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Gonadal dysgenesis and Y-chromosome abnormalities.

Fifteen subjects with chromosome mosaics were investigated: in ten of them a chromosome pattern 45,X/46,X,i dic(Yp) was found, in four 45,X/46,X,i dic(Yq), in one 45,X/46,X,Yq+.; the ratio of the two cell types varied between patients and between different tissues. The phenotype of patients ranged from female with some Turner's syndrome stigmata, to infants with ambiguous external genitalia, to azospermic male. Most patients presented short or very short stature. Laparatomy and hystologic studies revealed hypoplasic or dysgenetic or even normal testes and streak gonads variously combined; four patients were tested for H-Y antigen and found to be positive-reduced. No correlation was demonstrated between clinical data and chromosomal pattern. Phenotypic feature of mosaics with structurally abnormal Y-chromosomes are not different from feature found in 45,X/46,XY patients; probably no genetically active portion of Y is lost. The different proportion of 45,X cells in various tissues possibly has the greatest influence on the phenotype of these subjects.

This investigation was performed in collaboration with the Institute of General Biology and Genetics, University of Pavia.

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PROLACTIN AND TSH RESPONSES TO TRH IN 146 HYPOPITUITARY DWARFS.

Prolactin (PRL) and TSH were measured from 0 to 90-120 minutes after TRH 0.2 mg IV in 146 hypopituitary dwarfs : 76 with cranial radiotherapy and/or hypothalamic lesions (groupe A) and 70 without grossly detectable lesion (group B) including 14 after breech presentation and 19 isolated GH deficiency. Abnormal PRL responses, either delayed or outside the normal range, were found in 71 patients, 45 from group A (59 %) and 26 from group B (37 %). Abnormal TSH responses, delayed or outside the normal range, or in the normal range with low plasma T4, were found in 90 patients : 50 from group A (66 %) of which 26 had low T4 and 40 from group B (57 %) of which 19 had low T4. One of the responses at least was abnormal in 113 (77 %): 63 from group A (83 %) and 50 from group B (71 %), only 49 patients showing abnormal responses of both hormones. In group B, 19 patients with otherwise isolated GH deficiency and normal T4 had an abnormal response of one or two hormones. It may be concluded that 1/considering both TSH and PRL responses, TRH test is abnormal in most hypopituitary dwarfs including otherwise isolated GH deficient subjects. 2/ While the percentage of normal TSH response seems identical in the different etiological types an abnormal PRL response is more frequent in lesional hypopituitarism.

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Pituitary Gigantism : Apparent Response to Bromocriptine.

A.M., a male aged 7.7 years presented with rapid growth since the age of 5 years. His height was 151.1 cm, and weight 36.7 kg. There was no secondary sexual development. Investigation showed enlargement of the pituitary fossa, a slightly advanced bone age, elevated levels of plasma GH (30-70 mU/l) with no suppression after glucose. Plasma prolactin was also elevated (2000 mU/l) but pituitary function otherwise normal. Treatment with Bromocriptine (up to 20 mg/day) resulted in rapid suppression of prolactin but less rapid suppression on GH to near-normal. Treatment was associated with a fall in growth rate from 9.0 cm/year to 5.6 cm/year.

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Effect of LH-RH treatment for cryptorchidism on gonadotropine secretion.

Sixty-two cryptorchid boys aged two to six years were selected at random either for surgical or for hormonal LH-RH treatment. As all biopsies from boys operated upon showed typical histological and ultrastructural signs of cryptorchidism, it can be concluded that only true cryptorchid patients were included in our study. LH-RH treatment was successful in sixteen (55%) of thirty-one boys. Median plasma 30' response values to LH-RH of LH were initially normal in all boys. Those treated successfully, remained also normal after treatment, whereas unsuccessfully treated patients had significantly lower LH response values at the end of the 4 weeks LH-RH treatment. FSH response was not statistically different before and after treatment.

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Kallman's syndrom with high set osmotic threshold (OT) and deficient thirst.

A patient with Kallman's syndrom is presented, who had deficient thirst mechanism and high set OT for vasopressin secretion. Random determinations of plasma osmolality (Posm) revealed high values of 296-307 mOsm/kg. The OT defined by the isovolemic infusion of hypertonic saline, was 296mOsm/kg. This was significantly higher than the OT of 7 normal controls, 286.6±0.9 (M±SD). He had intact volume receptors, as shown by an appropriate fall of free water clearance during dehydration, from -0.3 to -1.25/min. His pressor receptors were shown to be intact by a normal response to the hypotensive agent trimethaphane with a fall of free water clearance from 10 to 1 ml/min. and a rise of plasma ADH from 1 to 26uU/ml. Indifference to thirst was expressed at plasma osmolality as high as 301mOsm/kg. It is suggested that deficient thirst and high set OT are additional manifestations of the hypothalamic involvement of Kallman's syndrom.

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The Childhood Prognosis for Congenital Hypothyroidism.

The neurological, intellectual and behavioural problems of 126 hypothyroid children were examined in a cross-sectional study designed to provide retrospective control data for a newborn screening programme.The mean I.Q.(WISC-R) declined progressively with diagnostic delay during the first year, but improved thereafter. 9 children with severe pre-natal hypothyroidism treated before 3 mths. had a mean I.Q. of 93. 35% of the children had I.Q.s over 90 but 23% were below 70.

Although 67% of the children were not walking by 18mths., walking delay was only a bad prognostic feature for I.Q. after 2 yrs. though even some very late walkers did well. Speech delay was a better indicator of poor intellectual prognosis. Of 100 school-age children, 27% required education in special schools and a further 15% remedial teaching.Behaviour disorders (assessed by teachers' ratings) were present in 45% compared with 12% of age and sex matched controls. These were found in many children with post-natal onset of the hypothyroidism.

The study confirms that new-born screening is likely to lead to a substantial improvment in the I.Q. prognosis though children may continue to have minor behavioural or motor problems.