Special report

Recognition and management of fetal abnormalities

MEMBERS OF THE JOINT STUDY GROUP ON FETAL ABNORMALITIES

1 Introduction

1.1 The detection and management of fetal abnormalities are of prime concern to paediatric surgeons because they are responsible for the postnatal care of a substantial number of babies with such defects. In order to encourage cooperation between the specialists involved with the antenatal and immediate postnatal care of the fetus, the British Association of Paediatric Surgeons convened a study group with representation from those colleges and associations whose members would have experience and knowledge in this field. The Department of Health was invited to send an observer who contributed to the discussions. This document has been compiled by the group and its purpose is to improve accuracy in the diagnosis of fetal abnormalities and raise the standard of subsequent care. Encompassed within this objective are aspects which concern doctors in several specialties, nursing and midwifery staff, and providers of health care resources, clinical and laboratory technicians and—most importantly—pregnant women and their families.

The factors are considered under two main headings: diagnosis and management.

1.2 An appendix summarising the results of the Northern Region Fetal Abnormality Survey for 1986¹ is given at the end of the paper. This gives the number and type of fetal abnormalities suspected by routine antenatal screening as a proportion of the number actually born. The number of erroneous diagnoses is also included. These figures illustrate the scale of the problem currently confronting antenatal diagnostic services. Some of the relevant factors were discussed in a previous publication.²

2 Diagnosis

2.1 The intention is to outline a standard for screening services for fetal abnormalities for which a number of techniques are available. There is a need for a two tier service comprising district and regional levels with perhaps an additional third supraregional level where intensive, finely focused teaching and research will be carried out and special skills developed.

2.2 All diagnostic units should be able to make immediate assessments of fetal abnormalities and be prepared to seek further opinions on diagnosis and treatment from a regional centre. Antenatal ultrasound at district hospital level provides one of the most important diagnostic methods available and its accuracy at the present time is sufficiently good to justify its increasing use.

2.3 We do not have the resources or the expertise to estimate the financial cost of the service we are recommending except to say that extra resources will be required. Neither is it possible to equate the outcome of antenatal ultrasound screening with precise financial gain. It seems clear to us, however, that money for antenatal screening services is very well spent. They can relieve the burden of continuing pregnancy when the fetus is severely malformed, assist in the management of pregnancy, forewarn of some of the impending obstetric complications, give early warning of the need for urgent medical and surgical postnatal care, and offer substantial improvements in maternal and neonatal morbidity.

2.4 It is the duty of the team involved in antenatal diagnosis not only to work closely together so as to create a rational plan of management but to communicate sympathetically and comprehensively with the parents. Clearly, the consultant obstetrician or his deputy should be responsible for informing the mother of the results and implications of the investigations. Experience indicates that most parents desire antenatal screening for fetal abnormalities.

3 Recommended screening

3.1 Structural malformations of the fetus are a major cause of morbidity and mortality. Screening of all pregnancies is necessary as most (about 80%)

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congenital abnormalities occur in the absence of recognised risk factors. Ultrasound examination and measurement of maternal serum α fetoprotein are at the present time the principal means of diagnosis and can achieve a high level of detection of serious abnormalities.

3.2 Structural abnormalities are recognised mainly by ultrasonography and appropriate staff and facilities for ultrasound examination should be available for all pregnant women at an appropriate time in the second trimester.

3.3 With presently available equipment, expertise, and facilities, the minimum gestation at which a detailed fetal scan will provide maximum information is 18–20 weeks. We believe that normally all pregnant women should be offered a detailed fetal anomaly scan by 20 weeks' gestation.

3.4 Some centres are offering screening for α fetoprotein to pregnant women at around 16 weeks. If the recommendation in paragraph 3.3 above is accepted, serum screening at 16 weeks will remain important in the earlier detection of neural tube defects. Furthermore, in the light of recent evidence that maternal serum screening at 16 weeks may lead to the recognition of most cases of Down's syndrome, this technique is likely to be employed more widely.

3.5 An explanatory leaflet given to each mother at her first antenatal clinic attendance would be most helpful. Mothers must be made aware in advance of the limitations and implications of screening and their consent sought. If the offer of either of the examinations is declined an appropriate comment should be recorded.

4 Pregnancies at high risk of fetal abnormality

4.1 In this numerically small group of pregnancies, management should be tailored to specific needs. This category includes:

(a) Women of 35 years and over.
(b) Fetal abnormality in a previous pregnancy or a poor obstetric history—for example, recurrent abortion, growth retardation, or rhesus haemolytic disease.
(c) A family history of genetic disease identifiable by prenatal diagnostic techniques.
(d) Coexistent maternal disease liable to affect fetal development—for example, diabetes mellitus, hypertension, or renal disease.
(e) Maternal exposure to rubella or other infection liable to affect fetal development.
(f) Maternal drug or alcohol abuse.
(g) Maternal exposure to certain prescribed medications and other suspected teratogens.
(h) Complications during pregnancy—for example, intrauterine growth retardation, oligohydramnios, and polyhydramnios.

4.2 Preconception counselling and early booking for pregnancy care is particularly desirable in such patients. Other techniques may include:

(a) Chorionic villus sampling.
(b) Amniocentesis.
(c) Early anomaly scanning.
(d) Fetal blood sampling.
(e) Fetal tissue biopsy.

5 Clinical management

5.1 Once a fetal abnormality is suspected, a fundamental requirement for good care is close communication among those with diagnostic expertise or potential involvement in neonatal care. This includes neonatologists and specialists in paediatric cardiology, urology, nephrology, neurology, surgery, clinical genetics, cytogenetics, radiology, and fetal/perinatal pathology. All obstetricians in a district or region should have available the names, hospital locations, and telephone numbers of colleagues in the above specialties so that they may be contacted when the need arises.

5.2 The mother and her family should be given the opportunity of a consultation with any of the specialists who will be involved in the management of the pregnancy and postnatal care of the baby. If an abnormality is suspected which will require neonatal surgery, a visit to the neonatal surgical unit where the baby will be nursed postoperatively is helpful.

6 Regional referral centres

6.1 One or more such centres should be made available to each health region to offer training, advice, and support in prenatal diagnosis and clinical management. They should be equipped to provide a full range of prenatal diagnostic techniques including facilities to permit rapid karyotyping when abnormalities are detected in the second three months of pregnancy.

6.2 The main attribute of a regional centre is to bring together the following experts: obstetricians with a special interest in fetal medicine, radiologists practising antenatal ultrasound, neonatologists, clinical geneticists, cytogeneticists and specialists in
paediatric surgery, fetal/perinatal pathology, neurology, nephrology, cardiology, and endocrinology.

6.3 It is recommended that a regional advisory committee on fetal abnormalities with members from each of the appropriate specialities be established in each region. This would advise on all aspects of antenatal screening and make recommendations with regard to equipment, staffing, and management. It should make arrangements for collection of data and encourage the development of a regional malformations register. To this register should be added details of anomalies detected in spontaneous abortions and stillbirths.

6.4 Regular meetings of all those involved in the prenatal diagnosis and management of malformations should be set up at regional or subregional level (Area Health Boards in Scotland). It is envisaged that the results of prenatal investigations and detailed pathology findings would be available at such meetings.

6.5 It is recommended that the long term outcome of children with congenital abnormalities diagnosed antenatally be carefully documented.

7 Specific actions

7.1 The first and most important step is to establish as firm a diagnosis as possible. Repeat ultrasound scanning by an appropriately experienced obstetrician or radiologist using more sophisticated equipment may be necessary. Other diagnostic tests such as amniocentesis and fetal blood sampling as detailed in the section on diagnosis may be appropriate.

7.2 Those specialists likely to be involved in the perinatal care of the fetus should be informed so that a plan of management for the remainder of the pregnancy and for the delivery of the baby can be formulated.

7.3 Consultations between the obstetricians, other specialists, and the parents should be arranged at this stage so that the parents understand how the fetus is affected and the options for the further conduct of the pregnancy (see section 8).

7.4 It is important that the team primarily concerned with antenatal care continue to give advice, support, and information even though specialists in other fields become involved. The family practitioner should be kept constantly informed of the patient’s progress and consulted over the handling of relevant domestic issues.

8 Alternative courses of action

8.1 TO CONTINUE WITH THE PREGNANCY

Postnatal investigation and treatment are then undertaken as soon as indicated. For some mothers, arrangements should be made for the baby to be delivered in a maternity unit located as close as possible to the regional neonatal surgical unit.

8.2 TO ATTEMPT PREGNATAL TREATMENT

Prenatal treatment requires invasive procedures which should be undertaken only in regional centres where the equipment and skilled personnel are available.

8.3 TO TERMINATE THE PREGNANCY

This may be offered for serious fetal abnormalities incompatible with postnatal life and for abnormalities which carry a substantial risk of serious mental and/or physical handicap. It is essential that the mother and her family have the opportunity to discuss every aspect of the problem in full with her obstetrician and all other appropriate experts, to express her wishes, and make an informed choice.

8.4 Adequate provision should be made available for the counselling and care of couples before and after such terminations. Midwives, health visitors, general practitioners, genetic nurses, and voluntary self help groups may all offer valuable support.

9 Fetal pathology

9.1 Each region should have at least one perinatal pathologist. We are aware of the difficulties encountered hitherto and support the Royal College of Pathologists and other interested bodies in pressing for more perinatal pathologists throughout the United Kingdom.

9.2 When a pregnancy is terminated because of a fetal abnormality the fetus should undergo careful examination by a pathologist with an interest and expertise in fetal pathology. If no such expert is available locally the fetus should be transferred to a specialist in perinatal pathology in the appropriate regional centre.

9.3 In appropriate cases, maternal serum and fetal tissue should be examined for TORCH (toxoplasma, rubella, cytomegalovirus, herpes) antibodies and parvovirus. Chromosomes should be examined and a photographic and radiographic record of the fetus should be obtained.

9.4 Products of conception from any pregnancy
which aborts after any invasive investigation should be examined by a pathologist (see 4.2).

9.6 The purpose of fetal examination is threefold: firstly, confirmation of the anomaly for which termination was undertaken; secondly, careful scrutiny of fetus, placenta, and membranes for any abnormality which might be related to invasive investigations; thirdly, careful documentation of all abnormalities present to enable a precise diagnosis to be reached so that parents may be advised about the possibility of recurrence.

10 Conclusion

10.1 The proposals set out in this document represent a considerable expansion of the service covering the antenatal diagnosis of fetal abnormalities. The need for additional manpower, training, and equipment will require special study and consultation.

10.2 These recommendations, if implemented, will lead to improvements in the accuracy and sensitivity of the antenatal diagnosis of fetal abnormalities. Better cooperation and more effective exchange of information should enable suspected fetal abnormalities to be managed on a more rational basis.

Appendix

11 The Northern Regional Fetal Abnormality Survey

11.1 This survey of suspected fetal abnormality was started as a pilot study in 1984 and because of its initial success became permanently established in 1985 under the auspices of the Northern Region Health Authority. The structure of the survey and the results for 1986 are available in print from the Northern Region Health Authority.† The following data are abstracted from that report with some additional analysis.

11.2 Multiple sources of data collection are employed (table 1) in order to identify major structural malformations presenting in the first year of life. Retrospective assessment of prenatal diagnostic procedures is recorded. In addition, the register records all suspected major malformations and chromosome defects notified through the confidential returns from the region’s obstetric units and cytogenetic laboratories. Thus it is possible to record true and false positives and false negatives from a representative population which will provide a measure of the sensitivity and selectivity of existing antenatal diagnostic services.

11.3 ANTENATAL DIAGNOSIS OF SELECTED FETAL ABNORMALITIES 1986
Diagnoses made before delivery in patients delivered in 1986 as a proportion of all cases coming to specialist attention within one year of birth are shown in table 2.

11.4 NEURAL TUBE DEFECT
Almost all anencephalics were identified by the end of the second trimester. Four were not terminated because of a healthy co-twin. Less than half the cases of spina bifida were identified.

11.5 Other anomalies of the central nervous system such as hydrocephalus were diagnosed accurately

Table 1 Sources of data used to identify major structural abnormalities presenting in the first year of life

<table>
<thead>
<tr>
<th>Source of Data</th>
<th>Number of Diagnoses</th>
</tr>
</thead>
<tbody>
<tr>
<td>Regional perinatal mortality survey</td>
<td>77/77 (100)</td>
</tr>
<tr>
<td>District health authority SD56 returns</td>
<td>14/14 (100)</td>
</tr>
<tr>
<td>Registrar General’s annual death tapes</td>
<td>25/27 (93)</td>
</tr>
<tr>
<td>Laboratories in which a foetal protein assay is done</td>
<td>7/8 (87)</td>
</tr>
<tr>
<td>District obstetric ultrasound services</td>
<td>10/12 (83)</td>
</tr>
<tr>
<td>Regional cytogenetic laboratories</td>
<td>9/13 (69)</td>
</tr>
<tr>
<td>Records of referrals of patients &lt;1 year from regional centres for:</td>
<td>11/16 (69)</td>
</tr>
<tr>
<td>Neonatal surgery</td>
<td>7/8 (87)</td>
</tr>
<tr>
<td>Paediatric cardiology</td>
<td>14/14 (100)</td>
</tr>
<tr>
<td>Paediatric urology</td>
<td>23/46 (50)</td>
</tr>
<tr>
<td>Neurology/neurosurgery</td>
<td>3/7 (43)</td>
</tr>
<tr>
<td>Regional genetics advisory service</td>
<td>1/3 (33)</td>
</tr>
<tr>
<td>Grapevine</td>
<td>1/5 (20)</td>
</tr>
<tr>
<td>Other</td>
<td>13/93 (14)</td>
</tr>
<tr>
<td>Chromosome abnormality</td>
<td>1/10 (10)</td>
</tr>
<tr>
<td>Oesophageal atresia</td>
<td>1/6 (6)</td>
</tr>
<tr>
<td>Diaphragmatic hernia</td>
<td>0/13 (0)</td>
</tr>
<tr>
<td>Gut atresia below duodenum</td>
<td>0/4 (0)</td>
</tr>
<tr>
<td>Lethal skeletal abnormalities</td>
<td>1/16 (6)</td>
</tr>
<tr>
<td>Non-lethal skeletal abnormalities</td>
<td>10 (10)</td>
</tr>
<tr>
<td>Isolated renal abnormalities</td>
<td>43 (33)</td>
</tr>
<tr>
<td>Isolated cardiac abnormalities</td>
<td>34 (26)</td>
</tr>
<tr>
<td>Termination of pregnancy</td>
<td>46 (36)</td>
</tr>
<tr>
<td>Other</td>
<td>13/93 (14)</td>
</tr>
</tbody>
</table>

Table 2 Diagnoses made before delivery in patients delivered in 1986 as a proportion of all cases coming to specialist attention within one year of birth

<table>
<thead>
<tr>
<th>Diagnoses</th>
<th>No (%) of patients</th>
</tr>
</thead>
<tbody>
<tr>
<td>Unilateral renal damage treated by nephrectomy</td>
<td>7/7 (100)</td>
</tr>
<tr>
<td>Obstruction of pelviureteric junction treated by pyeloplasty</td>
<td>14/14 (100)</td>
</tr>
<tr>
<td>Anencephaly</td>
<td>25/27 (93)</td>
</tr>
<tr>
<td>Bladder outlet obstruction</td>
<td>7/8 (87)</td>
</tr>
<tr>
<td>Hydrocephalus without spina bifida</td>
<td>10/12 (83)</td>
</tr>
<tr>
<td>Bilateral renal agenesis or lethal dysplasia</td>
<td>9/13 (69)</td>
</tr>
<tr>
<td>Exomphalos/gastrochisis</td>
<td>11/16 (69)</td>
</tr>
<tr>
<td>Non-rhesus hydrops</td>
<td>4/6 (67)</td>
</tr>
<tr>
<td>Spina bifida with or without hydrocephalus</td>
<td>23/46 (50)</td>
</tr>
<tr>
<td>Encephalocoele</td>
<td>3/7 (43)</td>
</tr>
<tr>
<td>Chylous pleural effusion</td>
<td>1/3 (33)</td>
</tr>
<tr>
<td>Duodenal atresia</td>
<td>1/5 (20)</td>
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<tr>
<td>Chromosome abnormality</td>
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</tr>
<tr>
<td>Lethal skeletal abnormalities</td>
<td>0/4 (0)</td>
</tr>
</tbody>
</table>
but in most at too late a stage in the pregnancy to offer termination. Nearly all the survivors of these and other anomalies of the central nervous system are handicapped.

11.6 Urinary Tract
The accuracy of antenatal diagnosis was good though how many children with unsuspected anomalies presented during the first year of life is unknown at the moment. Two children were identified in 1986 where mild hydronephrosis detected antenatally was not apparent at birth but in whom subsequent investigation showed significant pelviuretic obstruction.

11.7 Abdominal Wall
Nearly 70% of these were correctly diagnosed antenatally with no false positives. Retrospective analysis indicates a substantial improvement in diagnostic sensitivity when compared with the period 1980-3. Prenatal diagnosis permits an examination for associated anomalies and multidisciplinary counselling of parents. If the pregnancy goes to term the mother can be admitted for her delivery to the maternity unit nearest the regional paediatric surgical centre.

11.8 Diaphragmatic Hernia
The accuracy of antenatal diagnosis was poor but there is as yet no evidence that prenatal recognition would have improved survival.

11.9 Chromosome Abnormality
Amniocentesis was carried out in only a small proportion of the pregnant women over the age of 35 years. Only six of 52 fetuses (11.5%) with Down's syndrome and 14% of the total number of whom chromosome anomalies were identified antenatally.

11.10 Congenital Heart Disease
Seventy one infants with cardiac malformation delivered in 1986 have been recorded to date but only three were suspected antenatally. Despite subsequent improvements in routine screening for heart defects in this region, it is apparent that the sensitivity of antenatal diagnosis for cardiac malformations remains poor.

11.11 False Positives
This survey is unique in providing information on false positive as well as false negative diagnoses. While antenatal diagnosis was often imprecise there were very few examples of erroneous diagnosis of a serious abnormality. Three suspected cardiac anomalies, one suspected duodenal atresia, one suspected porencephalic cyst, and one spontaneous abortion with suspected spina bifida constituted the principal examples. Caution in interpretation of false positives is necessary, particularly in relation to renal and cardiac defects. The former may not be apparent in the live neonate and cardiac defects may not be recognised without specialist pathological assessment.

11.12 The register contains details of 491 defects, approximately 1.2% of total births in 1986. This figure falls short of the 3% of births associated with congenital anomalies in large epidemiological surveys. In part this is due to incomplete ascertainment of, for example, anomalies which become apparent in later childhood. Furthermore, certain categories such as metabolic defects and isolated non-specific mental retardation have not been included.

11.13 Of recorded cases, 248 (50.5%) received some form of prenatal diagnostic assessment but no abnormality was suspected. Seven (1.4%) received no antenatal screening and in eight (1.6%) no details of the pregnancy were obtained. Abnormality was suspected in 228 (46.4%) but the diagnosis was incomplete or inaccurate in many cases and was often made too late for appropriate intervention.

11.14 A chromosome abnormality imposes a considerable burden on affected families. Rapid chromosome examination when anomaly scanning shows major malformations, together with current developments in the biochemical analysis of maternal serum, have the potential to increase the proportion of chromosomally abnormal fetuses recognised in the second trimester.

References

1 Northern Region Health Authority. A regional fetal abnormality survey. First progress report. Newcastle upon Tyne: Northern Region Health Authority, 1988.

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