

## PRACTICE METHOD

### A METHOD FOR THE DETECTION OF CONGENITAL ABNORMALITIES

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THERE ARE NO OCCASIONS IN a doctor's life when the responsibility upon him is so great or the opportunity for early diagnosis so golden, as when he attends the birth of a baby. He has two lives in his hands. The awesome responsibility for ensuring the safe delivery of both is matched by another; that of detecting congenital abnormality.

Little more than a decade ago the majority of congenital abnormalities were of scarcely more than academic interest. If they were discovered early the prognosis was unlikely to be much better than if they were not. Tremendous advances in neonatal surgery and our enlightenment about treatable conditions like phenylketonuria have changed all that. Now it is beholden on all who do midwifery to search for and find any structural, functional or biochemical abnormality at the earliest moment and to take effective action as a matter of urgency.

#### *A special record card*

In order to make this task easier, we in Bridgnorth have evolved a card that serves the dual purposes of permanent record and *aide-memoire* to screening procedures in the neonatal period. It has been on trial for a year and is easy to use. It fits neatly into the N.H.S. record folder so that in our practice a clear summary of birth and the crucial first month of life forms a permanent part of every young patient's medical chronicle. The importance of this becomes clear when a problem of development arises later in childhood.

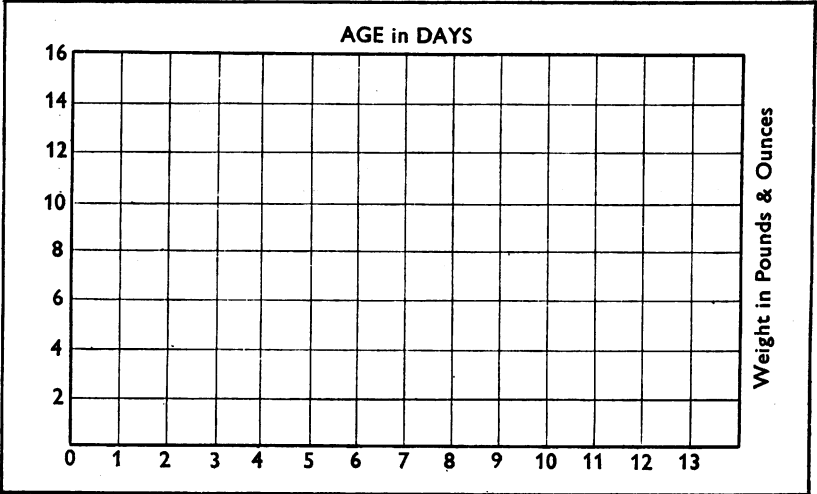
The design of the card is shown in figures 1 and 2. On the face, beneath identification details, is a chart for recording weight gain which is a sensitive pointer to faults in metabolism or management. Beneath this is the screening check on reflex activity and biochemical tests in common use.

The examination of an infant's reflexes is important in detecting brain damage and the persistence of primitive reflex activity or lateral asymmetry is likely to be of significance.

The biochemical tests ensure that disease due to rhesus incompatibility, congenital diabetes mellitus and phenylketonuria are not overlooked. Although the test for the latter is electively done at six weeks and there-

NEONATAL EVALUATION

Name:	B.W.	D.O.B.
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REFLEX ASSESSMENT		Date		
ROOTING	<input type="checkbox"/>	L	R	
MORO	<input type="checkbox"/>			KJ
GRASP	<input type="checkbox"/>			Biceps
SUCKING	<input type="checkbox"/>			Triceps
TONIC NECK	<input type="checkbox"/>	Walking Reflex		

BIOCHEMICAL ASSESSMENT			
Blood Tests		Urine Tests	
Haemoglobin .....		Clinistix .....	
Coombs Test .....		Phenistix .....	
Mothers Rh. ....			

Figure 1.

FUNCTIONAL ASSESSMENT			
APGAR SCORE:			
STRUCTURAL ASSESSMENT			
HEAD	{	Skull Circumf. <span style="float: right;">Fontanelles <input type="checkbox"/></span>	
	{	Palate <input type="checkbox"/> Nares <input type="checkbox"/> Other <input type="checkbox"/>	
Cardio-Respiratory			
		Lung Aeration <input type="checkbox"/> Cardiac Bruits <input type="checkbox"/>	
		Femoral Artery Pulsation <input type="checkbox"/>	
G.I.T.			
	{	Oesophagus <input type="checkbox"/> Abdominal wall <input type="checkbox"/>	
	{	Anus (P.R.) <input type="checkbox"/> Hernial orifices <input type="checkbox"/>	
	{	Meconium <input type="checkbox"/>	
Genitalia			
	{	Phallus <input type="checkbox"/> Labia <input type="checkbox"/>	
	{	Gonads <input type="checkbox"/> Scrotum <input type="checkbox"/>	
Spine: (Esp. Spina bifida) <input type="checkbox"/>			
Lower Limbs: (Esp. CDH & Talipes) <input type="checkbox"/> Digits: <input type="checkbox"/>			
Upper Limbs: (Esp. Digits) <input type="checkbox"/>			
SUMMARY OF ABNORMALITIES:			
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.....			
.....			
COMMENTS ON DELIVERY:			
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.....			

fore not strictly in the neonatal period, it is convenient to include it in the record of neonatal life.

As nearly as possible to one minute after birth, when the mother has been made safe and the infant's airway cleared we evaluate and record cardiorespiratory function (Apgar 1953). Experience shows that infants with an Apgar score of less than 7 require especially careful attention and that if the score is 5 or less, resuscitation is urgently needed.

The least difficult phase of neonatal assessment is physical examination for structural abnormalities. This follows a systematic routine and, with practice, takes less than two minutes to perform. Two especially important tests are the measurement of skull circumference and the testing of oesophageal patency by the passage of a rubber catheter. The latter is essential to the early recognition of oesophageal atresia and the former helpful in evaluating development progress in later childhood. A summary of abnormalities and comments on the delivery completes the record.

Finally, the card is transferred to the child's N.H.S. file so that for ever after we have a permanent note of the most noteworthy of all events; birth and the start of a new life.

### Results

During 1964 my partners and I were responsible for the delivery of 274 infants in our maternity unit and 34 in their own homes. There were no stillbirths, three premature infants and seven cases of congenital abnormality. The latter group comprised one case of oesophageal atresia, one of multiple defects including polycystic kidneys, one of severe pulmonary artery stenosis, one of polydactyly, two of talipes and one with a naevus. All these defects were recognized within the first 12 hours of life and two infants were transferred to a specialized unit for surgery.

The infant with oesophageal atresia diagnosed at birth, unfortunately died after operation. The infant with pulmonary stenosis met a similar fate, and the infant with multiple defects died within four hours of birth. These results are disheartening but we feel reassured that diagnosis was prompt and every chance of successful surgery was given at the earliest opportunity.

The infants with less serious abnormalities were treated by us and with the help of our visiting surgeons and physiotherapists at Bridgnorth Infirmary.

### Summary

A method of screening the newborn for congenital abnormality has been evolved. A special card is used as an *aide-memoire* and permanent record of neonatal period. The results of using this for a year are summarized and the importance of early diagnosis emphasized.

### Acknowledgements

I am indebted to my partners for their enthusiastic co-operation in evolving this method of neonatal screening and to Dr J. C. Macaulay, consultant children's physician at the Royal Salop Infirmary for much encouragement and helpful advice. Nicholas Laboratories Limited of Slough undertook the printing of prototypes of the record card and have produced a large quantity of the final

design. Without their help this work would not have been possible.

#### FOOTNOTE

Since this work was begun a national scheme for the central recording of congenital abnormalities has been introduced. It is interesting to note that the incidence of abnormalities in our small series corresponds with that revealed by this scheme (Ministry of Health, 1964). Thus the figure of 24 per 1,000 total births (live and still) for the country as a whole approximates to ours of seven in 308 births, neither of which is unexpectedly higher than the value of 15 per 1,000 derived by Lamy and Frezal (1961) from several surveys of 'major' abnormalities detected soon after birth.

During 1964 the County Medical Officer of Health for Shropshire was notified of 129 congenital abnormalities occurring in 95 children out of a total of 6,021 born (O'Brien, 1965).

Thus congenital abnormalities can no longer be regarded as rare curiosities devoid of practical significance. They must rather be sought out meticulously as common, in many cases amenable to treatment and in all worthy of note.

#### REFERENCES

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## A SYSTEM OF RECORDING INFORMATION IN GENERAL PRACTICE

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WHEN A PATIENT IS REFERRED to a consultant, the general practitioner often summarizes the relevant past history and treatment; this paper describes certain conventions and in this paper in order to simplify this process only National Health Service cards and folders are mentioned but the principles may suit any list.

#### Letters

All letters concerning a patient were kept together in the folder (E.C.5, 6). Each letter was filed so that the information was outermost (if folded) and the date facing the front of the folder; if folding was necessary the letter was folded once, or at most, twice with all edges dressed in line, so