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Facilities for prenatal diagnosis

The practical and ethical difficulties of providing a prenatal diagnostic service for the whole population have recently been widely discussed, but the facts needed for rational planning have not been available. The council of the Clinical Genetics Society has now published a booklet[1] which provides much of this essential information. The report is based on a detailed survey in 1977 of 16 000 pregnancies tested by amniocentesis before 20 weeks' gestation. Of the 69 laboratories in Britain which provide prenatal diagnostic services, 51 replied to questions on the indications for referral, the needs of the community, the quality of the service, staffing, equipment, and funding.

The results of the survey suggested that women should be referred for prenatal diagnosis if they have had an infant with a neural tube defect or trisomy 21; if the maternal age is over 35 years; if there is a family history of chromosomal translocation, a detectable metabolic disorder, or neural tube defect; if the mother is a carrier of x-linked disease; or if she has a raised serum concentration of alphafetoprotein (AFP). A previous infant with autosomal aneuploidy other than trisomy 21, a family history of trisomic Down's syndrome, and maternal anxiety were not seen as strong indications for prenatal diagnosis.

In 1976 the prenatal diagnosis clinics identified 110 fetuses with open neural tube defects and 33 with Down's syndrome. At the other end of the scale only about 100 pregnancies were known to be at risk for the 37 different metabolic diseases that can be diagnosed prenatally, and 12 different laboratories had detected 19 affected fetuses among 70 pregnancies tested—most of the tests being undertaken by four NHS-funded laboratories.

Testing for amniotic AFP was cost-effective when performed on every specimen, but cytogenetic studies were not being done in several laboratories unless there was an additional indication such as maternal age. Every regional health authority (with one exception) now had at least one laboratory undertaking prenatal diagnostic tests. Most of the investigations were handled by one or two larger laboratories, but some laboratories, says the report, were offering a cytogenetic service before they had reached a sufficient level of skill. Many were unable to follow up continuing pregnancies to determine the false-negative rates for the various tests. Wrong sexing due to growth of maternal rather than fetal cells was the most common error. The minimum requirements for quality control, says the report, should be laboratory confirmation of the prenatal diagnosis in all terminations and verification of the birth of a baby of the predicted sex.

Prenatal diagnosis has now passed out of the research stage and has become entirely a service responsibility; yet in many regions the work is still being supported by research funds. The 27 of the 45 laboratories that gave information on finance received most of their running expenses from the area health authority. Nevertheless, at least part of the work was usually undertaken in university accommodation by staff holding university or research appointments funded exclusively from outside the NHS. Six laboratories were totally dependent on research grants. In general the laboratories believed that the serum AFP screening test should be undertaken by regional laboratories (using semi- or fully-automated procedures). That should allow adequate communication between the laboratory and the obstetric and counselling services in the region, so that no time would be lost in recalling patients for ultrasonography, a second serum AFP test, and if necessary diagnostic amniocentesis.

Clearly what we need is a comprehensive service funded by regional health authorities and integrated with genetic advisory centres. The Clinical Genetics Society suggests that a standing committee on genetic services should be set up to ensure adequate facilities and laboratory standards. The report estimates that facilities for prenatal diagnosis are needed in about 8% of all pregnancies; ultrasound facilities will need considerable expansion if all these patients are to be screened safely. The report also suggests that a national programme should be set up as soon as possible for detecting open neural tube defects by screening all women using estimations of the serum AFP, and that a national register of amniocentesis results should be established to monitor the reliability of the diagnostic tests and the safety of amniocentesis.

In concentrating on the laboratory diagnosis of neural tube defects and chromosomal abnormalities the report may not have emphasised sufficiently the work of the clinical geneticist. Prenatal diagnosis has helped only a minority of his patients: there are no prenatal tests for several common conditions including congenital heart disease and cystic fibrosis, but even so patients at risk can still be given valuable, practical guidance.

This careful and detailed survey should be read by all concerned with planning obstetric and paediatric services. If the policy of screening by regional centres is to be put into effect these must be funded by regional boards and not merely designated—for otherwise a service now available to the privileged few will disappear as laboratories become submerged in a sea of specimens.

[1] *The Provision of Services for the Prenatal Diagnosis of Fetal Abnormality in the United Kingdom*. Report of the Clinical Genetics Society Working Party on Prenatal Diagnosis in Relation to Genetic Counselling. Obtainable from the Eugenics Society, 69 Eccleston Square, London SW1V 1PJ. Price £1·50.