All cases had one or both ions hypermagnesaemia and hypocalcaemia. The osmolality. 39 of the 75 cases syndrome (OFD) is one of a few inherited disorders 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


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. Vertebral 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 4 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 cell number and chronological age in children of normal weight. Preliminary observations in a few obese children from whom...