The best prospect of a permanent cure occurs in those cases seen in the first year of life and diagnosed early, where the tumour can be completely excised and is well differentiated.

Teratomas arising in other situations may be noticed at, or soon after, birth. This is particularly true of those arising in the thyroid, mediastinum, or stomach, and of the nasopharyngeal teratomas (epignathi). Teratomas of the testis or ovary, on the other hand, may present throughout childhood, and their histological structure is usually devoid of embryonic tissue.

Factors affecting the form of medullocervical dislocation deformity in relation to meningo-myelocele and spina bifida. J. L. Emery and N. MacKenzie. The Congenital Anomalies Research Unit, University of Sheffield, Thornbury Annexe, Sheffield 10.

Earlier studies indicated that with the Cleland-Arnold-Chiari deformity there is compression of the cranial segments of the cervical cord, a compression that is dispersed by the level of C8.

The present study is based on a complete dissection of 100 cadavers with meningo-myelocele and hydrocephalus. The lesions were first classified into degrees of deformity, and measurements of the deformities were recorded. The extent and type of the deformity was correlated with other anatomical features in the children.

The extent and degree of the medullocervical dislocation are not related to the site of the open meningo-myelocele, but are directly related to its extent. Evidence also suggests that the form of the deformity, i.e. the dorsal spur of 'knickung', and cyst are also related to the laxity of the cranial end of the dentate ligaments.

It would seem most likely that the medullocervical deformity in children with meningo-myelocele is secondary to the open spinal dysraphism.

Uhl's anomaly. W. A. Aherne. Department of Pathology, Newcastle General Hospital, Westgate Road, Newcastle upon Tyne NE4 6BE.

The patient was born normally at term, of young and unrelated parents, after an uneventful pregnancy. She was admitted to hospital at the age of 7 weeks with an upper respiratory infection from which she made a good recovery. A chest x-ray at this time showed some enlargement of the heart shadow. She was admitted again at the age of 10 months for investigation of increasing general oedema, restlessness, and irritability. On examination the apex beat was not palpable; the heart rate was 125/minute; the blood pressure 110/70 mmHg. There was considerable enlargement of the liver, which was firm and acutely tender on palpation. X-ray of the chest showed gross enlargement of the heart, especially on the right side, and oligemic lung fields. There was incomplete right bundle-branch block. Cardiac catheterization showed (among other features) only a 3 mm pressure difference between the right atrial A-wave and the right ventricular systolic peak. Cineangiography showed a huge right atrium and ventricle with very poor contractions of the right

ventricle. She deteriorated and died in congestive heart failure.

At necropsy there was evidence generally of congestive heart failure, and, apart from this, the significant findings were in the heart and liver. The heart was greatly enlarged, due mainly to dilatation of the right ventricle and dilatation with hypertrophy of the right atrium. The free wall of the right ventricle appeared pale and fibrous except for a narrow zone, anteriorly and posteriorly, where it joined the interventricular septum. The cavity of the right ventricle was enormously enlarged, and though the anatomical configuration was normal, the papillary muscles in particular were extremely thin.

Sections taken from various parts of the right ventricle showed almost complete absence of myocardium in the greater part of the free wall; there were occasional small bundles of muscle fibres. There was a well-established endocardial fibroelastosis which was in direct contact with the epicardial and subepicardial tissues. At the junction of the right ventricular free wall and the septum there was a quantity of very small and only occasionally striated muscle fibres, which strongly resembled embryonic myocardium. There was no evidence of past myocarditis or of ischaemic damage; the appearances in general suggested rather a failure of myocardial development. Apart from the hypertrophy of the right atrial muscle, the rest of the heart appeared macroscopically and microscopically normal. It was concluded that propulsion of blood through the right heart was due almost entirely to atrial contractions.

Sections of the liver showed a cardiac cirrhosis, presumably due to the backward pulsations from the right atrium.

Studies on GM2 type 2 gangliosidosis. A. D. Bain. Department of Pathology, Royal Hospital for Sick Children, Sciennes Road, Edinburgh EH9 1LF.

Necropsy diagnosis of fructosaemia in the newborn. J. S. Wigglesworth. (Nuffield Neonatal Research Unit, Institute of Child Health, Hammersmith Hospital, Du Cane Road, London W.12.

Lipid histochemical study in Fabry's disease. B. Ivemark. Department of Pathology, Karolinska Sjukhuset, Stockholm 60, Sweden.

Hirschsprung's disease: experience with some enzyme histochemical techniques. J. D. Elema. Department of Pathology, University of Groningen, Oostersingel 63, Groningen, Netherlands.

Acetylcholinesterase (AChE)-positive nerve fibres are increased in number in the mucosal layer of the bowel of patients suffering from Hirschsprung's disease.

To decide whether rectal suction biopsies stained for AChE could be used in the diagnosis of this condition, 36 patients with constipation were investigated for AChE activity.

29 patients ultimately proved not to be suffering from Hirschsprung's disease; none of the rectal biopsies

showed increased AChE activity. 7 patients ultimately proved to be cases of Hirschsprung's disease; all but one of the rectal biopsies showed an increased AChE activity in the mucosa, and all biopsies showed an increased activity in the submucosa.

From the investigation of resected bowel segments it was shown that the strongest activity occurred in the most distal part of the aganglionic bowel and that the proximal extension of the enhanced activity into the aganglionic segment increased with age. Rectal suction biopsies stained for AChE activity therefore seem to be a valuable diagnostic aid for investigation of constipation. False negative results are possible in young infants, but can be avoided by taking the biopsy immediately proximal to the pectinate line.

Large-scale investigation of 2 phenylketonuria screening methods and factors affecting blood phenylalanine levels in the newborn. J. B. Holton. Department of Pathology, Southmead Hospital, Bristol BS10 5NB.

Bloods taken from 22,365 babies between the 5th and 20th day of life were analysed for phenylalanine by the Guthrie inhibition assay and by an automated fluorimetric method (Hill *et al.*, 1965). 6 phenylketonurics were found and both methods were equally effective in detecting the disease. The fluorimetric method showed a more consistent pattern of rising phenylalanine levels before treatment. Using an acceptable upper limit of mean $+2$ SDs, the fluorimetric method gave more 'false positive' results. Two-thirds of these high levels were shown by a thin layer chromatogram to be associated with tyrosinaemia. The distribution of blood phenylalanine by both methods was non-Gaussian, and a small shift in the accepted upper limit would eliminate many 'false positives' without significantly altering the detection of phenylketonuria.

An attempt has been made to identify in normal infants some factors which influence blood phenylalanine in the newborn period. The level is raised at day 5 then remains constant up to day 12. After the 6th day of life the phenylalanine level of boys tends to be higher than girls. Babies with a birthweight below 2.5 kg have significantly higher phenylalanine levels than heavier babies, presumably due to immaturity of liver enzymes. However, very low birthweight babies do not show as high levels as those between 2.0 to 2.5 kg, perhaps because of poorer feeding. A big difference was observed between babies born in rural areas and those from urban districts. The reason for the lower phenylalanine level in rural areas is unknown.

REFERENCE

- Hill, J. B., Summer, G. K., Pender, M., and Roszel, N. O. (1965). An automated procedure for blood phenylalanine. *Clinical Chemistry*, 11, 541.

Quantitative study of vesical ganglia in children with neurospinal dysraphism. M. Forbes. The Congenital Anomalies Research Unit, University of Sheffield, Thornbury Annexe, Sheffield 10. This project was undertaken to determine if neurogenic dysfunction

of the bladder in children with spina bifida and meningocele was in any way related to the population of vesical neurones present.

Two quantitative histological studies were performed. In the first, 10 whole bladders (3 normal and 7 spina bifida) were serially sectioned at 10μ . Every 50th section was counted and the total number of ganglion cells calculated per block of bladder from the fundus to the urethra. The results showed an overall reduction in the number of ganglion cells in the bladders of spina bifida children, but there was a particularly striking depletion in the region of the trigone.

In the second study, a single midtrigone block was taken from 100 bladders (50 normal and 50 spina bifida) and again the total number of ganglion cells estimated per block. The mean values for ganglion cell counts related to age showed a marked reduction in the spina bifida cases as compared with the controls.

The possible pathogenesis of these findings was discussed.

Brain swelling in the newborn: artefact, development, or pathology? J. Pryse-Davies. Bernhard Baron Memorial Research Laboratories, Queen Charlotte's Maternity Hospital, Goldhawk Road, London W.6.

Brain swelling was studied prospectively in 183 perinatal deaths of 20 to 42 weeks' gestation. Cerebral flattening in 67 babies obviously correlated with maturity, a factor difficult to exclude in any other analysis; there was also an apparent association with birth asphyxia and intrauterine growth retardation.

Macroscopically there was a progressive series of findings to suggest genuine brain compression. Cerebellar herniation of varying degree was found in 22 cases and slight herniation of uncus gyri in 30: both features occurred in 16 brains. Such herniation phenomena were only found in association with cerebral flattening, which was not always marked. Reduced cisternal CSF in 46 cases and skull moulding in 21 usually correlated with other features of cerebral compression, but also occurred without brain swelling; 18 swollen brains showed no confirmatory evidence of compression. Prolonged body storage and survival time were probably related to an increased incidence of cerebral flattening but not to herniation phenomena.

Babies showing cerebellar herniation have been described in detail, as this was considered the main indication of pathological brain swelling. This finding was associated with death before (3 cases), during (10), and after labour (9). Apart from gross trauma or a softened macerated skull, moulding alone was thought unlikely to initiate herniation. However, constriction of a swollen brain during intrapartum hypoxia might increase the lethal potential of compression and exaggerate the appearances in the dead fetus. In 9 babies cerebellar coning was probably unrelated to skull pressure and due to hypoxia or intracranial bleeding occurring after delivery.

Histological evidence of nerve cell damage showed no consistent relation to brain swelling. Purkinje cell