Paediatric Research Society

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Renal Function Studies in Girls with Asymptomatic Bacteriuria. D. C. L. Savage. The renal function in 5-year-old girls with asymptomatic bacteriuria was studied. No abnormality was found in creatinine clearance, and the results of urinary acidification were within normal limits.

The concentrating ability of these children after 19-20 hours of water deprivation was significantly different from that of controls ($p < 0.01$). In asymptomatic bacteriuria the urinary osmolality achieved by children with radiological evidence of urinary tract abnormality was significantly different from those with normal renal tracts ($p < 0.01$). This defect of concentrating ability was corrected by therapy.

Neonatal Polycythaemia. Peter M. Dunn. Neonatal polycythaemia may be arbitrarily defined as a venous packed cell volume (PCV) of 70% or more, or a capillary blood PCV of 75% or more, during the first month of life.

The known and suspected causes of neonatal polycythaemia were briefly reviewed. In particular, the ability of the placental transfusion and postnatal plasma shift to induce an intense transitory polycythaemia immediately after birth was discussed. In a study of 86 unselected normal term infants no cases of polycythaemia were detected at delivery, though the umbilical vein PCV was over 60% in 8% of cases (mean value 52%, range 37%-66%). Nor were polycythaemic values encountered during serial observations on 17 normally delivered term infants whose cords were clamped immediately (mean delay 1.1 sec.); for example, the mean PCV values and ranges for the cord blood and for the capillary blood at 5 and 24 hours were: 52% (45-62), 62% (56-68), and 55% (48-65). In contrast, half the infants in a matched group whose cords were left intact for at least 3 minutes after delivery (mean delay 4.7 minutes—infants lying on bed) became polycythaemic; the comparable mean values and ranges to those given above were 52% (43-64), 73% (63-86), and 65% (60-75) ($p < 0.001$). Further studies demonstrated a direct correlation between the PCV and the clinical estimation of blood viscosity ($p < 0.001$) and an indirect correlation between PCV and pH of capillary blood 4 to 12 hours after delivery ($p < 0.01$). The main clinical associations and complications of neonatal polycythaemia were briefly mentioned, as was the benefit to be obtained in selected cases from dilution

exchange transfusion with plasma or saline (20-30 ml./kg. body weight).

Neurological and EEG Findings in Infants with Disordered Calcium and Magnesium Metabolism. J. Keith Brown (*introduced by F. Cockburn*). Out of a group of 142 neonates with convulsions, 75 were due to abnormalities of calcium or magnesium metabolism—an incidence of 7 biochemical fits per 1000 deliveries. There was an increased incidence in the months of March to May, the commonest day of onset was the sixth, mean gestational age 39 weeks 6 days, mean weight 3345 g.

They were investigated neurologically, biochemically, and by EEG. The neurological examination consisted of the accepted three parts, assessment of gestational age, state of alertness, and examination proper. The jittery baby with exaggerated phasic reflexes was found in 39/75 cases and jitteriness was severe in 26. Carpopedal spasm was not an accompaniment and Chvostek's sign was not reliable. Varying degrees of extensor hypertonus were found in 27 cases and this was severe in 14, producing metabolic decerebration. In 14 cases there were splayed sutures and in 7 'sunsetting' of the eyes. There was no relation between decerebration and signs of raised intracranial pressure, the former appearing to be due to the metabolic lesion and not to pressure. No infant was hypotonic and none was apathetic.

The EEG was recorded using a temporal ring for 6 channels and the 7th and 8th as vertex electrodes over the motor area. The importance of this *montage* was stressed. 60% of the cases had an abnormal EEG, and in 40 it was frankly epileptic. Spike discharges were seen in all 53 ictal episodes which occurred during the EEG recording. No infant had a clinical fit with a normal EEG at that time, though an epileptic EEG could occur without a clinical fit. The site of the spike, configuration, amplitude, spread of discharge, and rate of discharge varied within the same fit. Two fits could occur at the same time clinically and electrically or a full blown hypsarrhythmic pattern develop. Focal myoclonic fits at a rate of one or three to four cycles per second were the commonest type seen; no infant had a classical *grand mal* fit.

Calcium and magnesium estimations were made by atomic absorption spectrophotometry. The lower normal for calcium was taken as 7.4 mg./100 ml. (mean—

2SD for breast fed) and for magnesium as 1.42. All cases had one or both ions below these levels. 39 of the 75 cases showed hypomagnesaemia with hypocalcaemia and 5 had hypomagnesaemia alone. 33 were treated with magnesium and calcium and 9 with magnesium alone. There were 9 infants who showed the hypotonic normo-alerted clinical picture of hypermagnesaemia as a result of therapy. CSF calcium and magnesium levels were significantly lowered ($p < 0.005$ Mg, < 0.0025 Ca).

The infants were fed on full-cream Regal milk which has a high phosphate (90 mg./100 ml.) and a high osmolality.

OFD Syndrome. J. A. Dodge. Oral-facial-digital syndrome (OFD) is one of a few inherited disorders which are virtually limited to the female sex, and at least 2 of the 7 reported male cases had XXY karyotypes. Rimoin and Edgerton (1967) drew attention to a very similar disorder (OFD II, or Mohr syndrome) which is inherited as an autosomal recessive and may, therefore, occur in males. Examples of both syndromes have been encountered in Northern Ireland. 'Classical' OFD was seen in the first and third generations of a pedigree, but the single living patient in the second generation had very minor dental and dermatoglyphic abnormalities which would have been overlooked or disregarded but for the manifestations in her daughters. She is, therefore, regarded as a true '*forme fruste*'. Her mother and four daughters all had lobulated tongues with hamartomata, supernumerary frenula, and gross dental malformations. Another female in the second generation is reported to have had typical OFD features, but she died at the age of 8 months; she had the same mother as the '*forme fruste*', but a different father. This is further evidence for genetic transmission as a dominant characteristic, through the female—a rare mode of inheritance which may be explained by postulating that the gene is X-linked, lethal when hemizygous. This hypothesis is supported by the finding of Klinefelter's syndrome in some of the affected males. In order to investigate further the suggestion of X-linkage, the Xg blood groups of the pedigree were determined. The findings indicated that recombination had occurred in 3 out of 4 patients in the third generation. If the OFD gene is carried on the X chromosome, it is likely to be situated a long way from the Xg locus.

REFERENCE

- Rimoin, D. L., and Edgerton, M. T. (1967). Genetic and clinical heterogeneity in the oral-facial-digital syndromes. *Journal of Pediatrics*, 71, 94.

Vertebral Abnormalities in First Degree Relatives of Cases of Spina Bifida and of Anencephaly. K. M. Laurence. The parents and sibs aged over 5 years of 100 cases of anencephaly, spina bifida cystica, and encephalocele were x-rayed to determine spinal abnormalities. All these individuals were matched for age and sex with cases seen in the accident unit. Verte-

bral abnormalities found were divided into the mild and severer groups.

Sibs showed 54% mild and 26% severer forms of spina bifida occulta, as compared with 36% and 8% respectively, amongst the controls under 20 years of age. Parents had 20% mild and 4% severer abnormalities, and adult controls had 20% and 1%, respectively.

The results suggest that the severer variety of spina bifida occulta only is probably part of the dysraphic neural tube syndrome.

Hand and Foot Preference in Thalidomide Children. R. W. Smithells. Of 197 thalidomide children, 34% were left-handed and 37% were left-footed. Of those with no arms but normal legs, 70% were left-footed. There was no increase in left-side preference amongst their parents or sibs. The reason for this is obscure, but the observations support the view that a wholly genetic explanation for hand and foot preference is untenable.

Measurement of Adipose Cells in Children. C. G. D. Brook (introduced by June Lloyd). Early nutritional experiences may have lasting consequences on cell growth and behaviour. Animal experiments have demonstrated that different weaning diets have permanent effects on adipose cells in the rat. This paper presents preliminary observations on the measurement of adipose cells in children.

Subcutaneous adipose tissue was obtained at the time of operation from 34 children, aged 3 months to 42 years and of normal weight, who were undergoing elective surgery for various conditions. In 12 of these children additional samples were obtained from deep (intra-abdominal) sites. The lipid content and fatty acid composition were estimated by quantitative gas-liquid chromatography in all samples. In 21 of the children the number of cells was counted by method III of Hirsch and Gallian (1968), and total adipose tissue mass estimated from skin-fold thicknesses using the regression equations of Durnin and Rahaman (1967).

There was a small but statistically significant difference between the mean lipid content of adipose tissue (% wet weight) taken from subcutaneous and deep sites, though the fatty acid composition did not differ. In those children in whom biopsies were taken from both subcutaneous and deep sites the cell size of the deep adipose tissue was considerably less than that of the subcutaneous tissue. However, because deep fat forms only a minor part of the total adipose tissue mass the difference in cell size was ignored in estimating the total number of adipose cells in the body. This has been calculated from the mean cell size of subcutaneous adipose tissue; other workers have shown that subcutaneous cells from different parts of the body are of similar size (Hirsch, Knittle, and Salans, 1966), and this has been confirmed. Results so far obtained indicate a linear relation between total cell number and chronological age in children of normal weight. Preliminary observations in a few obese children from whom