REFRESHER COURSE FOR GENERAL PRACTITIONERS

EARLY RECOGNITION OF CONGENITAL DEFECTS

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Infants with congenital defects may be grouped as (a) those presenting an obvious anatomical lesion at birth—for example, cleft-lip or mongolism; (b) those presenting an anatomical lesion not immediately recognizable but discovered by routine examination—for example, congenital heart disease and imperforate anus; (c) those presenting with a functional disorder resulting from an anatomical defect the recognition of which may be delayed until the organ in question is put to the functional test—thus in oesophageal atresia vomiting will occur after the very first feed, a short oesophagus may not be recognized until later, and a congenital defect of the hip may not be diagnosed until the child attempts to stand; and (d) those presenting with an anatomical lesion made manifest by the onset of infection or, more rarely, of malignant change—for example, infective endocarditis and malignant change in an ectopic testis.

However, with the recent advances in our knowledge of the causation of some of the congenital defects an alternative grouping from the aetiological point of view might be (a) those of hereditary or familial aetiology—for example, achondroplasia and acholuric jaundice; (b) those due to noxious influences on the developing foetus—for example, maternal rubella and toxoplasmosis; (c) those due to intrauterine influences—for example, some cases of talipes; and (d) those due to maternal dietetic deficiencies, evidence for which is largely in the animal experimental stage.

Thus it might be said that some defects can be expected, some suspected, and that others are latent. If the practitioner is to prepare himself for the possibility of a baby having a congenital defect he must take a full antenatal history, make an adequate antenatal examination of the foetus as well as of the mother, and undertake a complete examination of the newborn baby.

Antenatal History

In taking the history there are two main lines of inquiry, the first into the family stock, and the second into the health of the mother. The family history must elicit the presence of any congenital defects in members of the families of both parents and information about their consanguinity. If blood relations marry there is a greater risk that both partners may be carriers of a recessive gene. A sex-linked defect may be revealed, for example, by a history of pseudohypertrophic muscular dystrophy in the male members of the mother's family.

Since the work of Gregg and Swan in Australia it seems to be well established that rubella occurring in the first three months of pregnancy may cause harm to the developing foetus, with subsequent disorder of its growth and development leading to such conditions as cataract, deafness, and possibly congenital heart disease and cerebral agenesis. This discovery has opened a vast field for exploration, but so far no other virus disease in the mother has proved to be a responsible factor for congenital defects; nevertheless the conception of foetal

damage as the result of a maternal viral infection must be regarded as established. The probability of one of the defects, particularly cataract or deafness, following maternal rubella in the first three months of pregnancy is high, and some obstetricians have even suggested that it is an indication for terminating the pregnancy. To justify this, however, there must be no doubt of the diagnosis of rubella and the infection must be within the first three months. Even then it should be remembered that the only legal indication for terminating is related to the health of the mother and not to the health of the foetus.

Another condition which, though rare in this country, leads to foetal changes is toxoplasmosis due to a parasitic involvement of the developing nervous system. The mother is the carrier of the disease and is usually unaware of its existence; when active in the early weeks of pregnancy the disease has been found to be responsible for such states as microcephaly, hydrocephalus, intracranial calcification, and eye changes. Skin testing may detect the condition in the mother in early pregnancy, but unfortunately there is no known method of treating this state of affairs.

It seems fairly well established from experimental and clinical evidence that radium or deep x-ray therapy applied to the uterine field during the early weeks of pregnancy may result in the baby being born with an anatomical defect affecting particularly the skull and nervous system.

Maternal dietetic deficiencies have not so far been shown to be clearly related to foetal abnormalities in man, but in animals on deficient diets, especially of vitamins A and D and riboflavin, foetal changes have occurred, and although these results may not be strictly applicable to human development they at least suggest a possible cause of defect which needs further study. In this respect iodine deficiency has long been known to play a part in the causation of cretinism in the baby.

The age of the mother and her parity must also be noted, for they may have some bearing on the possibility of a congenital defect. The classical example of this, though there are many exceptions, is the mother of a large family who produces a mongol at the end of her child-bearing age.

Finally, antigenic incompatibility, especially to the rhesus factor, though not producing any direct developmental change, may result in such conditions as kernicterus and familial cirrhosis of the liver, which in the past were thought to be due to direct genetic causes. The occurrence of previous pregnancies or miscarriages, or of jaundice or anaemia in the babies, should therefore be noted and the mother should be asked whether she has previously had any blood transfusions.

Antenatal Examination

It should be remembered that congenital defects are commoner in abnormal presentations, in premature

births, and when hydramnios is present. In examining the mother, therefore, attention should be directed to these points. A persistently abnormal lie should call for a radiological examination, which, with good films and expert interpretation, may show such changes as anencephaly, achondroplasia, or other bony dyscrasias. If a gross abnormality of this sort is discovered the practitioner may be well advised to warn the parents that he is not altogether happy about the future of the child or he may prefer to pass the problem on for a specialist opinion.

Congenital heart disease may be suspected and sometimes diagnosed with confidence if a murmur is heard in the foetal heart sounds, though it is impossible to be certain of the exact lesion. Thyrotoxicosis has been diagnosed in the foetus with a very rapid heart rate in a severely thyrotoxic mother, but such diagnostic feats are few and far between.

Immediate Examination of the Newborn Baby

Examination of the newborn baby is sometimes perfunctory, and indeed after a difficult midnight delivery the doctor may well postpone a detailed examination until the morning. He should, however, satisfy himself that no immediate catastrophe is imminent from any congenital defect and that no obvious deformity will be found by the mother before him. As a first measure he should note the presence or absence of cyanosis and determine its cause in relation to the heart. As a second measure he should determine, so far as is possible, that the alimentary tract is patent. This may not be easy, but if the baby has not passed meconium he should look to see that the anus is open, for an imperforate anus calls for immediate attention. Other obstructive conditions in the alimentary tract may not manifest themselves until feeding is begun. Finally he should look inside the baby's mouth, otherwise he will miss a cleft-palate and his reputation may not unnaturally suffer.

Detailed Examination of the Baby

In the more systematic examination it is useful to start with the skull, for more frequently than elsewhere in the skeleton the skull shows changes in its shape, and such conditions as microcephaly may indicate an underlying cerebral agenesis. The general shape and size of the skull should be noted and also the character of the sutures and fontanelles. The maximum circumference of the normal skull is $13\frac{1}{2}$ in. (34.3 cm.), but its shape may be temporarily modified by the caput succedaneum. The posterior fontanelle is normally almost closed, but the anterior fontanelle is open, with the sutures easily palpable. The size of the anterior fontanelle in particular and the width of the sutures may vary considerably within normal limits.

The abdomen needs careful palpation, though inspection in a good light is often more informative. Most causes of intestinal obstruction which need surgical relief will give rise to vomiting, visible peristalsis, abdominal distension, and failure of bowel action. If obstruction is suspected, as it may well be if vomiting persists from birth, a straight x-ray film of the abdomen and a study of the air shadows are more helpful than a lipiodol swallow and should always be done first.

A newborn baby's first feed should be carefully observed. Sudden distress, with cyanosis and frothing at the mouth, suggests an oesophago-tracheal fistula, which

calls for immediate surgery. It is the wise practice in many nurseries for the newborn to be given only sterile water or saline for the first feed, until the continuity of the oesophagus has been established.

Cyanosis, which may be cerebral, respiratory, or cardiac in origin, requires careful classification. If of cerebral origin it will tend to vary and to be accompanied by a greyish colour in a shocked ill-looking baby, and there may well be other cerebral signs such as twitching or stiffness of the limbs, a tense fontanelle, and vomiting. Cyanosis of respiratory origin will vary with the crying of the baby and improve with oxygen therapy: on the other hand cyanosis of cardiac cause will be more constant, will not respond much to oxygen, and will tend to increase when the child cries.

It is by no means uncommon for a baby with cyanotic heart disease to have no audible cardiac murmurs; this is sometimes forgotten. Persistent deep cyanosis is seldom due to anything but congenital heart defects. Although murmurs may be absent, often the heart sounds are tick-tack in character and the heart rate is rapid. Cyanosis and recession of the ribs during inspiration are more important as an indication of the severity of the condition than the presence or absence of a murmur. The common systolic murmur of a non-cyanotic heart lesion, especially those due to septal defects, is often missed at birth because its harshness and timbre are almost identical with that of the breath sounds in a newborn baby. It therefore requires careful listening to separate and distinguish such a bruit.

The genitalia need careful inspection, and the site and patency of the anus and urethra must be determined. The pseudo-hermaphrodite may suddenly develop a suprarenal crisis with vomiting and collapse, and such cases should not be dismissed too lightly as a future problem for the plastic surgeon. Most minor degrees of defect in the male, such as hypospadias, are best left for surgical repair until the child is older, but the mother must be told the reason for delayed treatment. In the female an ectopic anus may not be noticed at first, but it is best dealt with as soon as the child's feeding is established and she is gaining weight satisfactorily, but before mixed feeding is begun.

Most limb deformities are obvious and visible—for instance, polydactyly or syndactyly—but dislocated hips may pass unobserved for some time. Talipes, which in some cases, especially of the equino-varus type, is probably due to intrauterine posture, needs early treatment, and these cases should therefore be referred to an orthopaedic surgeon within the first two weeks. Talipes associated with a meningocele and spina bifida is a different problem, for then the integrity of the nervous system, as judged by the muscle tone of the legs and the shape of the skull, is the overriding factor in determining the treatment. Most meningoceles are best left without surgical removal for at least a year, and this is certainly so if the lower limbs are flaccid.

Examination of the eyes and ears is difficult but important. Such defects as cataract or retrolental fibroplasia in premature babies may easily be missed unless carefully looked for. Deafness is impossible to detect in the early months. As a rule it is delay or difficulty in speech that first arouses the mother's doubt about her child's hearing, and it is surprising how late parents may be in suspecting this disability. It is often more helpful to watch the mother unconsciously using sign language to her child than to watch the child.

There are, in addition to gross structural changes in particular organs, certain general congenital conditions which may be recognizable at birth. The most important, perhaps, of these, because it is the most frequent, is mongolian idiocy, which unlike cretinism can and should be diagnosed at birth in almost every instance. There is still no known cause for this tragic condition, but it would seem by analogy that it might be due to some noxious influence on the foetus in its early development. However, no direct evidence for this has been forthcoming, and we are still left with the view that it is related to chromosome changes associated with the age of the parents as the only clue to its aetiology.

Latent Defects

There are many congenital heart lesions that pass unnoticed until later in life, as every school medical officer can testify. Most of the children with these lesions are without symptoms of their disability, as, for example, those with a patent ductus arteriosus or the rarer coarctation of the aorta. These are commonly first found at routine examination, and so far as these two particular defects are concerned the first is usually missed because the child is not undressed and the stethoscope does not reach the pulmonary area, and the second is missed because the femoral arteries are not palpated as a routine. A patent ductus is relatively common and has assumed greater importance because it can be cured by ligation. The optimum age for this is between 7 and 9 years, but if there are symptoms of distress at an earlier age there is no need to delay surgery. The development of infective endarteritis is an immediate indication for operation. In this country we have hardly had enough experience of the surgical treatment of coarctation of the aorta in children to be able to lay down an optimum operation age, and decision must depend in each case on such factors as the symptoms, size of the heart, blood pressure, etc.

Perhaps the most difficult of the latent defects to recognize is mental deficiency. In cases associated with obvious physical signs, such as in mongols, there is seldom difficulty, but in others an associated physical defect may either not be recognized or its significance not be appreciated. In such cases there may also be difficulty in distinguishing the congenital or genetic defects from those that are acquired or due to environmental influences, as in some instances of cerebral palsy. In order to find these defects as early as possible the doctor must keep the newborn baby under frequent review, especially noting for this purpose his developmental progress. Gesell's studies have greatly helped us to appreciate the early developmental delays and deviations that portend poor mental progress.

Just as some congenital defects are recognized only by careful routine examination, others force their attention on the doctor by some functional or symptomatic disorder, often precipitated by infection. There are many examples of this; for instance, the development of infective endocarditis is the signal that there has been an underlying but latent congenital heart lesion; or an intractable diarrhoea and spasmodic cough in a baby with a good appetite but a coeliac contour suggest that the disease is due to congenital cystic changes in the pancreas and a congenital genetic dysfunction of the mucus-secreting glands. Other examples of conditions in which infection first attracts attention to a congenital defect are cystic changes in the lungs or kidneys, for so long as

infection is kept at bay quite gross deformity may pass unnoticed. It is true to say, especially of boys, that a persisting urinary infection nearly always indicates some congenital abnormality of the renal tract, and an intravenous pyelogram will probably be needed to disclose it.

What to Tell the Parents

Finally, it may be helpful to discuss the very practical problem of when and how much a practitioner should tell parents when he suspects or finds a congenital defect. This is never an easy decision to have to make. If the family history in this respect is a bad one the mother may already have had her suspicions aroused, and it is then as well to warn her if these suspicions are justifiable, for if the doctor is right in his prognostications the mother will be less disturbed by their realization and will be more able to accept the inevitable, but if he is wrong then she surely will be delighted. As in all comparable situations the way in which the news is likely to be received depends on the temperament and intelligence of the mother and father and on the manner of its telling by the doctor. However, if at birth a congenital defect is obvious the doctor should have no hesitation in presenting the known facts. If he is in doubt about the extent or significance of the findings he may justifiably modify his statement until the findings have been confirmed, but it is kinder to the parents, and it will earn him greater respect in the end, if he tells them of his suspicions as soon as he can. It is never an easy task for either party, but its tactful and smooth achievement may make all the difference to the future of the family.

Thus it will be seen that congenital defects may present themselves in the antenatal period, or immediately or soon after birth, or they may not be manifest for some years. The doctor should always be on the alert to detect them, for not only may some of them be amenable to surgery but with their recognition the child's life can be adjusted accordingly and the family life and happiness, so often in jeopardy when a deformity is found, safeguarded and preserved.

In an article in the *Journal of the American Medical Association* (August 25, p. 1589) Dr. John Flumerfelt draws attention to some of the problems raised when a physician wishes to refer a patient to a psychiatrist. Apart from the rather suspicious attitude towards psychiatrists still adopted by many otherwise well-informed people, there are certain other difficulties mainly arising out of the psychological effect that such a decision may have upon the patient, who may, for instance, think that "neurosis" means insanity, or that there is some weakness of character implied by the need for psychological treatment. On the whole these objections are made up of feelings of guilt, caused by impulses of which the patient is unaware. Even when the patient agrees to undergo such treatment the presence of these feelings must be borne in mind. Many patients require some help from the referring physician, and he is obviously the person best qualified to help, as he knows most about the patient. Dr. Flumerfelt stresses the importance of always telling the patient that the doctor to whom he is being referred is a psychiatrist, however opposed to this he may know the patient will be; if he does not tell him it is likely that the patient will erect such a defence against the psychiatrist that nothing will be achieved. Much delicacy must be exercised by the doctor when he suspects that the trouble is of mental origin, or the patient's condition may easily be made worse. In fact, it must be remembered that psychotherapy has already been started when a patient goes to his doctor for help, and continues when the doctor advises him to see a psychiatrist.