The First Year of Life

H B VALMAN

SOME CONGENITAL ABNORMALITIES

Myelomeningocele

A myelomeningocele is a flat or raised neural plaque in the midline over the spine due to abnormal development of the spinal cord and associated deficiency of the dorsal laminae and spines of the vertebrae. It is usually found in the lumbar region. The absence of the various coverings which normally protect the cord allows meningitis to occur easily. If there are no active movements in the legs and the anus is patulous the infant will probably be incontinent of urine and faeces for life and never be able to walk unaided. Thoracic lesions and kyphosis are signs of poor prognosis.

Infants with a good prognosis need urgent treatment, so all affected infants should either be seen by a consultant paediatrician without delay or sent to a special centre, where selection for surgery can be made. About 10% of the infants have surgery as a result of this policy. During the first operation the lesion on the back is covered by skin.

Most of these infants develop progressive hydrocephalus later and those considered suitable for surgery require insertion of a catheter with a valve from a cerebral ventricle to the lower end of the superior vena cava to reduce the cerebrospinal fluid pressure.

Progressive hydrocephalus is confirmed by measuring the circumference of the head at its largest circumference (occipitofrontal) every three days with a disposable paper tape measure, plotting these values on a growth chart, and showing that the head is growing faster than normal.

Raised concentrations of α -fetoprotein are found in the amniotic fluid when the fetus has an open myelomeningocele or anencephaly. In anencephaly there is absence of the cranial vault and most of the brain, and the infant dies before or shortly after birth. In some hospitals *maternal plasma* α -fetoprotein levels are measured in all mothers at the 14-16th week of gestation. The *amniotic fluid* concentration is estimated and an ultrasound examination is performed in the mothers with a raised plasma value who wish to have these investigations and are prepared to have a termination of an affected pregnancy.

Microcephaly

The signs of microcephaly are a small head and a forehead that is particularly small in relation to the face. The diagnosis is confirmed by showing a small head circumference in relation to the baby's weight and gestational age. It is invariably associated with mental impairment. Evidence of certain intrauterine infections such as toxoplasmosis should be sought to enable the parents to receive accurate genetic counselling.

Cleft lip and palate

Mothers are often severely disturbed by the appearance of the infants and may be reassured by seeing photographs of similar patients before and after their repair operations. Cleft lip and cleft palate are often associated. A cleft lip, caused by failure of the maxillary process growing towards the midline to fuse with the premaxilla, may be unilateral or bilateral. Minor degrees of cleft palate may be easily missed if the posterior part of the palate is not seen and palpated. Most of these infants feed normally from the breast or bottle. If there are feeding difficulties a special teat, a large normal teat, a special spade-like spoon, or an ordinary spoon may be tried. The lip is usually repaired at three months and the palate at one year. The value of an obturator before operative closure of the palate is controversial. Despite excellent operative results these children are prone to recurrent otitis media and problems with speech development.

Umbilical hernia

An umbilical hernia, usually containing omentum and gut is most common in African infants or West Indians of African descent. No treatment is needed, as the hernia usually disappears spontaneously by the age of 3 years, although in West Indian infants it may take a further three years.

In contrast to an umbilical hernia the sac of an omphalocele is covered by peritoneum but incompletely by skin. An omphalocele is a hernia into the base of the umbilical cord and contains gut and sometimes solid organs like the liver. Immediate transfer to a surgical unit is needed.

Oesophageal atresia

In a few units a tube is routinely passed into the stomach as part of the initial examination to exclude oesophageal atresia. In the absence of such a routine manoeuvre, the diagnosis will be suspected in any newborn baby who has a continual accumulation of frothy secretions in the mouth, sometimes with cyanotic attacks.

The first feed should be supervised and for breast-fed babies this should take place in the labour ward. In many units 5% glucose solution is given as the first bottle feed to reduce the dangers of choking and aspiration pneumonia from milk if oesophageal atresia and the associated tracheo-oesophageal fistula are present. Breast-fed babies do *not* need a first feed of glucose solution.

The diagnosis of oesophageal atresia is confirmed by attempting to pass a tube down the oesophagus. The tube used must be stiff enough to prevent coiling in the upper oesophageal pouch and should have a radio-opaque line so that the position can be checked by a chest radiograph. The tube should be aspirated every few minutes to keep the upper pouch clear until the infant reaches a specialised surgical unit.

Multiple abnormalities

Infants with multiple abnormalities should be examined and investigated by a paediatrician without delay to ensure correct management of the infant and to provide the information needed for genetic counselling. Some of the infants may have a recognisable syndrome and the investigations may show a chromosomal defect or evidence of intrauterine infection.

The most common problem is the infant with possible Down's syndrome (mongolism). Although a scoring system for a large number of clinical features has been used in the past, any infant with suspected Down's syndrome should have a blood chromosome analysis performed. The most useful features suggesting the diagnosis are the facial appearance with palpebral fissures which slant upwards and outwards, prominent epicanthic folds, and baggy cheeks. A flat occiput, poor limb tone, and single palmar crease are suggestive but not specific signs. About 2% of normal infants have a single palmar crease in one hand, and prominent epicanthic folds are even more common. If the infant has congenital heart disease it may be detected by performing an electrocardiogram as a definite murmur may not be present until several weeks after birth. All infants with Down's syndrome have mental retardation but it may be possible to reduce maternal depression and increase the infant's drive and ability to learn by introducing the parents to an occupational therapist in the neonatal period.

Dr H B Valman, MD, FRCP, is consultant paediatrician, Northwick Park Hospital and Clinical Research Centre, Harrow.

The photographs of cleft lip were reproduced by permission of Mr R Saunders.

Cot deaths

About one baby in every 500 born alive dies suddenly and unexpectedly between the ages of 1 week and 2 years. Typically an apparently healthy baby (or, occasionally, one with only minor symptoms) is put in his cot to rest and sometime later is found dead. Although an infant may be face down in the cot with the bedclothes over him, suffocation should not be assumed. Sometimes vomit, which may be blood-tinged, is found around the mouth or on the bedding, but regurgitation usually occurs after death and is not the cause of death.

In some cases necropsy discloses an unsuspected congenital abnormality or rapidly fatal infection. But usually there is no evidence of severe disease, though there might be slight reddening of the tracheal mucosa, which in other babies normally resolves spontaneously.

Parents often blame themselves and may worry that the infant suffocated as a result of neglect. They should be told that their feelings of guilt are a natural reaction, and the doctor should explain to them, and to anyone who was looking after the baby when death occurred, that cot death is a well-recognised but ill-understood condition and that no one is to blame for the infant's death.

Parents must also be told that because the death is of unknown cause the coroner will have to be told as a matter of routine and that he will arrange for a necropsy. If the parents want to see the infant's body, and they should be asked, the infant is clothed and a doctor or nurse should be present to answer questions and provide support. Discussion with the parents will determine who is best to help with their grieving. The Foundation for the Study of Infant Deaths (23 St Peter's Square, London W6 9NW, telephone 01-748 7768) produces a leaflet on cot death aimed at parents, which they may find helpful.

Once the results of the necropsy are known the family doctor should either explain them to the parents himself or arrange for a paediatrician to do this.