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STUDY OF 3ß-HYDROXYSTEROID DEHYDROGENASE ACTIVITY (3ß-HSD) IN PRIMARY CULTURE OF HUMAN TESTICULAR CELLS DURING THE FIRST MONTHS OF LIFE.
Berensztein, E., Saraco, N., Belgorosky, A., Rivarola, M.A. Laboratory of Investigation, Garrahan Children's Hospital, Buenos Aires, Argentina.
The activity of 3ß-HSD, estimated by the ratio of testosterone (T) + androstenedione (A) /dehydroepiandrosterone (DHEA) was studied in primary cultures of human prepubertal testicular cells (post mortem) in basal conditions and after stimulation with 40 U/L hLH, 1.5 U/L rhFSH or 0.12 U/L rhGH. T, A and DHEA were measured by RIA on day 6 of culture in 6 independent testes collected from 6- and 9-day-old newborns (n) and from 1-, 3-, 7- and 24-month-old infants (i). Basal ratio was (X+SD) 10.7 ± 2.28 and 16.2 ± 3.72 for the n and 0.60 ± 0.42, 2.63 ± 0.24, 2.30 ± 0.23 and 0.29 ± 0.23 for the i. Response to stimuli was evaluated by Student's t test as % increment over basal. It was found that testicular cells from n did not increase 3ß-HSD under any condition while those of i increased significantly under hLH (256 ± 5%, X ± SD), rhFSH (425 ± 194%) and rhGH (365 ± 125%). In conclusion, n testicular cells probably express maximal 3ß-HSD activity that cannot be stimulated further. This activity is lost after the n period when it becomes hormone responsive. The effect of FSH is probably mediated by a paracrine factor secreted by Sertoli cells. GH or prolactin can have an effect on the testes.

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SRY GENE SCREENING IN TURNER SYNDROME BY POLYMERASE CHAIN REACTION (PCR). Domenice, S., Nishi, M., Medeiros, M.A., Costa, E.M.F., Nicolau, W., Arnhold, I.J.P., Mendonca, B. B. Gonads and Intersex Unit, Division of Endocrinology HCFMUSP, Sao Paulo, Brasil.
The association between gonadal tumor and Y chromosome material in patients with dysgenetic gonads is well defined. Cytogenetic analysis sometimes is unable to detect low mosaicisms or to identify the origin of ring/maker chromosome fragments. Recently molecular techniques have determined Y chromosomal material in Turner syndrome. We studied 27 patients with Turner syndrome by PCR technique using XES10 e XSE11 primers that amplify the complete sex determining region (780bp) of the Y-chromosome gene (SRY) with the use of the following program: 32 cycles (94°C, 1 min, 55°C, 1 min and 72°C, 2 min). Only one patient had mild virilization (clitoromegaly). Cytogenetic analysis revealed: 16 cases 45,X, 6 cases 45,X + mar, 4 cases 45,X/46,XX, 1 case 45,X/46,X dic (Y) (qter -> p11::p11 -> qter) by C and G banding techniques. PCR amplification of the SRY gene was positive in 3 patients (11% of the cases). Two of these were 45,X, + mar (including the patient with clitoromegaly), and the third patient was 45,X/46,X dic (Y). Conclusion: Molecular techniques are faster and more sensitive than cytogenetic studies to detect fragments of Y chromosome. These findings further underline the relevance of performing molecular studies in patients with Turner Syndrome, which may help to define appropriate therapeutic strategies.

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FAMILIAL NEUROHYPOPHYSEAL DIABETES INSIPIDUS (NHDI) DUE TO A MUTATION IN THE VASOPRESSIN-NEUROPHYSINE II (VP-NPH II) GENE. Arriazu, M.C., Roubicceck, M., Isaac, G., Blanco, M. Community Hospital, Mar del Plata, Argentina.
Autosomal dominant NHDI is a familial form of antidiuretic hormone deficiency, usually expressed in early childhood with polyuria, polydipsia, and an antidiuretic response to exogenous vasopressin or its analog. We report a family with evidence of NHDI in four generations. Diagnosis was confirmed with plasma and urine osmolarity under water restriction, and a good response to DDAVP (VP analog) in the patients receiving treatment. The family, of Russian-German ancestry, lives in the southwest of Buenos Aires province, Argentina. Most affected members did not seek medical attention, as they viewed the symptoms as a natural occurrence in their family, in spite of diuresis over 12 liters a day, and single voiding volumes over 1.5 liters. Phenotypic expression was variable. Molecular studies were performed trying to characterize a mutation of the AVP-NP II gene, encoding AVP, its carrier protein NP II, and a glycoprotein of unknown function. A Guanine to Adenine mutation was detected in the nucleotide 1859, that encodes a change from Glycine to Serine at Aminoacid 57 of NP II. Five different mutations have been reported up to this time. The mutation in this family has also been described in a Japanese kindred.

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ETIOLOGIC DIAGNOSIS IN 126 CASES OF SEX AMBIGUITY.
Maciel-Guerra, A.T., Guerra Jr. G., Marques-de-Faria, A.P., Baptista, M.T.M., Silva, R.B.P. Interdisciplinary Group for the Study of Sexual Development (GIEDDS) - FCM - UNICAMP, Campinas, Sao Paulo, Brazil.
The complexity of human sex differentiation becomes manifest in the variety of diseases associated with sex ambiguity (SA). There are a few clinical centers especially dedicated to the managament of such patients, and hence, few large series have been published. We had the oportunity to evaluate 126 cases of SA. Among them, 57 (45.2%) were male pseudohermaphrodites (MPH), 35 (27.8%) were female pseudohermaphrodites (FPH), 23 (18.3%) had anomalies of gonadal differentiation (AGD), and 11 (8.7%) exhibited other genital abnormalities. Among MPH, 36 were considered idiopathic, due to our impossibility to study androgen/receptor binding in cultured fibroblasts to diagnose partial androgen resistance, 11 were syndromic, there were 5 anorchias, 3 had complete androgen resistance, 1 had mullerian persistence and 1 5-alpha-reductase deficiency. Congenital adrenal hyperplasia was the main cause of FPH (24); there were also 6 syndromic cases and 5 idiopathic cases. Among AGD, there were 10 cases of true hermaphroditism, 8 Klinefelter syndromes, 3 mixed gonadal dysgenesis and 2 testicular dysgenesis due to sex chromosome aberrations. There were also 4 cases of balano-prepucial hypospadias, 3 of cryptorchidism, 3 incarcerated ovaries and 1 idiopathic microrchidism. Such a large series could only be achieved as a result of an interdisciplinary approach to SA over approximately 5 years.