FREE COMMUNICATIONS. Brief Comments on each oral presentation and on awarded Posters.

Marco A. Rivarola y Alicia Belgorosky
Servicio de Endocrinologia, Hospital de Pediatría Garrahan, Buenos Aires, Argentina

FREE COMMUNICATIONS

During the XVIII Annual Meeting of SLEP, 133 scientific works were presented as free communications, either as Oral Presentations or Posters. Six Sessions of Oral Communications of 4-6 presentations each (n = 30), and 3 Poster Sessions of 33-38 (n = 103) presentations each, were presented. The majority of these works were carried out by members of SLEP or non-member Latin American professionals. These is a good demonstration of the vitality of Pediatric Endocrinology in Latin America. The abstracts of the Meeting were published in a Supplement of the Journal of Pediatric Endocrinology and Metabolism (volume 19, supplement 3, September 2006, Freund Publishing House Ltd, Tel Aviv, Israel).

Oral Sessión 1.
Oral Communication #1. The authors (Pepe C et al., Lab. Investigación, Hosp. Garrahan, Buenos Aires) proposed that a mRNA variant of aromatase, lacking exon 5 (-Ex5), and associated to aromatase deficiency, could be present in normal human steroidogenic tissues. They found that the –Ex5 variant, presumably generated by alternative slicing, was present in prepubertal testis, adrenal gland and placenta. They raised the question whether this alternative splicing is a mechanism of enzymatic activity modulation.

Oral Communication #2. The authors (Ropelato et al., División de Endocrinología, Hospital Gutierrez, Buenos Aires) studied the effects of testosterone on spontaneous LH secretion in eumenorrheic adolescents and in polycystic ovary syndrome. Since they found differences they conclude that an alteration of the mechanism of feedback is present in this syndrome.

Oral Communication #3. The authors (Berensztein E. et al., Lab. Investigación, Hosp. Garrahan, Buenos Aires) found immunoexpression of IGF-II and insulin receptor in Leydig cells and in germ cells of the post natal human testis. They propose that IGF-II would stimulate cell proliferation and would inhibit apoptosis in the interstitium, and furthermore they would contribute to preserve an adequate pool of spermatogonia in seminiferous cords.

Oral Communication #4. The authors (Iñiguez G et al., IDIMI, Escuela de Medicina, Universidad de Chile) found an increase in the concentration of IGF-I and IGF-II in placenta (maternal and fetal sides) collected from childbirths of small for gestational age newborns. There was an inverse correlation with birth weight. Different from babies born adequate for gestational age, placental fetal side concentrations of IGF-I and IGF-II were higher than maternal side concentrations.

Oral Communication #5. The authors (Hernández MI et al., IDIMI, Escuela de Medicina, Universidad de Chile) studied the relationship among gonadal function, insulin secretion/sensitivity and a polymorphism in the promoter region of the CYP17 (T—C, -34 bp) gene, during early puberty in healthy girls, born either small or adequate
for gestational age. They found girls with the polymorphism had lower serum testosterone and greater insulin sensitivity. Testosterone levels were higher in girls born small for gestational age.

Oral Communication #6. The authors (Grispon R. et al., División de Endocrinología, Hospital Gutierrez, Buenos Aires) studied the diagnostic value of serum gonadotrophins measures by ultrasensitive methods in boys with doubtfully palpable gonads. They concluded that high serum levels are useful to predict hypo/agonadism, but not to differentiate between anorchia and damaged testis. Lack of elevation ascertains the presence of testes but does not differentiate normo from hypo function.

Oral Session 2.

Oral Communication #7. The authors (Scaglia P et al., División de Endocrinología, Hospitales Gutiérrez, Tornú, Fernández de Buenos Aires, and MGB, NHGRI, NIH, USA) studied the diagnostic value of serum gonadotrophins measures by ultrasensitive methods in boys with doubtfully palpable gonads. They concluded that high serum levels are useful to predict hypo/agonadism, but not to differentiate between anorchia and damaged testis. Lack of elevation ascertains the presence of testes but does not differentiate normo from hypo function.

Oral Communication #8. The authors (Iñiguez G et al., División de Endocrinología, Universidad de Chile, and INSMED, Glen Allen, Virginia, USA) studied the acute effect of sc IGF-I/IGFBP-3 complex administration on serum nocturnal concentrations of GH and Ghrelin in children born small for gestational age. They found that, while the complex decreased GH concentrations (negative feedback), it increased Ghrelin (positive feedback) in children born small for gestational age.

Oral Communication #10. The authors (Jorge AAL et al., Unidade de Endocrinologia do Desenvolvimento, LIM 42, Disciplina de Endocrinologia, Hospital das Clínicas, São Paulo, Brazil) evaluated the frequency of SHOX gene mutations in idiopathic short stature. They concluded that frequency is low (3.2 %), even after extensive molecular studies. However, among patients selected because of sitting height SDS/height SDS ratio > +2 DS, the incidence increased to 22 %.
compensatory growth suggesting that fetal malnutrition might program the body for poor growth.

Oral Communication #13. The authors (Della Manna T. et al., Pediatric Endocrine and Pulmonology Units, Instituto da Crianza-HCFMUSP, São Paulo, Brazil) compared metabolic responses to mixed meal and to oral glucose tolerance tests in 52 children and adolescents with cystic fibrosis. Mixed meal test produced an earlier hormonal (insulin and glucagon) secretion. Insulin resistance, in addition to beta cell dysfunction was also detected, in the complex pathogenesis of glucose alterations, in cystic fibrosis.

Oral Communication #14. The authors (Iñiguez G et al., IDIMI, Escuela de Medicina, Universidad de Chile, Department of Paediatrics, University of Cambridge, UK) determined longitudinal changes in IGF-I levels, insulin secretion and insulin sensitivity in 50 children born small and 14 adequate for gestational age, from birth to 3 years of age. They found that IGF-I was associated to catch up growth and to insulin secretion, in small for gestational age, during the first year of life. After the first year of life, insulin resistance and decreased sensitivity to IGF-I developed. This might generate a risk of type 2 diabetes for the future.

Oral Communication #15. The authors (Eyzaguirre F. y col., Clínica Santa María, U. de Los Andes, Clínica Las Condes) studied the prevalence of having been born small for gestational age (SGA) in 1002 patients with overweight or obesity. The prevalence of SGA in children and adolescents with overweight/obesity was similar than in the general population, but the former had higher triglycerides.

Oral Communication #16. The authors ((Oliveira A.M. y col., State University of Feira de Santana, Federal University of Bahia, y Cleriston Andrade, Brazil) studied non-alcoholic fatty liver disease and its relationship with insulin resistance and the metabolic syndrome in children and adolescents. They concluded that non-alcoholic fatty liver disease might be a component of the metabolic syndrome, but that this metabolic alteration develops after many years.

Oral Session 4.

Oral Communication #17. The authors (Salazar T. et al., IDIMI, Facultad de Medicina, Universidad de Chile) studied the expression, and post GH stimulation phosphorylation and nuclear/cytoplasmic distribution of STAT-5b in fibroblasts from short prepubertal children with normal GH pharmacological tests and low serum IGF-I (possible GH insensitivity). Post GH stimulation phosphorylation and nuclear/cytoplasmic ratio of STAT-5b in GH insensitivity was lower than in controls, suggesting that this might be a mechanism for poor growth.

Oral Communication #18. The authors (Gutierrez M. et al., División de Endocrinología, Hospitales Gutiérrez, Tornú y Fernández, Buenos Aires) studied GHR (Ex3) and IGFBP-3 (-202 C/A, -185 C/T) gene polymorphisms and their relation to the formation of the ternary complex IGF-I/IGFBP-3/ALS in vitro. They found that the ternary complex changes with age and that it is decreased in association with the two polymorphisms.

Oral Communication #19. The authors (Corner E.et al., IDIMI, Facultad de Medicina, Universidad de Chile y CEDIE, Hospital Gutierrez, Buenos Aires, Argentina) measured antimullerian hormone (AMH) in adolescents and adults with polycystic ovary syndrome and type 1 diabetes. They found high levels of AMH suggesting that folliculogenesis might be abnormal in these patients.

Oral Communication #20. The authors (Marino R. et al., Servicio de Endocrinología, Hospital de Pediatria Garrahan, Buenos Aires) analyzed the GH-1 gene coding region in 16 argentinian patients with isolated GH deficiency. They found that 8 patients belonging to 6 families had deletions or point mutations in GH-1. These results suggest
that the promoter region of this gene and other genes need to be studied to find out other
gene anomalies.

**Oral Session 5.**

Oral Communication #21. The authors (Susperreguy S. et al, CIBICI-CONICET, Fac. 
Cs. Químicas, Univ. Nac. de Córdoba, Hospital de Niños de la Santísima Trinidad, 
Córdoba, Argentina) found that treatment with rhGH in patients with Turner syndrome 
decreased markers of the effect of thyroid hormones. They proposed that this effect 
could decrease growth response to treatment with rhGH in Turner syndrome.

Oral Communication #22. The authors (Chiesa A. et al., División de Endocrinología, 
Hospital de Niños Gutierrez, and Catedra de Genética y Biología Molecular, Fac. de 
Farmacia y Bioquímica, UBA, Buenos Aires, Argentina) analyzed the thyroglobulin 
gene in 11 patients with congenital hypothyroidism and goiter, as well as low serum 
thyroglobulin, detected mostly by newborn screening. They found gene mutations in 7 
patients, the most frequent mutation being R277X.

Oral Communication #23. The authors (Herzovich V. et al., Servicio de Endocrinología, 
Hosp. Garrahan, Buenos Aires) report on the finding of unexpected peripheral markers 
of the action of thyroid hormones in a patient with a new mutation of the thyroid 
hormone transporter gene MCT8. While in the brain the mutation blocks the entrance of 
thyroid hormones resulting in severe brain damage, excessive T3 would enter into 
muscle, liver and bone.

Oral Communication #24. The authors (Almeida M.Q. et al., Unidade de 
Endocrinologia do Desenvolvimento, LIM 42, Disciplina de Endocrinologia, Hospital 
das Clínicas, São Paulo, Brazil) found overexpression of IGF-II transcripts in adrenal 
adenoa and carcinoma, while type 1 IGF receptor in carcinoma was greater than in 
adenoa, resulting in a new pronostic marker, and a new potential therapeutic target.

Oral Communication #25. The authors (Baquedano M.S et al., Lab. Investigación, 
Hosp. Garrahan, Buenos Aires) studied a possible role of estrogens in adrenarche, by 
detecting the expression of estrogen receptors and aromatase in adrenal 
tissues. They found evidences that estrogens might be produced in the adrenal medulla 
to regulate zona reticularis differentiation or function via estrogen receptor . 
Furthermore, the aromatase gene probably has a specific promoter in this tissue.

Oral Communication #26. The authors (Jorge A.A.L. et al., Unidade de Endocrinologia 
do Desenvolvimento, LIM 42, Disciplina de Endocrinologia, Hospital das Clínicas, São 
Paulo, Brazil) studied ERK and AKT phosphorylation under IGF-I stimulation in 
fibroblasts of patients with Noonan syndrome and activating mutations of the PTPN11 
gene (coding for a SHP-2 tyrosine phosphatase protein). However, they did not found 
differences between Noonan syndrome and control fibroblasts.

**Oral Session 6.**

Oral Communication #27. The authors (Silveira L.G. et al., Unidade de Endocrinologia 
do Desenvolvimento, LIM 42, Disciplina de Endocrinologia, Hospital das Clínicas, São 
Paulo, Brazil) described a heterocigote KISS1 gene mutation in a 1 year old boy with 
idiopathic precocious puberty. The mutation was present in his mother and maternal 
grandmother. This mutation (P74S) would induce a longer half life of kisspectin 
increasing its bioactivity.

Oral Communication #28. The authors (Marino R. et al., Servicio de Endocrinología, 
Hospital de Pediatria Garrahan, Buenos Aires) studied the expression of the SRY gene 
blood leukocytes and in gonadal tissue in 5 patients with true hermaphroditism. They 
conclude that true hermaphroditism is an heterogenous condition and that gonadal 
mosaicism is not frequent.
Oral Communication #29. The authors (Hernandez MI et al., IDIMI, Facultad de Medicina, Universidad de Chile) evaluated serum levels of antimullerian hormone (AMH) in adolescent girls (Tanner II) born either small (n = 30) or adequate (n = 35) for gestational age. They found that in small for gestational age girls AMH was slightly higher, and that in them, there was a positive correlation between AMH and ovarian diameter or number of follicles.

Oral Communication #30. The authors (Hitchsfeld C et al., West Division, School of Medicine, U. Chile, e IDIMI, Facultad de Medicina, Universidad de Chile) studied gonadal function and AMH concentration during the first 3 months of age in children born small for gestational age. Girls, and not boys, had increased serum AMH and estradiol after GnRH stimulation comparing to controls, suggesting that ovaries are more vulnerable to intrauterine growth restriction.

Posters

Oral Communication #68. The authors (Menezes Filho HC y col., Unit of Pediatric Endocrinology Instituto da Criança/HCFMUSP*, Unit of Endocrinology of Development, Laboratory of Hormones and Molecular Genetics, LIM42/HCFMUSP**; Unit of Bone Mineral Diseases-HCFMUSP) report on a case of Barakat syndrome (hypoparathyroidism, sensorineural deafness, renal dysplasia). This 19-year-old boy was evaluated at the age of 12 years due to seizures associated to episodes of tetany and muscle weakness. A heterozygote mutation in exon 3 (709) of the GATA3 gene was detected. This results in a premature stop at codon 302 with a loss of the zinc fingers domains.

Oral Communication #69. The authors (Da Silva AG y col., Department of Endocrinology, Heart Institute (InCor), Medical School, University of São Paulo, São Paulo, Brazil and School of Physical Education and Sport, University of São Paulo, São Paulo, Brazil) studied the association between Glu27 beta2-adrenoceptor polymorphism and augmented muscle vasodilatory responses. They found that the augmented muscle vasodilatory responses to mental stress and exercise in humans who are homozygous for Glu27 of the beta2-adrenoceptor are already expressed in chidhood.

Oral Communication #109. The authors (Berensztein E. y col., Research Laboratory, Endocrinology Department, Garrahan Pediatric Hospital, Buenos Aires, Argentina) found high expression of aromatase and estrogen receptors in 3 patients with Leydig cell tumors. They propose that estrogens could participate in tumoral growth and, may be, in normal Leydig cell growth.

Oral Communication #110. The authors ((Trarbach E.B. y col., Unidade de Endocrinologia do Desenvolvimento, Laboratório de Hormônios e Genética Molecular - LIM42, Hospital das Clinicas, São Paulo, SP, Brasil.) carried out an mutational analysis of gonadotropin-releasing hormone gene 1 in 30 patients with normosmic isolated hypogonadotropic hypogonadism without GnRH-R, GPR54 or FGFR1 mutations. Most cases of normosmic isolated hypogonadotropic hypogonadism do not carry mutations in this gene.

Oral Communication #122. The authors (Balbi V y col., Hospital de Niños “SSM Ludovica” La Plata, Fundación Bioquímica Argentina (FBA), Hospital “J.P. Garrahan”, Argentina) report the results of 7 years of screening for congenital adrenal hyperplasia in 219924 neonates born in Buenos Aires Province (Argentina). They found an 1:8145 incidence of the classical form. Screening improved age of diagnosis, salt wasting and correct sex assignment.