

RESUMEN DE TRABAJOS PRESENTADOS A CONGRESOS INTERNACIONALES

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SEXAGESIMAL SCALE FOR MAPPING HUMAN GENOME.

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Six years ago I designed a diagram of the Human Genome based in a circular ideogram of the haploid set of chromosomes, using a low resolution scale of Mb Units (J. Med. Genet, 1990 ; 27 388-89). Now. I am designing a new scale that may cover the whole physical structure of genome from low to high resolutions levels. The entire length of the haploid genome of males is deployed in a circumference, marked with a sexagesimal scale of 360 degrees and 1.296.000 arcseconds. The radius display a semilogarithmic metric scale from 1 meter to the angstrom level(10¹⁰). The base pair level is measured by the « milliarsec » unit (Mas) equivalent to a thousand of arcsecond. The unit "MAS" covers 1.27mm and 0.432 bp and is the framework of DNA sequence. Thus, the 3 billion base pairs may be identified by 1.296.000.000 mas units in a continuous correlation from number 1 to number 1.296.000.000. This universal and absolute scale covers all the levels of genetic material, and can position every gene and every codon. I show an example with the allele DQA1-101 of locus DQ of Class II, of cluster MHC of chromosome 6, located between degrees 119 and 139. This allele may be located at position 441200001 mas .

We do not know the exact number of base pairs and

length for human genome. Mb and kb scales measured only partial fractions of the genome. On the contrary, the arsec scale with a given fixed length of 1.296.000.000 milliarsecs unit (mas) can measure the whole structure of genetic material.

PATERNITY ANALYSIS BY DNA FINGER- PRINTING: PRELIMINARY EXPERIENCE IN CHILE.

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Direct DNA-based testing is useful for paternity determination. The simultaneous detection of several highly informative VNIR loci (DNA fingerprinting (is used successfully in paternity analysis Jeffreys A, Nature 316:76,1985). This communication shows a preliminary sample of paternity cases solved by multilocus DNA fingerprinting in Santiago.

A group of 141 individuals, who asked for paternity analysis, at the University of Chile Clinical Hospital, between 1992 and 1995 were studied. The DNA characterization was obtained after digestion with Hae III, separation of fragments by electrophoresis in 0.7 % agarose gel and hybridization with (CAC) 5 probe marked with 32 P. Bands of size greater than the lambda/Hind III fragment of 4.3 kb, were considered. We found 13.1 % of bands shared between unrelated people and a exclusion probability of 99.99%. Only one mutant band was found (mutation rate 0.00136). Paternity exclusion was made when two or more child's bands were absent in their parents. This occurred in 10 of the 49 cases analyzed. Attribution of paternity was made by means the index proposed by Pena and Chakraborty with probability higher than 99.9 % in almost all the cases .

TRACE ELEMENTS STUDY IN PREGNANT WOMEN LIVING IN THE COPPER MINE TOWN CHUQUICAMATA: RELATIONSHIP WITH REPRODUCTIVE FAILURE AND PREVALENCE OF CONGENITAL MALFORMATIONS.

Dra. S. Castillo, C. Astete, R. Alfaro, C. Ortiz, J. Araya, J. Castillo, M. Mendoza, I. López, E. Condemarin, N. Pereira, N. Ramírez, M. Cortés, J. San Martín, P. Sanz, L. Monasterio.

Servicio de Genética- Hospital Clínico U. de Chile, Santiago, Chile (Proyecto Fondecyt 1940555)

For the evaluation of the teratogenic impact of contaminants on the inhabitants of the copper mine town Chuquicamata, we are looking for a correlation between urine levels of the following trace elements: As, Cu, Te, Bi, Sb, Se, Pb and Zn in pregnant women and a pathological outcome of the pregnancy: spontaneous abortion and/or malformations in stillbirths or newborns. The copper mine company, Codelco, has undergone a number of measures for diminishing the environmental contamination, it is possible that we are in between of these effects. In 461 urine samples we have found no traces of Te, Bi, Sb and Se (in parts per billion (The range for the others is: As 0.015-0.648ug/mg creatinin, Cu 0.031-0.823ug/mg, Pb 0.15- 0.95ug/mg and Zn 0.063-4.677 ug/mg. Chromosome studies since August 1994 reveal 36.4% aberrations in trophoblast tissue from abortions and one trisomy 21 and one sex reversed 46,XY female with campomelic dysplasia in 173 newborns. The reproductive failure rate was 14.8% in 676 births in 1994 and 17.1% in 509 births in 1995, if we add ectopic pregnancies it grows to 17.6% and 18.5% respectively. Another remarkable finding is ten twin births in 509. The malformations rate is 3.46% in 173 newborns: one oto-palato-digital II syndrome, one cavernous hemangioma, one microcephaly one congenital heart malformation, one soft palate fissure and one cryptorchidism. We believe that our studies will be useful as preliminary information for evaluating progressive effects of decontamination measures.

CHROMOSOME REARRANGEMENTS AND FLOW CYTOMETRY IN GALL BLADDER

ADENOCARCINOMA.

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Gall bladder cancer is one of the major malignant pathologies in Chile. Our country has the highest mortality rates caused by this type of cancer. It is the first cause of death due to malignant tumors among Chilean women. Up to now, the few cytogenetic studies performed have not reported specific or primary chromosome anomalies in these tumors. Also no direct connection has been reported with a given oncogene that might be responsible for the development of these cancers. There are, however, some studies on the DNA content by flow cytometry where hyperploidy is present. We analyze chromosomes and DNA content in 22 samples of gall bladder adenocarcinoma. Results were obtained in 68% of the cases (15). In 3 out of the 15 cases we found only one type of cell population with the normal karyotype due to the proliferation of supporting tissue cells in the culture. The other 12 showed two different populations, one with normal karyotype, and the other with hyperploidy (neartriploidy, neartetraploidy). This was confirmed in the cytometry study which also showed two cell populations one carrying normal DNA content and the other with hyperploidy. This shows that hyperploidy detected with both techniques represents an alteration that occurs in advanced stages. No chromosome anomalies nor DNA content alterations were found in 6 controls. In 11 out of the 15 cases, multiple structural chromosome anomalies were also found such as translocations, deletions, chromosome fragments, inversions, isochromosome, rings and markers. Some chromosome alterations appeared in two or more cases: del (4)(q32), del (17)(p12) and t (6;13) (p23;q14) which may represent primary or specific alterations in this tumor, and should be confirmed with further studies. There are some genes and oncogenes related with some of the above mentioned chromosome break points that could be explored with molecular

techniques. Supported by Fondecyt proyect 1940567-1994.

BARDET-BIELD SYNDROME (BBS) AND MONOSOMY 21pter →q21: CASE REPORT.

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The BBS, an autosomal recessive disease, is characterized by mental retardation, retinitis pigmentosa, polydactyly, obesity and hypogenitalism. Three locus BBS have been defined: type 2 locus has been related to chromosome 16q21; type 1 has been related to 11q13 and type 3 to 3p13, and in other families to none of the above. The purpose of this paper is to present an 11 year old male with BBS and partial 21 monosomy. The patient was born after a term pregnancy, with a weight of 3200 g and a length of 51 cm. Post-axial polydactyly of the left hand and both feet was noticed at birth. Psychomotor development is retarded; growth is normal and he is obese since his first infancy. Hypothyroidism was diagnosed at age 7 and at 10 he was diagnosed with retinitis pigmentosa and myopic astigmatism. Clinical findings include: obesity, weight 85 kg; length 145 cm, cranial circumference 56 cm, narrow forehead; horizontal palpebral fissures; small ears, cleft uvula; high-arched palate, small penis and testes; excision scars at V finger on the left hand and V toes on the both feet; distal narrowing fingers, brachydactyly of hands and syndactyly of II and III toes of both feet. Carpo x-ray studies showed a bone age concordant to the chronological, short metacarpal bones and irregular phalangeal metaphysis. The karyotype was 46,XY, del (21pter →q21).

Parents are normal, young and non-consanguineous. He is the second liveborn child to the couple; an older sister is normal. The mother has had two spontaneous abortions.

The karyotypes of the parents will discard an inherited unbalanced translocation and in that case a

best delineating of the patient's chromosomal aberration and the relation with his phenotype will be possible.

AUTOSOMAL DOMINANT INHERITANCE IN A FAMILY WITH OROCRANIODIGITAL SYNDROME.

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The craniodigital syndrome was described by Juberg and Hayward in 1969 as an autosomal recessive syndrome with variability in expression. Verloes (1992) suggest that the extreme variability in expression and the advance paternal age in some reports could be pointers to autosomal dominant inheritance with reduced penetrance. This syndrome presents cleft lip/palate, hypo/aplasti thumbs and mild microcephaly as the main clinical features.

The purpose of this communication is show a chilean family with three affected generations with craniodigital syndrome. In this family the grandfather, his four sons and four of his seven grandsons are affected with variable intensity since hypoplastic thumbs as only manifestation to severe mental retardation, cleft lip/palate, microcephaly and aplastic thumbs. In this family the syndrome have an autosomal dominant inheritance with variable expressivity.

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SINGLE AGENT SULPERAZONE VS. TWO AGENT CEFTAZIDIME-AMIKACIN IN HIGH RISK FEBRILE NEUTROPENIC PATIENTS

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Forty febrile episodes in 25 patients with hematological malignancies (35 with acute leukemia, 5 with others (with high risk neutropenia, median minor than 100u/l, and a mean duration of 16 days are

presented. Of those, 19 episodes received, empirically and randomized, 1g Ceftazidime 8-hourly in combination with 15 mg/Kg/day Amikacin 12-hourly (CA), and 21 episodes Cefoperazone/Sulbactam (Sulperazone (S), 3g 12 hourly, these latter were weekly administered prophylactic K-vitamin. 11/19 cases of the CA group (57.9 %) and 11/21 cases of the S group (52.3%) responded to treatment with an odds ratio= 1.25, (95% IC=0.3-5.3) ,p=0.72. In 18 unresponsive patients 1 g Vancomycin 12-hourly was added; 1 CA patient and 2 S patients responded; another S patient responded after Amphotericin B administration. In the remaining 14 cases, the antibiotic therapy was completely changed. Gram-negative bacilli were isolated in 11 cases and Gram-positive cocci in 7. In 3 CA-cases and 9 S-cases blood cultures were positive. One patient of each group died. Amikacin was ceased in 3 cases due to multifactorial renal failure; rash was present in 1 CA-case and 1S-case. Sulperazone is an alternative choice of single antibiotic treatment with no adverse effects for febrile neutropenic high risk patients.

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ANNUAL MEETING 1996 NEW RESEARCH PROGRAM & ABSTRACTS AMERICA'S MENTAL HEALTH, AMERICAN PSYCHIATRIC ASSOCIATION, ANNUAL MEETING MAY 4-9,1996 NEW YORK.

COMPLEX SEGREGATION ANALYSIS OF SCHIZOPHRENIA

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Objective: Genetic epidemiologic studies have provided evidence that genetic factors contribute to familiar aggregation of schizophrenia. However, the precise mode of inheritance has not been elucidated. Most such studies correspond to reports of Caucasian populations. The present study was performed in Santiago, Chile, whose population stems from the admixture of Amerindians and Spaniards.

METHOD: The sample consisted of 44 randomly ascertained schizophrenic probands (22 males and 22 females) with ages ranging between 20 and 48. Extended pedigree information was thus obtained. The diagnosis was made according to DSM-III-R criteria. Both probands and relative were interviewed using a structured interview (CIDI) and the DSM-III-Checklist. Complex segregations analysis was carried out using the computer program POINTER. **Results:** The nontransmission model ($Q=h=0$) was rejected as was the recessive single locus ($h=0, Z=1$). The multifactorial, the single codominant, the nonmajor locus component, the nonpolygenic component transmission model and the nontransmission of a major effect could not be rejected.

CONCLUSIONS : The most likely model that fits the data of the present study is that of a mixed model with a substantial environment component (93.12%). The frequency of the major gene was estimated at 0.000155. Our results are similar to those previously reported in ethnically different populations.

WHAT DETERMINES PRIMARY CARE PHYSICIANS DETECTION OF PSYCHIATRIC MORBIDITY?

Ricardo Araya, M.D., Psychiatry, University of Chile, Avda. La Paz 1003, Santiago, Chile; Graciela Rojas, M.D., Julia Acuña, M.D.

SUMMARY :

Objetives: To examine primary care physicians (PCPS) detection rates of psychiatric morbidity and patients and doctor's variables which influence these rates.

METHOD: Psychiatric morbidity was assessed using General Health Questionnaire (GHQ) and Clinical Interview Schedule-Revised (CIS-R). PCPS assessed patients' mental health blindly with rating scales. Patients and doctors variables influencing the detection of psychiatric morbidity (DPM) were examined using univariate and multivariate methods. **Results:** 4,079 randomly chosen patients (95% response) and 67 PCPs (93% response) from 23 PHC clinics participated, 53% of patients presented a psychiatric disturbance and approximately half were detected by PCPs. according to PCPs, 42% of their

patients showed a psychiatric disturbance. Variables related to patients, particularly reason for consultation, showed the closest association with the DPM. Undetected «cases» consulted more frequently and took more tranquilizers than detected "cases".
Conclusions: This is the largest study ever done on this subject in South America. Our results confirm previous findings. Chile shows the highest prevalence rate of psychiatric morbidity in PHC in the world. Almost one of every two patients suffering from a psychiatric disturbance goes undetected by the PCP. Detection rates are more influenced by variables related to the patient than those related to doctors.

WORLD'HIGHEST PSYCHIATRIC MORBIDITY PREVALENCE RATES IN PRIMARY HEALTH CARE.

Ricardo Araya, M.D. Psychiatry, University of Chile, Avda. La Paz 1003, Santiago, Chile; Graciela Rojas, M.D. Julia Acuña, M. D.

Summary:

Objectives: To estimate the prevalence rate of psychiatric morbidity among Primary health care (PHC) patients and its association with socio-demographic factors and frequency of consultations.
Method; A cross-sectional survey of psychiatric morbidity was made of randomly chosen attenders to PHC clinics. A structured interview which contained the General Health Questionnaire (GHQ-12), Clinical Interview Schedule-Revised, and other question was used. Reported frequency of consultations over last six months was confirmed with medical records. Variables were examined using univariate and multivariate (logistic regression) methods.

Results: 4,079 randomly chosen patients (95% response) from 23 PHC clinics participated in this study. The prevalence rate of psychiatric morbidity was 53%. Five percent of the sample gave a psychological reason for consultation. Anxiety was the most prevalent symptom. All individual symptoms were more prevalent among women. Previously married women with poor income and education were at higher risk. Attenders with psychiatric morbidity consulted more often even

after controlling for physical illness.

Conclusions: This is the largest study ever done on this subject in South America. Our results confirm previous findings. Chile shows the highest prevalence rate of psychiatric morbidity in PHC in the world. Our findings contradict the assumption that people from "developing countries" report more somatic than psychological symptoms.

PRIMARY CARE IN SANTIAGO, CHILE: MENTAL HEALTH AND PSYCHOSOCIAL PROBLEMS.

María G. Rojas, M.D. Psychiatry, University of Chile, Avda La Paz 1003, Santiago, Chile; Rosemarie Fritsch, M.D. Isabel González, M.D. Berta Díaz, S.A., Fernando Lolas, M.D.

Summary:

Objective : To study the prevalence of psychiatric disorders in primary care users and their association with socio demographic variables, social problems, life events, and social support.

Method: 815 consecutive patients from 15 to 50 years were interviewed. A structured interview was applied that included: sociodemographic data, the Clinical Interview Schedule-Revised (CIS-R), a social problem questionnaire, a life event list based on Holmes and Rahe, and social support questions.
Results: The prevalence of psychiatric disorders was 49,4%. Higher scores in CIS-R were associated significantly with being women ($t=5.38$; sp 0.01); being part of a couple ($F=5.2378$; p 0.001); being less educated (t : 4.38; p 0.01); having lower income ($F= 3.2281$; sp 0.05); having more social problems ($F=51.056$; sp 0.01); no affiliation to community organizations ($t=2.6$; $p=0.010$) having problems with their couple ($t=6.73$; $p0.01$); couple separation ($t=3.01$; p 0.01); suffering an accident or an illness themselves ($t= 2.85$; sp 0.01) or a relative or friend ($t= 2.84$; p 0.01); suffering the death of a relative or friend ($t=2.63$; p 0.01) or of the spouse ($t=13.44$; sp 0.01); having retired or being fired ($t=2.28$; p 0.05) and having an important income decrease ($t=8.54$; p 0.01). Finally there was a significant correlation between CIS-R and age, and number of close persons living in the house ($r=1288$; p 0.001).

Conclusions: This study contributes important information to the planning of actions that could improve mental health at the primary care level.

ABSTRACTS DE PSIQUIATRIA Y SALUD MENTAL EN EL "1996 ANNUAL MEETING" DEL AMERICAN PSYCHIATRIC ASSOCIATION ANNUAL MEETING, NUEVA YORK Y EN EL DECIMO CONGRESO MUNDIAL DE PSIQUIATRIA MADRID, 1996, AGOSTO 23-28.

SUICIDE ATTEMPTERS BY SELF-MUTILATION.

Alejandro Gómez Chamorro, Alvaro Barrera, Eduardo Jaar, Fernando Lolas, Carlos Nuñez, Grisel Orellana, University of Chile, Department of Psychiatry.

Self-mutilators have been descriptively studied, or compared with other groups, such as psychiatric controls or personality-disordered patients. This study aimed to explore psychometric and clinical differential characteristics of suicide attempters by self-mutilation.

Among 124 female suicide attempters, there were 19 who mutilated themselves, and 89 who took overdoses. Subjects were assessed by means of semistructured interviews. Pierce's Suicide Intent Scale, SCID-P, EPQ-R, and other psychometric scales. As a group, self-mutilators differed on a less severe attempt, more frequently impulsive, committed more commonly under the effects of substances, and having mixed motivations (self-destructive/interpersonal). Diagnostically, self-mutilators had higher rates of major depression, personality disorders, and alcohol/substance abuse. No differences were observed on depressiveness, hopelessness, and suicidal ideation. On Eysenck personality dimensions, self-mutilators evinced higher neuroticism scores. Clinically, self-mutilators appear as a more psychiatrically and personality-disordered group than a representative group of suicide attempters.

BIOETHICAL DECISION-MAKING IN THE

TREATMENT OF SEVERE MENTAL DISORDERS.

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Psychiatric treatment of severe mental disorder poses particular problems and strains the patient-doctor-family relationship to a high degree. One of the main tenets of informed consent doctrine—autonomy—can be said to be absent or at least impaired on the basis of a technical labelling process. The principle of beneficence, frequently invoked for implementing therapeutic or preventive measures, may collide with the views of the patient, the family or other social agents. Justice may be invoked to justify certain practices or forms of resource allocation. On occasion, surrogate decision-making is possible and necessary to justify intervention.

On the basis of an examination of actual treatment practices, a three-step decision-making process is proposed. Diagnostic labelling should include, from the outset, a bioethical appraisal of its effects and consequences. Second a determination of disability, impairment, and handicap should be based on a contextual evaluation, beyond the purely medical or psychological data-gathering process. Third, should the patient be unable to collaborate, a surrogate decision making should be instituted, guided either by the best interests principle or the principle of substituted judgement, both of which may be applicable in psychiatry.

OCD TREATMENT AND SPECT COURSE IN ONE ADOLESCENT PATIENT

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This work describes the clinical course, treatment and SPECT correlates of a 13-year-old boy who presented a severe OCD.

David a 13-year old boy presented severe obsessional

thinking related with fears of parents accidents or death and compulsive rituals of touching and checking. He presented associated symptoms: refusal to attend school and isolated conducts. His Yale Brown Score was 27.

Neurological and EEG examination was normal. SPECT findings showed an incremental perfusion of cingulate gyrus and hypoperfusion in the left dorsal frontal lobule compared with a normal sample ($+2SD$).

He began a multimodal treatment: education, fluoxetine (60 mg) and behavioral techniques and social approaches.

At the 6th week began to show clinical improvement: 10th week significant improvement, Yale Brown Scale was 7 and SPECT was normalized: 15th week he returned to school (fluoxetine 40 mg): 26th week was asymptomatic.

This work demonstrates abnormal SPECT in a severe OCD 13 year-old patient and its normalization according to clinical improvement with an appropriate treatment. There are few studies about OCD and SPECT in children and adolescents and this study is a contribution in this field.

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DIFFERENT CONCEPTS OF PERSONALITY AND DEPRESSION

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In recent years, the different DSM concepts of «personality disorders» have strongly influenced the development of psychopathological models.

These concepts have contributed to a significant increase in research on the field of personality disorders, but have unfortunately had the effect of relieving clinicians and researchers of the obligation to take into account the complex interacting psychological, environmental and historical factors that characterize patients who suffer more chronic conditions. The DSM-IV proposal of a «Depressive personality Disorder» seems to be a interesting contribution to the field of personality research and its relationship to mood disorders. Nevertheless, despite the substantial overlap that may exist among this concept and the concept of Dysthymic Disorder, it may also introduce confusion into research and clinical work on mood disorders. German and Japanese authors have described independently a specific personality structure related to unipolar-melancholic depression, characterized by being rigid, dependent, very orderly and ambiguity intolerant. The relationship of these features with unipolar major depression has been demonstrated in several psychometric studies. Personality features of individuals with Dysthymic and depressive Personality Disorders have been suggested as being significantly different to the traits mentioned above. The meaning of these discrepancies and the consequences of the inclusion of a concept like "Depressive Personality Disorder" for the future of research on these topics will be discussed.

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CULTURAL MODELING OF QUALITY ASSURANCE STANDARDS IN MENTAL HEALTH CARE.

Fernando Blas, University of Chile. Dept. and Chair of Psychiatry.

One of the best known hindrances to appropriate epidemiological data on mental health problems and the consequent design of services is the lack of culturally fair data bases. Several attempts have been made to improve communication among mental health professionals and to properly evaluate needs in a quantitative

sense. Cultural expectations shape basic demographic characteristics screened and reported by experts type and nomenclature of neuro-psychiatric diagnoses, manifestations of distress in primary care settings, impairment, disability, and handicap associated with morbidity and care-seeking and illness behaviours. A qualitative approach to dealing with these issues is needed in order to devise appropriate intervention strategies and to design evaluative programs geared towards establishing quality of mental health care. After reviewing these aspects case-studies will be proposed and described.

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SUICIDE ATTEMPTERS BY SELF-MUTILATION

Alejandro Gómez Chamorro, Alvaro Barrera. Eduardo Jaar, Fernando Blas. Carlos Nuñez, Grisel Orellana. University of Chile, Department of Psychiatry.

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PROLACTIN RESPONSES DEXFENFLURAMINE IN BPD

Sonia Jerez Concha, Hernán Silva Ibarra, Elisa Paredes Merino, Alejandra Ramírez Coronel, Patricia Rentería Cruz. Universidad de Chile, Psiquiatría y Salud Mental, Avda. La Paz 1003. Santiago, Chile.

Objective. This study investigates the serotonergic central function through prolactin responses to a challenge with dexfenfluramine in a group of patients with Borderline Personality Disorder. **Method:** 37 patients with Borderline Personality Disorder were selected using DSM-III-R and Diagnostic Interview for Borderlines (DIB-R) and another 20 control subjects were selected using Goldberg Inventory. Two blood samples were taken for each subject to determine basal prolactin levels, after rest and fast. They were then given 60 mg of dexfenfluramine. Blood samples were taken for each of the next five hours. The samples were centrifuged and frozen. Prolactin levels were measured using radioimmunoanalysis (RIA). **Results:** The response of prolactin to the challenge of dexfenfluramine was less in Borderline Personality Disorder than in the control group. The differences between the basal prolactin level and the level after 5 hours was significantly bigger in the control group than in the Borderline patients. **Conclusions:** Prolactin's lower response to the challenge of dexfenfluramine in the group of BPD is compatible with the hypothesis of serotonergic dysfunction.

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Indicaciones

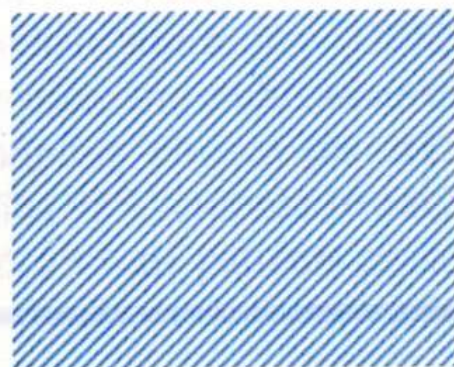
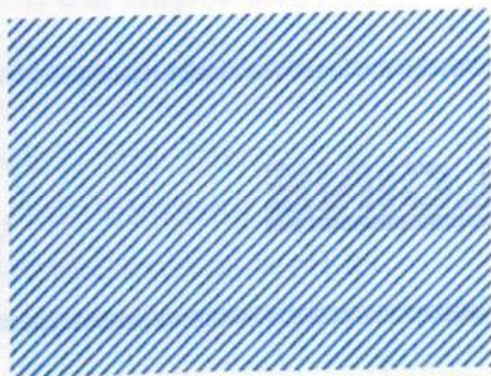
- Colopatía funcional
- Síndrome de colon irritable
- Alteraciones del tránsito intestinal y vaciamiento gástrico
- Dispepsia: Meteorismo, flatulencia, sensación de plenitud estomacal
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Synthélabo

PSYCHOLEXICOLOGY

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Nosological taxonomy and nomenclature constitute essential aspects of psychiatric classification, a prerequisite for sound scientific development.

Lexicology is a discipline aimed at the study of words according to their meaning, etymology, and social legitimacy. As a sociality constructed discourse, psychiatry depends on shared meanings established under some form of orthodoxy, which includes rhetoric, mode of knowledge production, preferred or specific audience, and ways of handling discrepancy and heresy. A psycholexicological enterprise is geared towards establishing the boundaries of proper psychiatric discourse, its social use, and its relation to professional jurisdiction.

An examination, on a comparative basis, of current terminology and classification, uncovers the structure of disciplinary discourse and leads to proposals for further study. It is hypothesized that psychiatric discourse, as a space of collective representation, should be viewed, in both its instrumental and its hermeneutic aspects, as reflection of the disciplinary and professional process of reality construction.

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COMMUNICATION OF EMOTIONAL MEANING. A DIAGNOSTIC AXIS RELEVANT IN BIOPSYCHOSOCIAL

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Emphasis on primary care and on the importance of the humanities and the social sciences for the practice of psychiatry characterize recent developments in medical theory. Revised model, the bioethical discourse, the influence of systemic thinking and of economic theory can be considered cause as well as

outcome of these developments. The social construction of illness, sickness and disease and the heavy impact of somatoform disorders upon health care systems underscore the need for a reappraisal of communication and emotional expression in diagnosis, prognosis, and therapy.

Work done on the basis of content analysis of verbal behavior during the last years will be reviewed, aiming at reformulating its basic tenets and the scope of its applications within a biopsychosocial framework. The proposal to establish «communication of emotional meaning» between health care giver and patient as an objectifiable axis or diagnostic dimension, comparable to the ones already in use for DSM-IV and ICD-10, derives from the results of these studies. Data stressing its relevance are used to substantiate the proposal and its further development and application.

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DETECTION OF PSYCHIATRIC MORBIDITY

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Objetives: To examine primary care physicians (PCPs) detection rates of psychiatric morbidity and patients and doctors variables which influence these rates.

Method: Psychiatric morbidity was assessed using General Health Questionnaire (GHQ-12) and Clinical Interview Schedule Revised (CIS-R). PCPs assessed patient's mental health blindly with rating scales. Patients and doctor's variables influencing the detection of psychiatric morbidity (DPM) were examined using univariate and multivariate methods. Results: 4.079 randomly chosen patients (95% response) and 67 PCPs (93% response) from 23 PHC Clinics participated. 53% of patients presented a

psychiatric disturbance and approximately half were detected by PCPs. According to PCPs, 42 % of their patients showed a psychiatric disturbance. Variables related to patients, particularly reason for consultation, showed the closest association with the DPM. Undetected "cases" consulted more frequently and took more tranquilizers than detected "cases".

Conclusions: This is the largest study ever done on this subject in South America. Our results confirm previous findings. Chile shows the highest prevalence rate of psychiatric morbidity in PHC in the world. Almost one of every two patients suffering from a psychiatric disturbance goes undetected by the PCP. Detection rates are more influenced by variables related to the patient than those related to doctors.

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PSYCHIATRIC MORBIDITY PRIMARY CARE

Ricardo Araya, Graciela Rojas, Julia Acuña, Univ. de Chile, Department of Psychiatry, Av. La Paz 1003, Santiago, Chile

Objectives: To estimate the prevalence rate of psychiatric morbidity among PHC patients and association with sociodemographic factors and frequency of consultations.

Method: A cross-sectional survey of psychiatric morbidity was made of randomly chosen attenders to PHC clinics. A structured interview which contained the General Health Questionnaire (GHQ-12), Clinical Interview Schedule-Revised and other questions was used. Reported frequency of consultations over last 6 months was confirmed with medical records. Variables were examined using univariate and multivariate (logistic regression) methods.

Results: 4,079 randomly chosen patients (95 % response) from 23 PHC Clinics participated in this study. The prevalence rate of psychiatric morbidity was 53% 5% of the sample gave a psychological reason for consultation. Anxiety was the most prevalent symptom. All individual symptom. All individual symptoms were more prevalent among women. Previously married women with poor income and education were at higher risk. Attenders with psychiatric morbidity consulted more often even after controlling for physical illness.

Conclusions : This is the largest study ever done on this subject in South America. Our results confirm previous findings. Chile shows the highest prevalence rate of psychiatric morbidity in PHC in the world. Our findings contradict the assumption that people from "developing countries" report more somatic than psychological symptoms.

DOES THE GHQ-12 SHOW A CULTURAL BIAS WHEN USED IN DIFFERENT COUNTRIES.

Ricardo Araya, Graciela Rojas. Univ. De Chile, Department of Psychiatry. Av. La Paz 1003. Santiago, Chile.

Objectives: To test the hypothesis that the GHQ-12 shows an ascertainment bias when comparing rates of psychiatric disorder between a European (U.K.) and a Latinamerican (Chile) country.

Method : 163 Chilean and 107 British primary care attenders completed GHQ-12 and Clinical Interview Schedule-Revised (CIS-R). The CIS-R and GHQ-12 scores were calculated for both samples. The differences between GHQ scores in the Chilean and British samples were adjusted for several variables using multiple regression techniques.

Results: Our hypothesis was confirmed. The mean CIS-R score was similar for British and Chilean but the mean GHQ-12 score was significantly higher in the Chilean sample. This difference was explained almost entirely by the difference in the negative scale ("yesness") of GHQ-12. Opposite to the British sample, subjects in the Chilean sample had higher scores on the negative scale than the positive scale. There was only a modest change in the size of the differences between the samples after adjustment.

Conclusions: Responses to GHQ are probably influenced by cultural factors. Latinamericans have higher scores on the part of GHQ assessing negative aspects of mental health. This limits the usefulness of GHQ in cross-cultural comparisons and possibly when comparing population from different countries within a single country as well.

GENDER DIFERENCES IN ONSET OF SCHIZOPHRENIA AND PUBERTY

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Gender differences have been observed consistently in age of onset and frequency of schizophrenia, males presenting an earlier onset and a bigger frequency. Some authors have suggested that sex differences in age of puberty could explain the earlier onset of schizophrenia in males. In Chile, a bigger sexual dimorphism in age at puberty has been reported. If a relationship exists between sexual dimorphism in schizophrenia and puberty, a bigger sexual dimorphism in schizophrenia would be expected in Chile. **Method:** The study was conducted in 400 schizophrenic according to DSM-III-R criteria. Age of onset was defined as the emergence of psychotic symptoms. Information was obtained from medical records and by direct interviews. **Results:** Males (68.5%) showed a higher frequency of schizophrenia than females (31.5%) both in the total sample and in all subtypes. The mean age of onset in females patients (21.1;SD=6.7) was significantly greater than in males (18.8;SD=4.5) ($p<0.05$). Males had an earlier onset in all subtypes of schizophrenia and showed an incidence peak between 15 and 19 years. Though most of the females became psychotic before age 30 the percentage of them which became ill thereafter was significantly greater than males. **Conclusion:** The sexual dimorphism in schizophrenia observed in Chile both in frequency and age of onset does not differ from that reported for caucasian populations.

RELATIONSHIPS BETWEEN PUBERTY AND ONSET OF SCHIZOPHRENIA.

Aída Ruiz, Rafael Blanco, Jaime Santander, Claudia Almonte, Eduardo Miranda, University of Chile, Medical School, Department of Psychiatry. (1),Cell Biol & Genetics (2) Avenida La Paz 1003, Santiago, Chile

According to some neurodevelopmental hypothesis of schizophrenia, puberty triggers the neuromaturation events that produce psychosis. Sexual dimorphism in age of onset of puberty could explain the later onset of schizophrenia in females. Chilean girls have an earlier age at menarche than their european counterparts. Therefore, schizophrenic chilean females would have a later onset of psychosis than european females. The objective of this study is to analyze the relationship between puberty (age of menarche) and the onset of schizophrenia.

Method: A sample of 105 schizophrenic females, according to DSM-III-R criteria, were selected. The information was obtained from medical records and by medical interviews. Age of onset of psychosis was defined as the emergence of psychotic symptoms and age of menarche as onset of menstruation. **Results and Conclusion:** The mean age of onset of psychosis was 19.92 (SD=5.12) and the mean age at menarche was 12.99 (SD=1.49), significantly later than the general population of Santiago, Chile ($p<0.01$). The subtype which had the later onset (paranoid) also had the later age at menarche. However, no correlation was observed between them either for the total sample or for each subtype. According to these results it is likely that sex differences in schizophrenia is not due to sexual dimorphism in puberty.

PSYCHIATRIC EDUCATION IN THE AMERICAS: PSYCHIATRIC TRAINING IN SOUTH AMERICA

Rodolfo Fahrner, Fernando Lolas-Stepke. University of Buenos Aires. Department of Mental Health. J. Salgueiro 24368

Objectives: Current aspects of psychiatric training in several Latin American countries, particularly Argentina and Chile where residency training programs do have a well-established identity, will

be presented and discussed. Method: Review of curricula, personnel, training guidelines and comparative analysis of the same.

Results: Psychiatric training in the eve of 21st century in Latin American countries will have to be based on the following principles. 1) The revolutionary effects of molecular genetics and diagnostic technology, in particular neuroimaging techniques that permit the immediate study of structure and function of the living brain, 2) the emergency of dichotomus criteria that challenge the traditional psychodynamic approach and are reflected in the strong resurgence of neuropsychiatric; 3) impact of economic factors in the provision of mental health care, i.e. cost benefit analysis; 4) the various levels of medical and psychiatric care; primary care, primary psychiatric care and the subspecialties.

Conclusions: The world of culture and science has entered the 21st century. Psychiatry is the branch of medicine that has probably experimented the fastest pace of development and transformation. These changes will not stop as they will modify concepts and contribute to the development of new techniques. New criteria and methodologies to solve the psychiatric and psychosocial needs of the general population will be required. In many Latin American countries, post graduate training is not systematized and there exists several teaching methodologies, some of them still outside the residency system.

THE CHILDREN PSYCHIATRIC CARE SYSTEM-FUTURE PERSPECTIVE

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According to the mode of financing three sectors in the Chilean health care system can be identified: a public, semi-public and a private. Psychiatric care on the whole is organized around ambulatory services. Psychiatric hospitals and psychiatric departments in general hospitals. The latter are fairly new in Chile and growing, what concerns number of beds and number of units. In 1990 psychiatric beds in general hospitals for acute patients have been 239 whereas in former years psychiatric services concentrated on

the capital Santiago in 1970- 75% of all psychiatrists on Chile resided in Santiago-the equipment with psychiatric facilities in the rural areas is improving-now only 59% of all psychiatrists work in Santiago. Compared to international bed rates for psychiatric patients the bed rate in Chile is very low: 21 for 1000 inhabitants. In 1970 the rate has been 0.454 per 1000 inhabitants. This process reveals the effect of improving outpatients care and building up extramural facilities for chronic patients replacing hospital beds. The Chilean psychiatry is traditionally related to European and North American concepts of psychiatry. Undergraduate teaching emphasizes. Postgraduate training at university centres lasts 3 years. The scenario of psychiatric care in Chile is quickly changing to modern service structures similar to those being found in Western European countries.

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MENTAL HEALTH AND PSYCHOSOCIAL PROBLEMS

Graciela Rojas, Ricardo Araya, Rosemarie Fristch, Isabel González, Fernando Lolas, Berta Díaz. University of Chile. Department of Psychiatry and Mental Health. Avenida La Paz N 1003, Santiago, Chile

Objective: To study the prevalence of psychiatric disorders in primary care users and their association with sociodemographic variables, social problems, life events and social support.

Method: 815 consecutive attenders to general morbidity from 15 to 50 yrs. were interviewed. A structured interview was applied which included, sociodemographic data, the Clinical Interview Schedule-Revised (CIS-R), a social problem questionnaire, a life event list based on Holmes and Rahe and social support questions.

Results: Psychiatric disorders prevalence was 49,4%. Higher scores in CIS-R were associated significantly to: being women ($t=5.38$; $p<0.01$) having a couple

($F=5.2378$, $p < 0.01$): being less educated ($t=4.38$; $p < 0.01$); having lower income ($F=3.2281$; $p < 0.05$); having greater social problems ($F=51.056$; $p < 0.01$); no affiliation to community organizations ($t=2.6$; $p < 0.01$); having problems with their couple ($t=6.73$; $p < 0.001$); couple separation ($t=3.01$; $p < 0.01$); suffering or an illness themselves ($t=2.85$; $p < 0.01$); or a relative or friend ($t=2.84$; $p < 0.01$); suffering the death of a relative or friend ($t=2.63$; $p < 0.01$) or of the spouse ($t=13.44$; $p < 0.01$); having retired or being fired ($t=\text{income decrease}$ $t=8.54$; $p < 0.01$). Finally there was a significant correlation between CIS-R and age, and number of close persons living in the house ($r=0.1288$; $p < 0.01$).

Conclusions: This study contributes with important information to the planning of actions that could improve mental health at primary care level.

SERVICE USE, HEALTH, PSYCHOSOCIAL FACTORS

Graciela Rojas, Ricardo Araya, Rosemarie Fritsch, Isabel González. University of Chile, Psychiatry and Mental Health, Avenida La Paz N 1003. Santiago. Chile

Objective : To study the association among frequency of medical consultations physical and mental disorders, sociodemographic factors and social problems.

Method: Across-sectional survey was undertaken of 815 consecutive attenders to four primary care clinics. These patients answered a structured interview which included: a Sociodemographic Questionnaire, the Social Problems Questionnaire, the Clinical Interview Schedule-Revised, the Whitley index and a Service Use Questionnaire.

Also patients were assessed on various dimensions by physicians using rating scales.

Results: The use of medical services was significantly higher in women ($p < 0.01$); widower and separated ($p < 0.01$); older ($p < 0.01$); less educated ($p < 0.01$); poorer ($p < 0.05$); subjects. Also patients with high scores on the CIS-R, the Whitley index and physician's rating scale of physical illness had a significantly higher frequency of consultations.

Conclusions: Psychiatric and sociodemographic

factors are as important as physical illnesses to determine the frequency of medical consultations. This important finding must be considered when planning health services.

USE OF PSYCHOACTIVE DRUGS IN PRIMARY CARE

Graciela Rojas, Isabel González, Rosemarie Fritsch. University of Chile. Psychiatry and Mental Health. Av. La Paz 1003, Santiago. Chile.

Objective: To study the prevalence of psychoactive drug use and some of its characteristics at the primary level.

Method: Across-sectional survey was undertaken of 815 consecutive attenders to four primary care clinics. These patients answered a structured interview which included: a Sociodemographic Questionnaire the Clinical Interview Schedule-Revised and a Psychoactive Drug Use Questionnaire. Also patients were assessed on various dimensions by physicians using rating scales.

Results: The annual prevalence for psychoactive drug use was 26.6% from which 66.8% was with medical prescription. Benzodiazepines were the most frequent drug used by the patients (76.4%). Positive associations with statistical significance were found with sex (being women. $OR=2.8$; $1.49 < OR < 5.32$; $p < 0.001$) and with psychiatric disorder ($OR=3.0$; $1.82 < OR < 5.00$; $p < 0.001$)

Conclusions: General physician training could be very important for the correct use of psychoactive drugs by the population.

FAVORABLE OUTCOME TO FLUOXETINE IN BPD.

Sonia Jerez Concha, Hernán Silva Ibarra, Angélica Paredes Merino, Jezabel Salvo Hormázabal, Cristian Montes Aguirre. Universidad de Chile Psiquiatría y Salud Mental. Avda. La Paz 1003, Santiago, Chile.

Objective: This study evaluates the therapeutic effect of Fluoxetine, a selective serotonin uptake inhibitor, in Borderline Personality Disorder. **Method.** 37 patients with Borderline Personality Disorder according to DSM III-R and Diagnostic Interview

for Borderlines (DIB-R) criteria, were given fluoxetine 20-60 mg. for six weeks. They were evaluated each week using Brief Psychiatric Rating Scale (BPRS), Global Assessment of Functioning Scale (GAF), Hamilton Depression Rating Scale (HDRS) and a clinical Impulsivity Scale. Results: There were significant improvements in BPRS, HDRS, GAF and Impulsivity Scale from the first week of the treatment. These improvements continued until the sixth week of treatment. The favourable outcome was not only due to the improvement in depression and impulsivity scores, but also to the decline of global psychopathology. Conclusions: The data suggest that Fluoxetine is an effective pharmacologic treatment for Borderline Personality Disorder. These findings support the hypothesis of a 5-HT dysfunction in Borderline Personality Disorder.

PIMOZIDE IN THE TREATMENT OF DELUSIONAL DISORDER.

Hernán Silva Ibarra, Sonia Jerez Concha, Alejandra Ramírez Coronel, Patricia Rentería Cruz, Nelly Aravena Alarcón. Universidad de Chile, Psiquiatría y Salud Mental. Av. La Paz 1003, Santiago, Chile.

Objective: Evaluate the pimozide effect on the psychopathology of Delusional Disorder.

Method: seven patients with Delusional Disorder according to DSM-III-R criteria, participated in 16-week trial with alternative periods of pimozide treatment (2-12 mg/d) and placebo. They were evaluated every week using the Brief Psychiatric Rating Scale (BPRS), the Global Assessment of Functioning Scale (GAF) and the Kendler scale for delusions (modified). Plasma levels of pimozide were determined each week by High Performance Liquid Chromatography (HPLC). Correlations between clinic and pharmacologic variables were studied by Spearman Rank-Order Correlation. Results: All patients improved with pimozide treatment. A good correlation between pimozide plasma levels and clinical response was found in five of them. The measures of the Kendler scale for extension and pressure of delusional beliefs improved, but the conviction and sistematization

remained unchanged. A good correlation between the GAF, BPRS and Kendler scales was found. Conclusion: this study indicates that pimozide is an effective treatment for Delusional Disorder with differential effects on the various dimensions of delusion. The measurement of plasmas levels of pimozide may be useful for monitoring treatment.

PHOTOTHERAPY IN 20 DEPRESSED PATIENTS.

Luis Risco Neira, Fernando Lolas Stepke. Universidad de Chile. Psiquiatría y Salud Mental. Avda. La Paz 1003, Santiago, Chile.

Objective: To evaluate the response of 20 depressed patients (Hamilton Depression Scale Score more than 20), 13 with seasonal pattern, to phototherapy (2.500 lux-2hrs/day-10 days).

Method: 20 depressed patients were recruited following DSM-IV criteria for Mood Disorders. 17 were Unipolar and 3 Bipolar. 13 with the Rosenthal criteria for Seasonal Affective Disorder (1). All of them were exposed to similar phototherapy schedule, and evaluated with HDS in the days 0.5 and 10.

Results: 14 patients responded with a fall of 50% or more of initial HDS score at 10th, day, 10 with seasonal pattern, 12 unipolar. Six patients were non-responders, 3 with seasonal pattern, 5 unipolar. Differences in age and sex between both groups were not statistically significant. Conclusion: This is a sample obtained in the South Hemisphere and exposed to phototherapy between June and September. Not all the patients with seasonal pattern presented a positive response to phototherapy, and there were patients without seasonal pattern with a good response. Probably the response to phototherapy could not depend only on seasonality.

GENDER, AGE, AND EFFECT EXPRESSION.

Teresa Sanfuentes, Irene Schiattino, Sonia Jara, Marcela Larraguibel, Fernando Lolas. University of Chile, Department of Psychiatry, Avenida La Paz 1003, Santiago, Metropolitana, Chile.

This paper examines the influence of gender and age on hostility and anxiety scores derived from written

response to the instruction to narrate a drama event in the person's life using the method of Gottschalk and coworkers.

429 subjects (288 male, 141 female) provided verbal samples for scoring forms of anxiety: death, mutilation, separation, guilt, shame, and diffuse anxiety by reliable raters employing a clause-by-clause method. Data from 376 subjects (260 male, 116 female) were used for evaluating hostility directed outwardly, overt and covert, directed inwardly, and ambivalent. All belonged to the middle socioeconomic level as assessed by a modified Graffar index and were under evaluation for job allocation.

Using non-parametric statistics, main results can be summarized as follows: Male subjects present significantly higher guilt anxiety scores; no significant gender differences were evinced in hostility. Between 19-24 years, men express higher Ambivalent Hostility and between 45-49 years higher Hostility Direct Outwards than women. Shame anxiety was higher in men aged 30-34 years than in women within this age range.

The discussion considers cultural influences on gender-related verbal expression of affect, modulated by age and the context of study. Processes relate to the organization of experience for its written expression should be further explored. Data presented are relevant for diagnostic and prognostic purposes.

THEMATIC CHOICE SOCIODEMOGRAPHIC VARIABLES.

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One of the aims of using the Content Analysis of Verbal Behaviour in the study of the relationships between characteristics of the message and sociodemographic variable of the individual.

A non-clinical sample of 377 subjects (115 women and 288 men) rated through the Graffar Social Scale and the method of Content Analysis of Verbal Behaviour, permitted its classification according to socioeconomic status and thematic choice (accidents,

death, illness, family, couple, paternity, education, work, military, service, religion, recreation, others) in spontaneous written responses to a standardized instruction. The purpose of this work is to describe and evaluate the association between these variables. This study was complemented with a multivariate statistical analysis (Analysis of correspondence, aimed at obtaining a graphic representation of data which reveal association between socioeconomic group and thematic choice. Gender influences on thematic content indicated that women belonging to higher and lower socioeconomic strata deal preferentially with family issues, whereas men allude more frequently to accidents and life events.

Data presented underscore the importance of thematic choice for characterizing subjects' samples in psychological research.

AMERICAN SOCIETY OF CLINICAL ONCOLOGY

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PRIMARY CARCINOMA OF THE GALLBLADDER (PCG): A RETROSPECTIVE STUDY
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Fifty-three cases of PCG were referred after surgery to the SO-HCUCh, over a period of 9 years. Impact of age at diagnosis, sex, histology, stage and treatment modalities on survival was retrospectively studied. Average age at diagnosis was 63 y. for men and 57 y. for women, 88% of the patients (pts) were females.

Commonest symptoms were those of cholestasis and cholecystitis and no specific pattern of presentation was observed. 48 pts were studied with ultrasound and in 35% the examination was suggestive of PCG. In 12% the diagnosis was made by extemporaneous histopathologic study of the specimen. There were no pts with Nevin I and 34/53 had Nevin V lesion. In this series 12/53 pts consulted only 1 time, in the remaining 41 pts the median survival time (MST) from the date of diagnosis was 8.3 months. Male pts

had a MST of 4,2 m, while females had MST 8,3 m. Pts who presented with stage V disease did poorly (6,9 m.> compared to all others stages, pts with Nevlin II had a MST of 23 m. Adenocarcinoma was the only histological tipe. Pts with papillary subtype had MST of 3 m. while the others with tubulo-papillary subtype had MST of 11 m. There wasn't any difference on MST among the differents histologic grades. Pts treated with 5-FU had MST of 13,5 m.

Since at present our lab. test and imaging studies are not very helpful in diagnosis of PGC we believe that a high index of suspicion should be maintained to diagnose this disease early and have a better outcome. It's necessary to test new and different therapies for pts with advance disease.

INMUNOLOGIA

Clin Exp. Immunol 1996; 105: 39-45

COMPLEMENT-DEPENDENT CYTOTOXIC ANTIBODIES TO HUMAN T LYMPHOTROPIC VIRUS TYPE I (HTLV) - INFECTED CELLS IN THE SERA OF HTLV-I-INFECTED INDIVIDUALS.

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* Hospital Clínico U. de Chile, Serv. Inmunología.

SUMMARY

To investigate wheter HILV-I induces the development of complement-dependent cytotoxic antibodies in humans, sera of asytmomatic HILV-I carriers and of patients suffering from tropical spastic paraparesis/HIVL-I associated myelophaty (TSP/HAM) or adult T cell leukemia (ATL) were used in a cytotoxicity assay against a panel of target cells. This panel included uninfected cell lines (CEM, Jurkat, Molt and H9), cell lines chronically infected with HTLV-1 (MT2, MT4, C91PL and HUT 102), as well as lines H36 (H9) infected with HTLV-1), H9-IIIB (H9 infected with HIV IIIB) and H9-MN (H9 infected with HIVMN). HTLV-I+ sera induced lysis of H36 and of lines expressing HTLV-I antigens in the presence of rabbit complement, but did not lyse cells in presence of human complement. The HTLV-I sera

also failed to lyse the HTLV-I lines and H9 cells, suggesting that lysis was specific for HTLV-I. H36 cell lysis was prevented by IgG depletion of the sera and by dialysis of rabbit complement against EGTA or EDTA. Rabbit complement-dependent cytotoxic antibodies were present in the sera of the TSP/HAM patients. Such antibodies were also detected in 5/5 individuals coinfectd with HIV-1 and HTLV-I, although no cytotoxic antibody could be found against HIV-infected cells. Vice versa, sera of HIV-1 infected individuals did not exert a lytic effect in the presence of complement (of human or rabbit origin) against HIV-1 or HTLV-I infected cells. Incubation of the sera of four HTLV-I infected patients with HTLV-I env-specific synthetic peptides demonstrated that some of the complement-dependent cytotoxic antibodies recognized epitopes located on gp46 between amino acids 190 and development of disease.

LINFOMA NO HOGDKIN PRIMARIO DE VEJIGA, REPORTE DE UN CASO.

Dres. C. Camargo, J. Gallardo, B. Comparini, H. Harbst*, E. Mehre, M. Fodor, P. Salman, M. Yañez. (Servicio de Oncología y *Departamento de Urología, Hospital Clínico Universidad de Chile).

INTRODUCCION: Los Linfomas primarios de vejiga son una enfermedad muy infrecuente, con un número pequeño de casos reportados en la literatura internacional. Nosotros describimos un caso de linfoma no Hodgkin primario en vejiga.

CASO CLINICO: Paciente de 70 años con infecciones urinarias a repetición y hematuria macroscópica. Se pesquisó al examen físico masa sólida, pélvica. Hemograma: anemia leve, función hepática, renal, radiografía de tórax y Papanicolau normales. Ecografía ginecológica y transvaginal demostraron una masa pélvica, sólido quística, de 8x7x8 cms, hipervascularizada. CA-125: 7 U/ml. TAC de pelvis evidenció una masa sólida uterovesical de contornos irregulares, de 9 cm de diámetro, sin otras alteraciones. Cistoscopia mostró una mucosa engrosada e irregular. Biopsia: Linfoma no Hodgkin difuso, de grado intermedio. La etapificación confirmó un estadio I-AE. La paciente fue tratada

con 6 ciclos de quimioterapia, esquema CHOP- Bleo, seguido de radioterapia. En control con TAC y cistoscopia, después del tercer ciclo se evidenció una remisión completa, la cual se mantiene hasta la actualidad.

CONCLUSION: Los Linfomas no Hodgkin primarios de vejiga son vistos raramente y son responsables de al menos 1% de las neoplasias primarias de vejiga. Característicamente es más frecuente en mujeres con cistitis crónicas. El tratamiento asociado quimio-terapia más radioterapia puede otorgar curación y preservación del órgano.

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II JORNADA ARGENTINO - CHILENA DE GENETICA.

GENETICA HUMANA "A"

MAPA GENETICO DE LA HIPERTENSION ARTERIAL(1995). (Gene map of arterial hypertension). Cruz-Coke, R. Servicio de Genética, Hospital Clínico J.J.Aguirre, Universidad de Chile, Santiago.

La hipertension arterial es una enfermedad multifactorial producida por la interacción de múltiples genes con variados factores ambientales. En la última década se ha identificado dos tipos de genes que participan en la etiología de esta enfermedad. a) Los genes primarios o reguladores de la presión arterial que codifican enzimas (renina, kalikreína, kininasa, aminopeptidasa); hormonas (angiotensina, vasopresina, aldosterona, prostaglandinas) y el péptido atrial natriurético); sustratos (angiotensinógeno, kininógeno). Sus efectos son vasodilatadores o vasoconstrictores en la arteriola y retenedores o excretos de sodio en el espacio extravascular. Se están describiendo los polimorfismos alélicos de estos genes que estarían asociados a la hipertensión asencial, b) los genes tensionales secundarios producen enfermedades hereditarias de

baja prevalencia que presentan hipertensión arterial entre el 20% y 80% de los casos (riñón poliquístico, nefroblastoma, feocromo-citoma, hiperplasia adrenal, nefritis hereditaria). En total se han identificado más de 40 genes ubicados en todos los cromosomas y que se transmiten en forma dominante, recesiva y ligada al x. Se presentan dos mapas genéticos con todos los genes hipertensores.

TRISOMIA 8: REPORTE DE CUATRO CASOS CLINICOS (Trisomy 8: four cases report). Mellado, C., Moreno, R *, López, F.**, Sanz, P., Castillo, S., Vilaseca, C., Daher, V., Tobella, L., Salazar, S. Servicio de Genética Hospital Clínico de la Universidad de Chile. *Hospital Exequiel González Cortés; ** Hospital G. Fricke Viña del Mar.

La trisomía 8 es un cuadro cromosómico en que la mayoría de los casos descritos son en mosaico, su incidencia en la población general no ha sido establecida y está caracterizado clínicamente por: retardo mental leve a severo; dismorfias faciales típicas con ojos profundos, nariz bulbosa y labio inferior evertido; alteraciones esqueléticas especialmente vertebrales; función articular disminuida; camptodactilia, tórax largo; pliegues palmares y plantares profundos; anomalías renales y otras. Las características clínicas son variables desde dismorfias discretas hasta malformaciones severas. El objetivo de esta comunicación es presentar cuatro casos de individuos portadores de trisomía 8 con características clínicas del cuadro, corroborado citogenéticamente con cariograma en sangre. Tres de ellos con trisomía 8 en mosaico y uno de ellos con trisomía 8 completa, en este caso no se realizó estudio en otro tejido. Los motivos de derivación de estos pacientes para estudio en otro tejido. Los motivos de derivación de estos pacientes para estudio citogenético fueron diversos: el aspecto dismórfico, retraso del desarrollo psicomotor, retraso del lenguaje e hipotonía. Es importante tener en cuenta la existencia de variabilidad en las características fenotípicas de la trisomía 8, para sospechar el diagnóstico y solicitar el estudio citogenético.

BIOLOGIA

GENETICA HUMANA "B"

SITUACION DE LOS SERVICIOS DE GENETICA CLINICA EN CHILE (Clinical genetics services in Chile: current situation) Castillo, S. Servicio de Genética, Departamento de Medicina, Hospital Clínico Universidad de Chile.

La genética en el ámbito médico ha ido tomando mayor importancia como especialidad dados los cambios demográficos de nuestro país, que, al controlar causas prevenibles, han permitido que las malformaciones congénitas adquieran cada vez más preponderancia como causa de morbi-mortalidad en la infancia, y como causa de embarazo patológico y alteraciones del proceso reproductivo. Se revela la información obtenida de datos epidemiológicos generales y dirigidos a enfermedades genéticas y defectos congénitos. A través de una encuesta contestada por los trece centros de genética médica del país, se informa sobre la historia del desarrollo de esta disciplina en Chile, de los recursos disponibles, de los servicios existentes y de los principales problemas y eventuales soluciones. También se obtiene una visión respecto a la enseñanza de la genética médica a nivel de pre y postgrado, y del estado actual de la investigación en este tema.

FRECUENCIAS ALELICAS Y GENOTIPICAS DE MARCADORES MOLECULARES DE FISURA LABIOPALATINA EN LA POBLACION CHILENA (Allele and genotype frequencies of molecular markers of cleft lip/palate in the Chilean population). Blanco R. Depto. Biología Celular y Genética, Facultad de Medicina, Jara L. Palomino R. Villaseca C., Servicio de Genética, Hospital Clínico J.J. Aguirre. Universidad de Chile.

Recientes estudios sugieren que la fisura labiopalatina (FLP) no sindrómica se debe a la acción de varios loci de susceptibilidad. Ello debido a las diferencias interpoblacionales observadas para dichos loci. No obstante, pareciera que al fenómeno

interpoblacional mencionado se agregaría la situación de los casos simplex (un sólo caso en la familia) y multiplex (más de un caso).

En el presente estudio hemos analizado las frecuencias alélicas de MSX-1 (ROx-7), TGFA, D17S579 Y D6S89, tanto en casos simplex como multiplex, en sus respectivos parientes y en una muestra control. Los resultados muestran diferencias significativas para algunos de los marcadores mencionados al comparar casos vs controles (TGFA, D17S579 y D6S89). Cuando la información se analiza separadamente en 5 grupos, es decir casos simplex, sus familiares, casos multiplex, sus familiares y controles, las frecuencias alélicas de los marcadores mencionados muestran gradientes; en algunos casos esta es decreciente desde los casos multiplex que muestran los valores mayores hasta los controles que muestran los valores menores (los otros grupos estudiados muestran valores intermedios) en otros casos la gradiente muestra sentido inverso. Nuestros resultados apoyan la hipótesis de que en la etiología genética de la fisura labiopalatina existirían heterogeneidad genética.

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ABERRACIONES CROMOSOMICAS ASOCIADAS A FUSION INCOMPLETA DE CONDUCTOS DE MÜLLER

(Chromosomes anomalies and incomplete fusion of Muller ducts.) Figueroa, P.; Mellado, C.; Sanza, P., Castillo, S.; Salazar, S. Centro de Medicina Reproductiva del Adolescente (CEMERA) y Servicio de Genética. Hospital Clínico Universidad de Chile.

Existen diferentes malformaciones uterinas debidas a falta de fusión incompleta de los conductos de Müller. La prevalencia oscila entre 0.1 y 3%. No está indicado el estudio citogenético por esta causa y en general no se asocian con aberraciones cromosómicas, describiéndose en forma esporádica en algunos casos de trisomía 13 y delección del brazo largo del cromosoma 13. Estas anomalías uterinas son responsables del 1 al 3% de los abortos tempranos, asociándose con pérdidas del segundo trimestre. Presentamos dos casos clínicos de mujeres jóvenes con pérdida reproductiva del primer trimestre

y malformación uterina por fusión incompleta de los conductos de Müller, realizándose en ambas cariograma en sangre periférica. Una paciente fue referida al estudio citogenético por el antecedente de 4 abortos previos a la corrección quirúrgica de su defecto uterino y 2 posteriores a esta cirugía. El cariotipo muestra una translocación recíproca entre el cromosoma 1 y el 8 (46,XX,t(1;8)(q21;q24.1). La segunda paciente fue referida para consejo genético por el antecedente de 2 abortos tempranos y un cariotipo con mosaico 45,X/46,XX(15% 45,X). La paciente tiene talla baja y cuello corto. En ella se planteó la corrección quirúrgica y fertilización asistida.

En estos dos casos los abortos tempranos podrían ser secundarios a la formación de gametos portadores de aberraciones cromosómicas durante la meiosis. Esto demuestra la importancia del estudio citogenético en parejas con pérdida reproductiva aún con el diagnóstico de malformaciones uterinas.

ABSTRACTS REUMATOLOGIA 1996

ANALYSIS OF AUTOANTIBODIES TO PLASMINOGEN IN THE SERUM OF PATIENTS WITH RHEUMATOID ARTHRITIS. M. González-Gronow- M. Cuchacovich * . D.M. Grigg. S.V. Pizzo. Hospital, * Department of Medicine, Rheumatology Section. Clinical University of Chile, Santiago, Chile.

Abstract. Sera from patients with rheumatoid arthritis containing high titers of anti-streptokinase antibodies were found to contain anti-plasminogen antibodies of the IgG and IgA classes. High titers of antiplasminogen autoantibodies of the IgA class were also found in sera from patients with systemic lupus erythematosus and Sjögren Syndrome. Studies of the immune response to thrombolytic therapy with streptokinase in patients with no prior history of autoimmune disease suggest a strong correlation between streptokinase administration and the appearance of autoantibodies to plasminogen of the IgA class. The IgA anti-plasminogen autoantibody is specific for an epitope in a region of plasminogen which binds streptokinase and the IgG autoantibody reacts with an epitope in the C-terminal region

corresponding to the catalytic domain of the plasminogen zymogen. Our findings suggest a different origin for the two classes of antiplasminogen immunoglobulins in rheumatoid arthritis patients. Since plasminogen binding to rheumatoid synovial fibroblasts is enhanced, the high titers of both classes of anti-plasminogen autoantibodies may add to the localization and perpetuation of the immune response. We suggest that plasminogen may be a target of the immune response in autoimmune disease.

ABERRACIONES CROMOSOMICAS Y CITOMETRIA DE FLUJO EN ADENOCARCINOMA DE VESICULA BILIAR (Chromosome rearrangements and flow cytometry in gallbladder adenocarcinoma) Sanz P., Calvo, A., Castillo S., Tobella L., Salazar S., Daher V., Smok G., Bentjerodt MR, Csendes A., Nielsen E., Pruyas M. Serv. Genética, Anatomía Patológica y Depto. de Cirugía. Hosp. Clínico Universidad de Chile, 2 Cirugía. Anatomía Patológica Hosp. Sótero del Río Financiado con proyecto Fondecyt 194067.

Chile es el país con la tasa más alta de mortalidad por cáncer de vesícula biliar (10.6 por mil habitantes). Representa la principal causa de muerte por tumor maligno en la mujer chilena. Su etiología es actualmente desconocida. No se han reportado hasta el momento anomalías cromosómicas específicas ni primarias en estos tumores, ni directamente relacionadas con algún oncogen responsable de su desarrollo.

Realizamos simultáneamente el estudio cromosómico y de contenido de DNA (en citómetro de flujo) en 35 muestras de adenocarcinoma de vesícula biliar y en 24 contriales (vesículas sin lesión histológica cancerosa ni precancerosa). Empleamos técnicas directas y cultivos largos para obtener el cariograma. Para la citometría empleamos células desagregadas enzimáticamente a partir de muestras frescas o fijadas e incluidas en parafina. En 18 de los 35 carcinomas se obtuvo el cariotipo (63% de sucesos). En 3 de estos 18 se encontró una sola población con cariograma normal, debido a que hubo proliferación exclusiva de células del tejido de sostén en el cultivo. En los

restantes 15 casos se encontraron dos poblaciones distintas, una con cariograma normal y otra con la hiperploidías (neartriploidías, neartetraploidías). Esto fue corroborado en el estudio de citometría, que mostró dos poblaciones celulares, una con un contenido de DNA normal y otra con hiperploidías. Confirmamos que las hiperploidías evidenciadas con anabas técnicas constituyen una alteración que se presenta en estados avanzados de este cáncer. Tres de los 24 controles fuera de las células con cariograma normal presentaron 4 o más células tetraploides propias del cultivo. Dos de estos además presentaron una población mosaico 45,X/46XX y 45X/46XY cuyo significado no sería patológico. Los encontraron anomalías del contenido de DNA.

También encontramos múltiples anomalías cromosómicas estructurales en 13/15 carcinomas, tales como: translocaciones, deleciones, fragmentos cromosómicos, inversiones, isocromosomas anillos y cromosomas marcadores. Las alteraciones cromosómicas que se repiten en dos o más casos: del (4) (q³²) del (17)(p12) y t(6;13)(p²³;q/y) pueden ser específicas y/o primarias. Esto deberá ser verificado analizando un mayor número de casos, usando estrategias que permitan eliminar células cancerosas en estados muy avanzados.

ABORTO ESPONTANEO: ESTUDIO CITOGENETICO EN 640 CASOS. (Cytogenetic study of 640 spontaneous abortions). Velásquez, P y Youlton, R. Laboratorio de Citogenética. Clínica Las Condes.

El análisis cromosómico de los abortos espontáneos (AE) ha permitido establecer que las anomalías cromosómicas son causa de más de la mitad de las pérdidas embrionarias del primer trimestre.

Desde Septiembre de 1989 a Mayo de 1996 hemos realizado estudio cromosómico en 640 muestras

obtenidas de vellosidades coriales en casos de AE. En 609 de las 640 muestras procesadas (95.1%) se obtuvo células en mitosis aptas para estudio cromosómico. El porcentaje de éxito de los cultivos fue de 97.9% para las muestras recibidas del pabellón de Gineco-obstetricia de nuestra Institución, de 96.6% para las muestras de otras clínicas de Santiago y de 78.4% para aquellas derivadas de otras ciudades. De las 609 muestras estudiadas 388 presentaron un Cariotipo anormal (63.7%). En ellas la relación XY:XX fue 1.03 al igual que con cariotipo normal. Las trisomías autosómicas fueron la anomalía más frecuente (239/388 casos [61.6%]). 21 de ellas en forma de una doble trisomía y un caso con una triple trisomía. La trisomía 16 representó casi un tercio de las trisomías. Las triploidías se observaron en 62 casos (16%) y las tetraploidías en 17 casos (4.4%), diez de las cuales fueron en mosaico con una línea celular normal. Monosomías fueron detectadas en 41 muestras (10.6%). 39 correspondían a monosomía X y dos casos a una monosomía 21. Seis de las monosomías X eran en mosaico con una línea celular normal 46.XX. En diez casos se detectó una doble anomalía 2.6%) y 19 casos presentaron anomalías estructurales diversas (4.9%). El porcentaje de AE con anomalía cromosómica fue mayor a mayor edad materna con excepción de la monosomía X.

La trisomía 16 y las triploidías. La mayor proporción de estas anomalías se observó en embriones de una edad gestacional entre seis y doce semanas.

Conclusiones: 1) En nuestro estudio las anomalías cromosómicas afectaron al 63.7% de los casos estudiados. 2) Las anomalías más frecuentemente encontradas fueron las trisomías. 3) La trisomía 16 fue la más común y en ellas el promedio de la edad materna no fue diferente del promedio de la edad materna en los AE con cariotipo normal.