Report of Metabolic Disorders Working Group.

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Contents

Introduction

TERMS OF REFERENCE

1. We were appointed as a Working Group by the Minister for Health, Dr. Rory O'Hanlon, T.D., in September 1990 with the following terms of reference:

   - to establish where the responsibility lies for ensuring that all newborn infants are screened for metabolic disorders;
   - to recommend practices and procedures to ensure that all infants are screened for metabolic disorders;
   - to facilitate a co-ordinated approach by all health boards to the stated objective.

2. The members of the Working Group were:

Mr. Peter McQuillan	Chief Executive Officer, South-Eastern Health Board (Chairman)
Dr. E. R. Naughten	Consultant Paediatrician, Temple Street Hospital
Ms. Maura Breslin	Superintendent Public Health Nurse, Midland Health Board
Dr. Neville de Souza	Director of Community Care, South-Eastern Health Board
Ms. Catherine Craughwell	Matron, Regional Maternity Hospital, Limerick
Dr. T. V. O'Dwyer	Deputy Chief Medical Officer, Department of Health
Prof. Gerard Cussen	Erinville Hospital, Cork
Dr. Deirdre Murphy	General Practitioner
Ms. Dolores Moran	Higher Executive Officer, Department of Health (Secretary)

3. To undertake our task, we acquainted ourselves with all aspects of metabolic disorders and the testing arrangements in existence at present by exchanging the first-hand knowledge and experience of the Group members and from corresponding with public and private hospitals and health boards.

4. Attention was focussed on the medical consequences of a missed or delayed diagnosis of a metabolic disorder which could result in mental handicap, severe illness or even death of the infant. The importance of ensuring 100% coverage from the medico-legal point of view was also addressed.

5. The main thrust of our conclusions is that the procedures in place, to ensure neonatal screening is performed, are good but that there are areas which can be improved upon so that the objective of 100% coverage can be attained.

6. The financial implications of our recommendations are not significant. If our recommendations are not implemented, possible court awards could be much greater than the cost of implementing the recommendations.

7. The implementation of our recommendations should be initiated without delay.

Acknowledgements

We would like to thank everyone who helped us and we would particularly wish to express our appreciation to Dr. Seamus Cahalane, Consultant Pathologist; and Mr. Charles O'Neill, Chief Technologist, National Neonatal Screening Laboratory, Children's Hospital, Temple Street, and Mr. Raymond Smyth, Systems Analyst, Department of Health, who assisted the Group. We would like to thank, in particular, Ms. Dolores Moran for her excellence as Secretary to the Working Group and her skill in drafting the report.

MAIN FINDINGS

Neonatal Screening Procedures in place at present

1. The Group's objectives were to draw up procedures and practices to ensure the screening of all infants for metabolic disorders and to facilitate a co-ordinated approach by all health boards.

2. To determine the existing procedures in this regard, each health board and every hospital where births take place were asked to outline their practices and procedures in respect of hospital births and domiciliary births in their respective areas. The Group found that

   (a) in all hospitals it was indicated that the heel prick test was carried out between the 3rd and 5th day after birth and in most hospitals on the 4th day. However, from the analysis of data in the National Neonatal Screening Laboratory, it appears that not all the information given by the hospitals is correct.

   (b) if the infant is on antibiotics or had an inadequate protein intake, e.g. feeding not established, the test is repeated in some centres.

   (c) if the infant is discharged prior to the test being carried out, the test is carried out by requesting

      (i) the mother to bring the infant to the hospital for the test *or*

      (ii) the mother to bring the infant to the health centre to have the test *or*

      (iii) the mother to bring the infant to the local general practitioner for the test *or*

      (iv) the public health nurse to call to the infant's home to carry out the test.

(d) in most hospitals the midwife discharging the infant ensures that the test was carried out.

(e) a record that the test was carried out is made in

(i) the baby/birthday book *or*

(ii) a separate register for this purpose *or*

(iii) the child's record.

(f) in the case of domiciliary births, the public health nurse who is notified of the birth by the general practitioner or midwife, ensures that the test is carried out.

(g) if parents, following a request, refuse to allow the sample to be taken from the infant, they are requested to signify their refusal in writing.

(h) the test is forwarded to the National Neonatal Screening Laboratory, Temple Street Hospital, often by registered post.

(i) when the results are received from the National Neonatal Screening Laboratory, they are recorded in most hospitals.

(j) if no results are received, the ward sister in the hospital of birth usually follows up the case with the National Neonatal Screening Laboratory.

(k) if the result is positive, the parents are requested to bring the infant to Temple Street Hospital for confirmation and treatment immediately.

3. Information was also sought from the National Neonatal Screening Laboratory as to their screening procedures which are briefly outlined as follows:

   (a) 150-400 tests are received in the Laboratory each day.

   (b) the test on the specimen is carried out, incubated overnight and read the next day. However, there is no routine testing on Saturday or Sunday. Therefore, if a sample is received on Friday, it will not be read until Monday (or Tuesday in the case of a Bank Holiday weekend). There is an emergency call-out service (diagnostic) at week-ends as distinct from routine screening. Christmas also causes delays in screening as the Laboratory depends for the greater part on the postal service for delivery of samples.

   (c) if the test is definitely positive, the hospital is immediately contacted and the parents sought and requested to bring the infant to Temple Street Hospital for confirmation and treatment.

   (d) if the test is not definitive, a liquid blood sample is requested to be forwarded as soon as possible.

   (e) negative results are reported to sender.

4. Having acquired the information on the current procedures, the Group identified the areas in the system where improvement is necessary and a series of recommendations are made so that the objective of 100% coverage of infants can be attained. Recommendations are also made as to where the responsibility lies for ensuring that that stated objective is attained.

RECOMMENDATIONS

Screening for metabolic disorders must be considered an integral part of the management of newborn infants. Screening involves the co-operation of many agencies i.e. community care teams in health boards, maternity hospitals, the National Neonatal Screening Laboratory at Temple Street Hospital, and general practitioners.

1. Responsibility for ensuring that every infant is screened

1.1 Responsibility of health boards

The responsibility for screening for metabolic disorders should be placed on a designated person/authority. **The Group recommend that the ultimate responsibility to ensure that the highest possible number of infants are screened should be placed on the health boards. Thus, the health boards should be responsible for ensuring that the tests are carried out in accordance with the protocol set out in the following paragraphs.**

1.2 Responsibility of hospitals

The Group recommend that the hospitals should be responsible for ensuring that infants born in hospital are screened for metabolic disorders. If it is not appropriate to carry out the test before discharge from hospital, the hospital is responsible for ensuring that the child is screened by appointment at the hospital subsequently on the appropriate day or alternatively that the superintendent public health nurse is made aware that the infant has been discharged prior to the test being carried out. If no results are received from the National Neonatal Screening Laboratory by the hospital, the hospital is responsible for ensuring that omissions are notified to the superintendent public health nurse.

1.3 Responsibility of health board community care services

If it transpires that the **test is carried out in the community, the recording and follow-up of the case is the responsibility of the superintendent public health nurse.**

1.4 **Responsibility of general practitioners and midwives**

The responsibility for **performing the test in the case of home deliveries rests with the midwife and the general practitioner performing the delivery.**

Results of tests should be forwarded to the superintendent public health nurse and the general practitioner.

1.5 **Responsibility of parents**

In the case of parents who refuse to allow their infant to be screened, the responsibility for the possible adverse consequences of their decision shifts to them.

The parents should be requested to signify their refusal in writing.

In the case of infants born outside the jurisdiction to parents who reside in the jurisdiction the parents are responsible for ensuring that the infant is screened. If the parents notify the public health nurse of the birth the public health nurse will be responsible for ensuring that the test is carried out.

1.6 **Responsibility of National Neonatal Screening Laboratory, Temple Street Hospital**

The Group recommend that the National Neonatal Screening Laboratory, Temple Street Hospital, be responsible for

- **recording receipt of tests**
- **carrying out tests on specimens forwarded in accordance with acceptable medical protocol**
- **notifying results of tests to appropriate person/authority.**

1.7 The test should be taken in accordance with the medical protocol as set out in Appendix 3.

2. Recording of information

2.1 The Group recommend that the following steps be adhered to to ensure that adequate records are maintained in the hospital which will help identify any infant who has **not** had the test carried out before discharge from hospital:

(i) a person should be nominated by the hospital to take responsibility for checking records.

(ii) the details of each infant should be recorded in a single register. (In the case of 2.2 below, the mother may be in a ward other than the delivery ward where the birthday book is kept).

(iii) this register must indicate whether or not the test has been carried out.

(iv) when the results of the test are reported back from the National Neonatal Screening Laboratory, Temple Street Hospital, the entry should be ticked in a different colour pen – thus, any case where there is no report received from the Laboratory is noticeable at a glance.

(v) the person who checks the register to identify possible omissions should initial the entries to indicate that he/she has undertaken the exercise.

2.2 The above steps should be followed in the case of *all* infants who

- **are born in a hospital by normal delivery**
- **are born in a hospital by caesarean section**
- **are born outside a hospital and then brought to the hospital**
- **are born to a mother who is suffering from an infectious disease (e.g. hepatitis B and/or HIV/AIDS which prevents her from delivering in the delivery ward).**

3. Early Discharges from Hospital; Liaison with Health Board Community Care Services

3.1 The nurse/midwife discharging the infant from hospital before the test has been carried out should **ensure that the mother is clear about the following matters:**

- **the importance of the test**
- **when it should be done**
- **that the test should be carried out**
  (i) at the maternity hospital, or
  (ii) by a public health nurse, or
  (iii) by a general practitioner.

3.2 The **superintendent public health nurse** in the area in which the baby is born is considered by the Group to be the most appropriate person to be responsible for ensuring that infants, who are discharged from hospital prior to their having the heel prick test, have the test carried out subsequently.

3.3 If the test is to be performed by a **public health nurse**, the following procedures should be followed:

(i) the ward sister should ensure that the appropriate superintendent public health nurse is notified that the test is to be carried out.

(ii) the superintendent public health nurse should keep a register of requests from hospital/s.

(iii) the superintendent public health nurse should request the appropriate public health nurse to perform the test and send the sample by post (and obtain a receipt of postage) to the National Neonatal Screening Laboratory, Temple Street Hospital.

(iv) the superintendent public health nurse should notify the maternity hospital where the birth took place that the test has been carried out.

(v) the National Neonatal Screening Laboratory will then process the sample and send a copy of all results to the maternity hospital and the superintendent public health nurse.

3.4 If the test is to be performed by a **general practitioner**, the following procedures should be followed:

(i) the superintendent public health nurse should contact the general practitioner to ensure that the test will be performed and the sample sent by post (and obtain a receipt of postage) to the National Neonatal Screening Laboratory at Temple Street Hospital.

(ii) the Laboratory will then process the sample and send a copy of all results to the maternity hospital, the superintendent public health nurse and the general practitioner.

(iii) the general practitioner should contact the superintendent public health nurse in cases of non-attendance by the parents.

3.5 **The Group recommend that *all* early discharges be notified by hospitals to the superintendent public health nurse to enable the public health nurse to give priority to these infants. If it is not appropriate to carry out the test before discharge from hospital, the hospital should ensure that the infant is screened by appointment at the hospital subsequently on the appropriate day or alternatively notify the superintendent public health nurse that the infant has been discharged prior to the test being carried out.**

3.6 In the case of **travelling people**, the infant may have moved out of the area before the test has been carried out. **The Group recommend that the superintendent public health nurse be made aware of the circumstances at the earliest possible time so that particular care can be taken in the case of these infants.**

3.7 **The Group recommend that special care should be taken in the care of breast feeding infants to ensure that the test is taken when feeding is established and that the test should be taken in accordance with the medical protocol as set out in Appendix 3.**

4. Education

4.1 An explanatory leaflet for parents stressing the importance of the test, as mentioned in para. 3.1 above, should be published by the Health Promotion Unit.

4.2 An educational programme should be embarked upon under the auspices of the Department of Health specifically targetted at staff who organise the collection of specimens and at those who collect specimens. (Illustration of how the sample should be taken is at Appendix 3).

4.3 An information document giving medical detail concerning metabolic disorders (see Appendix 1) should be distributed through appropriate medical bodies to increase the level of awareness of the importance of the screening and the consequences if detection of the condition is delayed.

5. National Neonatal Screening Laboratory, Temple Street Hospital

5.1 It is agreed by the Group that the neonatal screening card be redesigned to include further information. (See sample at Appendix 2.)

5.2 The computer technology in the National Neonatal Screening Laboratory is considered to be inadequate and in need of upgrading. (See report on technology at Appendix 4). The availability of improved technology should eliminate delays in the issue of receipts and results of tests.

5.3 Negative results should be issued as soon as possible to the sender but in any case not later than ten days from the date of receipt of the sample at the Laboratory. If the results cannot be issued within ten days from the date of receipt, an interim report should issue to sender.

5.4 The code number on the neonatal screening cards should be different for each hospital.

5.5 It was further recommended that a line format should be used in reporting back results to the hospital/S.P.H.N./G.P. i.e. one line for each entry. The issuing of the results in both date of birth order and alphabetical order was recommended if this could be facilitated by the Laboratory's improved technology. If this is not possible, the date of birth order is preferable.

5.6 Senior Laboratory personnel should have a contact point in each community care area of a health board and relevant hospital to minimise delays.

6. Review of service

6.1 The Group recommend that the National Neonatal Screening Laboratory produce an annual report.

6.2 The Group recommend that the Department of Health review the system in place at regular intervals to evaluate the service being provided.

APPENDIX 1

MEDICAL INFORMATION ON METABOLIC DISORDERS

Phenylketonuria is common in the Irish community and has an incidence of 1 in 4,500. Without treatment this condition causes severe mental handicap. It results from lack of the enzyme phenylalanine hydroxylase. This enzyme normally changes phenylalanine into tyrosine, but in its absence phenylalanine accumulates and tyrosine becomes deficient. Tyrosine is one of the essential substrates for neurotransmittor formation. Phenylalanine has a direct toxic effect on the brain in high levels and also acts by competitive inhibition with tyrosine and tryptophan, a substrate for serotonin, another neurotransmittor. In Classical Phenylketonuria the phenylalanine hydroxylase enzyme is present in a concentration of $<1\%$ and as a result of this phenylalanine rises to toxic levels and tyrosine is deficient. Early treatment is beneficial and aims at giving a low intake of phenylalanine but a normal intake of overall amino acids and augmenting tyrosine. This diet has to be continued for life. Early detection leads to early treatment.

New born screening for phenylketonuria was established in Ireland in 1966.[1] Screening is based on a bacterial inhibition assay which was instituted initially by Guthrie.[2] This test depends on detecting a high level of phenylalanine in the blood and is carried out on the new born infant between day 3 and day 6 of life. The time for doing the test is aimed at picking up the largest number of infants who will have been established on a protein containing food as early as possible after birth. If the test is carried out before three days there is a possibility that the level of phenylalanine may not be elevated sufficiently to be detected by the bacterial inhibition assay test. If the test is carried out later than six days the blood level of phenylalanine may have become so elevated that there may be a deleterious effect on the infant's brain.

Guthrie screening has been internationally successful but is dependent on accurate methodology in taking the sample, labelling accurately the sample so that the infant can be identified, ensuring that the blood sample reaches the Screening Laboratory and subsequent notification of positive cases to the individuals looking after the infant.

The treatment for phenylketonuria has been one of the major successes in medicine since it was first instituted in 1953.[3] There is substantial evidence to show that the earlier treatment is started and the better the biochemical control throughout life the better the prognosis and the outcome will be for the affected individual.

Since 1966 other conditions have been added to the Screening Programme. The conditions are:

Galactosaemia 1972
Maple Syrup Urine Disease 1972
Homocystinuria 1971
Hypothyrodism 1979

Galactosaemia is a condition which arises as a result of a deficiency of the galactose – 1 – phosphate uridyl transferase enzyme. This condition occurs in an Irish population with an incidence of 1 in 22,000. It is particularly common among the itinerant or traveller community. As a result of the enzyme deficiency galactose builds up in the blood and galactose – 1 – phosphate within the red cell. Gal – 1 – p is an extremely toxic chemical which can cause a chemical hepatitis. This results in a coagulopathy and the infant may die within the first few days of life from a haemorrhage into the brain. The affected child may also present with jaundice, food intolerance or cataracts.

Most countries do not screen for galactosaemia as it is a condition which may present acutely before the results of the routine screening becomes available. In our community, however, the high incidence has justified the screening and the results of screening are often available before the clinician has considered the diagnosis. Ideally, galactose screening should be carried out on the second or third day of life, but this would be impossible to institute and so it has been piggy-backed onto the phenylketonuria screening. The method depends on detecting a high level of galactose in the blood and is therefore dependent on the child having some lactose or galactose containing feed. Lactose free feeds make this population screening method for galactosaemia ineffective.

Clinicians should never depend upon the general population screening for the diagnosis of galactosaemia, but should query this condition in any child who presents early with jaundice and other symptoms e.g. vomiting or floppiness. Similarly in a family where one child has been identified a *high risk screen* should be carried out e.g. the new born infant in a family already identified as having this condition should never be given a lactose-containing feed but should be fed from birth on a lactose and galactose free food or glucose and water until the specific test for the transferase enzyme has been carried out and shown to be normal. This test is called the Beutler Test and is available from the National Neonatal Screening Laboratory in Temple Street Hospital.

Meanwhile the routine population screening for galactosaemia has been successful in this country primarily because of the success of the phenylketonuria screening and children who have been identified by the galactose screening have been established on diet and have a reasonable prognosis for normal development.

The third condition screened for is **Maple Syrup Urine Disease.** This is a defect of amino acid metabolism in which three amino acids cannot be broken down by the affected baby. The incidence in Ireland is 1 in 110,000, and incidence alone would not justify the screening but there is a pocket of affected individuals in Ireland and the screening has been justified on the basis that chronic handicap and death have resulted in families with this condition where it has gone undetected. The amino acids involved are leucine, isoleucine and valine and they accumulate in the blood of the affected infant following the establishment of feeding towards the end of the first week of life. This accumulation results in severe brain damage in an infant who presents with signs of sepsis or collapse. A diet similar in constitution to the diet for phenylketonuria but with low levels of the offending amino acids is commenced urgently. Normal neurological development and good health result from establishment of diet early and life long adherence to the diet.

The fourth condition screened for is **Homocystinuria.** This condition occurs in the Irish population with an incidence of 1 in 49,000. Methionine is the essential amino acid which cannot be changed into cystine as a result in deficiency of the enzyme cystethionine synthetase. Methionine accumulates and is changed into homocystine which damages the endothelium of the blood vessels and also predisposes the individual to thrombus formation. The child can die in early life from intracranial bleeding or thrombotic episodes but may present later in life with dislocation of lenses, osteoporosis and tall stature. Severe mental handicap can result. The screening detects the high level of methionine. This is one of the more difficult conditions to screen for as methionine is low in most baby foods, particularly breast milk. Once the baby has been identified a diet similar to that used for the other two amino acidopathies is instituted with a very good outcome. In this condition the amino acid methionine is restricted and the amino acid mixture which the child takes is augmented with cystine which the individual cannot make. Diet is necessary for life.

Hypothyroidism has been screened for in Ireland since 1979. This is an endocrine condition which results from failure of the thyroid gland to produce thyroxine. As a result of this the pituitary secretes concentrations of thyroid stimulating hormone and this high level of TSH is the metabolite which is detected by immunofluorescence in the thyroid screening. The incidence in Ireland is 1 in 3,500, and early detection allows early treatment. This test is not dependent on the intake of food because of the methodology involved.

All of the above conditions fulfil the criteria which were set out internationally for newborn screening. These are that:

1. The conditions are treatable.
2. There is a test available which is easily applied to large population groups.
3. There are few false positives and false negatives i.e. the test is reliable.
4. The incidence of the conditions in the community is sufficiently high to warrant screening.
5. The cost of the screening makes the process cost-effective.

In all of the conditions profound handicap or death would result without early treatment. The newborn screening allows for early detection and thus for early treatment. Some of the conditions e.g. galactosaemia benefit from the earliest detection possible and it is hoped in this situation the screening will augment the clinical efforts in making the diagnosis. The other four conditions require a few days for the abnormal metabolite to accumulate in the blood and are therefore very suitable for the Guthrie Card Screening mechanism. In all of the conditions early detection may save the child's intellect or life and will allow treatment to commence which will be continued for life.

REFERENCES

1. Cahalane, S. F. (1968): Mass screening of newborns in Ireland. Arch. Dis. Child. 43, 141-144.

2. Guthrie, R. (1961): Blood screening for phenylketonuria. J. Am. Med. Assoc. 178, 863.

3. Bickel, H., Gerrard, J. & Hickmans, E. M. (1953): Influence of Phenylalanine intake on Phenylketonuria. Lancet 2, 812-813.

APPENDIX 2

Front of Card

NEONATAL SCREENING CARD

MOTHER'S SURNAME:

LAB No.

BABY'S FIRST NAME:

MOTHER'S FIRST NAME:

SEX M F

ADDRESS:

HOSPITAL: WARD

HOSPITAL NO.:

COMMUNITY CARE AREA:

	D	D	M	M	Y	Y
DATE OF BIRTH:						
DATE OF COLLECTION:						

GESTATIONAL AGE (   ) WKS.

PLEASE TICK	YES	NO
ANTIBIOTICS?		
TRANSFUSION?		
SOYA BASED FEEDS?		
BREAST FEEDING?		
REPEAT?		

TEST TAKEN BY:

DOCTOR/G.P.:

ADDRESS:

FILL CIRCLE WITH BLOOD THROUGH FROM REVERSE SIDE

Back of Card

NEONATAL SCREENING CENTRE
CHILDREN'S HOSPITAL, TEMPLE STREET, DUBLIN 1.

F № 99036

INSTRUCTIONS

PLEASE NOTE: —
A SEPARATE REPORT WILL BE POSTED TO HEALTH CENTRES IF THE APPROPRIATE ADDRESS IS SUPPLIED.

N.B. The blood specimen should be taken not earlier than 72 hours after the infants birth and when feeding is established.

1. After skin is sterilised, puncture with sterile disposable lancet.
2. Bleeding is facilitated by making limb dependent, and warming foot.
3. **Completely fill each circle** by soaking blood through from this side.
4. Please allow sample to dry completely at room temperature before posting.

APPENDIX 3

SCREENING FOR METABOLIC DISORDERS INSTRUCTIONS AND ILLUSTRATION FOR TAKING BLOOD SAMPLE

1. The blood sample should be taken not earlier than 72 hours and not later than 120 hours after the infant's birth and when feeding has been established.

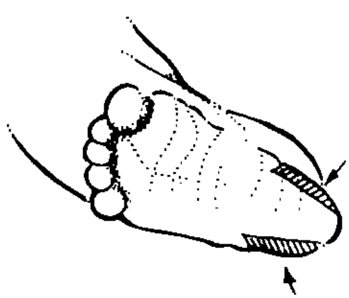
2. In a gallipot of warm water, heat the infant's heel.

3. Rub the skin for 1-2 minutes to increase blood supply.

4. Puncture the skin with sterile disposable lancet, depth 2-3mm only.

5. Gently massage the heel to encourage blood flow.

6. The blood drops are to be soaked through to the rear side of the neonatal screening card, filling all circles fully.



APPENDIX 4

METABOLIC SCREENING PROGRAMME
SUMMARY RECOMMENDATIONS ON USE OF NEW TECHNOLOGY

1. The Current Systems

1.1 The National Neonatal Screening Laboratory uses a proprietary laboratory package, called DATAPLUS*2/PC which runs in a MUMPS environment. Details of all Guthries are stored on this system. The package has been very reliable in maintaining the laboratory records and issuing standard results reports.

1.2 The Coombe, Galway Regional, and Letterkenny hospitals have computerised birth registers which have all births on file soon after birth. The Eastern and North-Western Health Boards have computerised registers.

2. Limitations of Current Systems

2.1 There is no linkage with hospital birth registers or with the Birth Notification System (**BNS**). The case reference numbers on the Guthrie are not those used for the **BNS**.

2.2 The laboratory package has limited reporting options and there are no reports on coverage or timings.

2.3 The laboratory package does not hold indicators to distinguish Guthries taken in the community, or those arising from repeat requests, unrequested repeats, or review tests on known positives: so none of these can be analysed or excluded from an analysis.

2.4 The laboratory package cannot use combined fields to generate unique codes e.g. hospital plus case reference.

2.5 The variety in hospital procedures is potentially crippling for national systems. At the basic level of case reference number there are problems including the use of the maternal chart number.

2.6 The Birth Notification System is not on-line early enough to serve as a register for the Guthrie screening.

3. Query Positive Cases

3.1 The response to these cases is immediate and direct, contact is made to all concerned by telephone.

4. Repeat Requests

4.1 It is important to minimise the time taken to collect a repeat sample when the laboratory fails to get a clearresult from the first sample. The volume of these is low enough to allow the use of the telephone. A call to the liaison person at the hospital should ascertain the location of the baby. This would then allow the faxing of formal requests to the hospitals and SPHN areas: fax could be automated and run overnight by the computer.

4.2 Many repeat requests now arise from 'no growth' caused by antibiotics: hospitals know this and routinely send repeat specimens; these could be classified as a separate category to reduce the number of repeat request calls.

4.3 The reporting of negative results for repeat cases should either be separate from the main negative reporting or be done so as to highlight these cases.

5. Negative Results

5.1 Negative results are reported separately. Their critical function is to allow the marking of registers to reveal those infants awaiting a report and ensure that none are missed.

5.2 Changes are desirable in format, sequence, and timing. There should be one line per case rather than one page per case. The sequence and some of the content should be hospital specific: some may want a strict number sequence, while most will want a date of birth sequence. In timing there should be the option of immediate ongoing reporting of negatives and a regular aggregate report.

5.3 A weekly report of all cases would greatly reduce the hospital task of checking the birth register. The report could show all results, with anomalies and results of repeats clearly marked. This could separate the response to repeat requests from the identification of possible misses.

5.4 If reports are delayed the laboratory could forward a partial report or receipt list which would serve the same checking function as the normal list.

6. Hospital Systems

6.1 A small number of hospitals already have computerised registers. **Linkage with these should be possible once the use of case reference numbers is consistent and harmonised.** The hospital should send the screening laboratory all births, within a day of birth. This would save the duplicate keying of reference information and should reduce errors in the identification of cases since all cases from such hospitals should find a match already on the laboratory machine. It would also allow the laboratory identify cases not yet received.

6.2 The reporting of cases for such hospitals should be on media or machine to machine.

7. Other Registers

7.1 While the development of child registers varies from area to area and the time taken to complete these has ruled them out as mainsprings in the screening programme there should be linkage to them. This would allow post-hoc confirmation that coverage is complete and highlight to the health board what kinds of delays are occurring.

8. Birth Notification

8.1 A recent study of the registration of births, deaths and marriages recommends the use of local microsystems to collect and validate the birth data at the hospital. The Department of Health plans to pilot such a system. This system should be able to provide a register of births early enough to be used as described above – sending an initial file to the screening laboratory, receiving results from the laboratory, and identifying delayed cases.

9. Immediate Steps

9.1 The laboratory has already moved their system to a more powerful machine which eliminates some of the previous holdups. Data can again be entered while reports are being printed. In addition, this machine can hold all the data on line for three years.

9.2 Laboratory staff are also assessing a report generator. This will provide lists and analyses for monitoring the scheme. Software is also available from the same supplier which will allow some extra flexibility in reporting within the laboratory package.

9.3 A more detailed analysis of the data would enable the laboratory assess some of the anecdotal claims about delays. This would allow the laboratory to respond with statistical measures and exceptions.

10. Longer Term Action

10.1 If the laboratory is to actively monitor the programme it needs systems designed for that purpose as distinct from managing the laboratory function per se.

10.2 It seems unlikely that the current laboratory system can meet the changing requirements. Key features in its upgrade or replacement should be linkage to existing and planned registers, a flexible modern user interface, and powerful report generator. Additional items of information should be held on file, e.g. second surnames, diet. Automation of some other tasks including fax operation will save effort.

AN ROINN SLÁINTE
DEPARTMENT OF HEALTH