Sickle cell screening makes genetic counselling everybody's business

As the world's first linked newborn and antenatal screening programme for haemoglobinopathies is now being rolled out in England, **Zosia Kmietowicz** looks at the challenges the programme faces

By the end of this year all pregnant women in England thought to be at risk of having a child with sickle cell disease will be offered a screening test for the blood disorder. This is in addition to universal screening of pregnant women for thalassaemia.

The introduction of a national antenatal screening test for the haemoglobinopathies should mean an end to surprise diagnoses among infants presenting with severe overwhelming infections and splenic crises, said Allison Streetly, director of the NHS sickle cell and thalassaemia screening programme. "Ultimately, every baby born with one of the blood disorders should be expected and be the result of an informed choice," she said. "And appropriate and comprehensive care should be available from birth."

The antenatal screening programme for the haemoglobinopathies runs alongside the national programme for screening newborn babies for sickle cell disease. The programmes are the world's first to link results from newborn and antenatal screening. It is also the first time that a national screening programme identifies not only those babies who are affected by a condition but also those who are carriers, and is also the first that systematically informs parents.

Dr Streetly explained the ethical basis for the programme: "Newborn screening identifies babies who would be at significant risk of death unless prompt prophylactic action is taken. However, the process also detects carrier status. The National Screening Committee took the view that we couldn't withhold the carrier information." She welcomed the recent report from the Human Genetics Commission on the genetic consequences of reproduction (*BMJ* 2006;332:

Allison Streetly says that screening will end late diagnosis

254, 4 Feb), as it "confirms the approach we have adopted as appropriate and valid."

Dr Streetly also welcomed the report's emphasis on the need to provide good quality services for people affected by the conditions. "Genuine informed choice must enable parents to choose to continue with an affected pregnancy in the knowledge that appropriate care is available."

The universal newborn screening programme for sickle cell disorders started to be extended beyond London and the West Midlands in 2003. It now covers about 90% of newborn babies in England, and the remaining areas are due to implement the programme by the end of April.

Screening of newborns is expected to identify between 250 and 300 babies with sickle cell disorders and between 7500 and 9000 carriers each year. These numbers make the condition at least as common in England as cystic fibrosis.

Since last year most trusts covering populations with a high prevalence of sickle cell disease

(areas where the condition is estimated to affect more than 1.5 per 10 000 pregnancies) have also been offering universal antenatal screening. By the end of 2006 areas with a low prevalence (fewer than 1.5 per 10 000 pregnancies affected by sickle cell disease) will have to offer a minimum of laboratory testing for the haemoglobinopathy, which will be based on an assessment of risk determined by women's family origins.

A questionnaire on family origins, designed to help health professionals detect women at risk, has been piloted by midwives at seven sites across the country. The questionnaire (which can be seen at www.kcl-phs.org.uk/haemscreening/publications.htm) asks about the ethnic origins of both partners and goes back two generations. It is hoped that eventually general practitioners and practice nurses will be able to use a similar tool when pregnant women first present. Dr Streetly said that midwives have found the questionnaire immensely helpful. "But it has also highlighted that there are training needs around the issue of ethnicity," she said.

The professional education for genetic assessment and screening training programme (the PEGASUS programme (www.pegasus.nhs.uk)) has been designed to fulfil some of those needs by building up the skills of local trainers, who can then train colleagues in their workplaces. The training package includes an element about cultural awareness. It also includes questions frontline staff should be asking to detect couples who might be carriers of sickle cell disease and might warrant screening.

Another challenge facing the antenatal arm of the screening programme is completing the antenatal screening process within a limited time frame, said Dr Streetly. "Blood for carrier screening can be taken at any time. But ideally this should happen before 10 weeks [gestation] so that there is time for counselling, testing the father, fetal testing, and the option, if the couple so chooses, of an early termination."

Guidelines on antenatal care produced by the National Institute for Health and Clinical Excellence say that pregnant women should be given information about screening tests the first time they present for antenatal care.

However, there is still a long way to go before awareness among the public and professionals of the haemoglobinopathies matches awareness of other genetic disorders such as Down's syndrome, said Dr Streetly.

Collis Rochester-Peart, a specialist nurse counsellor in haemoglobinopathies charged with developing screening services in southeast London, Kent, and East Sussex, said doctors can still be found who believe that "you can't have sickle cell disease because you're not African." But things are improving, she said, thanks largely to the linked screening programmes.

"We are no longer going into families to tell them their child has sickle cell disease and [finding that] they have no prior knowledge of the condition," said Ms Rochester-Peart. "They may not be aware of their own status, but they do have some awareness of the disease. Even a few years ago many families would never have heard of the condition."

Jane Logan, a general practitioner in Kennington, south London—an area with a high prevalence of the disease—has been offering preconceptual and antenatal screening for sickle cell disease and thalassaemia for men and women since 1994. She says the entire staff of her practice, from receptionists through to practice nurses, supported the decision to introduce the service. "Once you find out too late that a woman is carrying an affected baby and you can't do anything about it, it [introducing the service] is an easy decision to make," she said.

Dr Logan, who is one of a small number of people appointed as "GPs with a special interest in genetics" as a result of the government's white paper on genetics (*BMJ* 2003;326:1413), sees genetic counselling as an increasingly important issue for general practice, and not just in those parts of the country with large numbers of people at high risk.

On her role in the screening service she says: "It is very rewarding, especially as GPs' role in obstetrics has naturally been waning. When women present thinking they might be pregnant, it is a very critical and significant consultation. They ask lots of questions, and it is very easy to discuss possible inherited disorders and screening." □

Zosia Kmietowicz *London*