236 (20.3)	5 (3)
Practice 2	5000	71 (14.2)	82 (86)
Practice 3	9296	117 (12.6)	33 (73)
Practice 4	3258	60 (18.4)	38 (63)
Practice 5	11 821	150 (12.7)	42 (24)
Practice 6	17 110	213 (12.4)	41 (26)
Total practice	58 122	847 (14.6)	241 (35)

[a]*The 2000 birth rate per 1000 total population in England and Wales was 11.4. (Source: http://www.statistics.gov.uk/downloads/theme_population/Fm1_29/FM1_29_V3.pdf Table 1.1 page 1).*

RESULTS

Of the 173 pregnant women screened in general practice, 15 were found to have abnormal haemoglobin. Eleven of their partners were tested, none of whom had abnormal haemoglobin. No at-risk fetus was found. In the same period (from July 1999 to October 2000) there were 304 other referrals to the Brent Sickle Cell and Thalassaemia Centre of pregnant women with abnormal haemoglobins. In total, 153 partners were tested and 19 at-risk pregnancies were found. Two babies were born with

sickle cell anaemia, and one pregnancy was terminated. Sixteen others had relatively unimportant conditions or declined follow-up.

Difference between gestational age of screening

Table 1 shows that coverage by the practices varied greatly. Three practices without an existing system for screening (practices 1, 5, and 6) had great difficulty in setting one up and screened between 3% and 26% of pregnant women. Practices with an existing system screened between 63% and 86% of pregnant women. Practice 1 had particular difficulty due to long-term sickness of the practice nurse.

Table 2 shows the difference in mean gestational age of screening when done in general practice and in hospital/midwifery clinics. Screening in general practice took place 4.05 weeks earlier in general practice than in hospital clinics and 2.86 weeks earlier than in community midwifery clinics ($P<0.001$). These data were obtained through an independent sample *t*-test with equal variances.

Table 2. Difference of gestational age of screening when done in general practice compared with screening by midwives and at first hospital attendance.

	Practices	Hospitals	Midwifery clinics
Number of cases analysed	173	253	126
Mean gestational age of screening (weeks)	9.68	13.73	12.54
Difference in means	n/a	4.05	2.86
95% CI	n/a	3.41 to 4.68	2.07 to 3.65
P-value	n/a	<0.001	<0.001

The feasibility of antenatal screening in general practice

The community groups broadly welcomed the initiative. However, opinions voiced at the whole system events challenge the feasibility of widespread implementation:

- Successful start-up requires that new working practices are needed by each discipline in the care pathway. These need to happen at the same time so that all the links in the chain function as they should. This requires support for each discipline in understanding the rationale and practicalities of the change. For example, new blood forms, new places to send results, and modified roles for specialist nurses and midwives are needed. One anecdote from this project illustrates the dangers of leaving out certain disciplines, namely that the phlebotomists in one community centre were not informed of the project. This had the effect that they discarded the laboratory form devised for the project, excluding some women from practices 1 and 4 from the study;
- It was not easy to hold together such a diverse group of disciplines unused to participatory research. This is illustrated in the report of the external evaluator,[11] which describes problems arising from unfamiliarity with the research approach, including: 'difficulty to keep the whole picture in view'; 'when key individuals left or were off sick, it proved difficult to re-establish contact'; in respect of the facilitator, a 'lack of experience in primary care development'; in respect of the steering group, 'weak project management'; in respect of academic support, 'irregular attendance of a series of different public health specialists, each of whom proposed different ways forward';
- Successful maintenance of the new system takes more resources than initially appreciated. General practice team members reported that pre- and post-test counselling for antenatal screening took an extra 30 minutes for each pregnant woman. One reason for this is that the pilot practices came to realise that it makes sense to screen and counsel about other conditions at the same time (some practices undertook full antenatal screening including HIV, syphillis and rubella);
- At the second whole system workshop the participants formed the view that the required changes throughout the care pathway were too big. Small-group:large-group iterations allowed consensus to emerge about the meaning of the data in light of professionals' personal experiences. The group formed the view that preconceptual awareness of a carrier state would be ideal; women already known to have the heterozygous state could be fast-tracked to secondary services without any change in hospital and laboratory systems. A patient-held record of their genetic status might ensure that information was not lost in the event of the woman moving to another practice.

In the summer of 2004 all of the practices had reverted to screening at hospital or by community midwives. Several practices expressed heightened awareness about haemoglobinopathies and one practice had developed an in-practice midwifery clinic.

DISCUSSION

Summary of main findings

Antenatal screening for haemoglobinopathies in general practice is achievable with motivated

practices and support to develop in-practice systems. It can result in significantly earlier identification of risk. However, it is not easy to achieve adequate coverage because of the need to set up new practice systems, the expectation that other screening and counselling will take place at the same time, and the need to develop new working practices throughout the whole care pathway.

The multiple methods used, set inside a framework of whole system participatory action research, were able to help disciplines from the whole system to better understand the broader issues involved in this seemingly simple innovation. It succeeded in helping all to agree a common purpose (to identify at-risk pregnancies at an early stage), but challenged the originally proposed solution (that general practices should do the screening).

Comparison with existing literature

This study supports the findings of Modell *et al*[4] and Anionwu and Atkin[13] that implementing antenatal screening for haemoglobinopathies in general practice is difficult. It gives insight as to why organisational change can be 'chaotic, often involving shifting goals, discontinuous activities, surprising events, and unexpected combinations of changes and outcomes'.[14] It lends strength to the argument that integrating qualitative methods within pilot studies can help interpret the quantitative result.[15]

Strengths and limitations of the study

The participatory and multiple methods of this research approach enabled a breadth of insight. However, the people involved were unused to the participatory processes, and the coherence of the findings presented here retrospectively do not reveal the difficulties encountered when developing the project prospectively. Only at the final meeting did there develop a general recognition of the value of considering perspectives from throughout the whole system of care.

Implications for clinical practice and future research

More work needs to be done to explore other ways to identify at-risk pregnancies at an early stage. The value of whole system participatory action research in other complex health service developments should be tested.

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Ethics committee

Brent and Harrow Ethical Committee approval was gained (CMH 182)

Competing interests

Lola Oni works at the Brent Sickle Cell and Thalassaemia Centre. Alma Smith and Judith St Hilaire also worked at the Brent Sickle Cell and Thalassaemia Centre during the lifetime of the project

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