

# Resúmenes de publicaciones nacionales ISI 2009

## DEPARTAMENTO CARDIOVASCULAR

REV MÉD CHIL. 2009 OCT;137(10):1315-22.

### EFFECTOS DEL TRATAMIENTO PERIODONTAL SOBRE LOS MARCADORES DE INFLAMACIÓN SISTÉMICA EN PACIENTES CON RIESGO DE ENFERMEDAD CARDIACA CORONARIA. ESTUDIO PILOTO.

López NJ, Quintero A, Llancaqueo M, Jara L.

**BACKGROUND:** Studies investigating effects of periodontal treatment (PT) on markers of inflammation in healthy subjects show conflicting results. Few studies have investigated the effects of PT among subjects with coronary heart disease (CHD) risk factors. **AIM:** To report the results of a pilot prospective study on the effects of periodontal treatment on markers of inflammation among subjects with CHD risk factors. **MATERIAL AND METHODS:** Seventy three patients aged 53+/-6 years (25% males) with chronic periodontitis, dyslipidemia and other CHD risk factors were subjected to PT consisting on root planning and oral metronidazol and amoxicillin for 7 days. Periodontal clinical parameters, serum C-reactive protein (CRP), fibrinogen levels and erythrocyte sedimentation rate (ESR) were assessed before and at 6 weeks after PT. Polymorphisms at the IL1A-889 and IL1B+3954 genes were also genotyped. **RESULTS:** After the treatment period, CRP levels significantly increased from 3.6+/-3.7 mg/ L to 5.4+/-5.7 mg/L ( $p = 0.001$ ). No significant changes were observed in fibrinogen levels and ESR. Higher post-treatment CRP levels were significantly associated with the composite polymorphic genotype at the IL1A-889 and IL1B+3954 genes ( $p = 0.0001$ ), and extensive periodontitis ( $p = 0.005$ ). Moderate alcohol consumption appeared as a protective factor for CRP elevation ( $p = 0.029$ ). **CONCLUSIONS:** The increase of the CRP levels after PT in patients with CVD risk factors appeared associated with IL-1 gene polymorphisms and extensive periodontitis.

## DEPARTAMENTO DE CIRUGÍA

REV MÉD CHIL. 2009 APR;137(4):559-66.

### COMPARACIÓN DEL TRATAMIENTO MÉDICO Y QUIRÚRGICO EN PACIENTES CON OBESIDAD GRADO III (OBESIDAD MÓRBIDA).

Attila Cséndes J, Patricio Burdiles P, Karin Papapietro V, Ana María Burgos L.

This is a review of publications comparing the results of medical and surgical treatment of morbid obesity. An overall conclusion is that the frequency of cardiovascular complications or cancer is higher among patients receiving medical treatment. Surgical treatment is associated with a better weight loss, reduction in complications and quality of life. Mortality risk decreases significantly after surgical treatment, when compared with patients receiving medical therapy. Therefore, management of morbid obesity should be carried out by multidisciplinary teams with experience on gastrointestinal surgery. In this way the complications and mortality of bariatric surgery would be minimized.

REV MÉD CHILE 2009; 137: 928-935.

**LINFOMA PRIMARIO DE TIROIDES: REPORTE DE CUATRO CASOS.**

**Patricia Gac E, Patricio Cabané T, José Amat V, Ricardo Zamorano S, Pedro Pineda B, Claudia Morales H, Catalina Valenzuela V, Francisco Rodríguez M.**

Primary thyroid lymphoma is uncommon and usually of non-Hodgkin type. We report four female patients with thyroid lymphoma. An 81 year-old patient that was operated and received three cycles of chemotherapy and was lost from follow up. A 16 year-old patient that is operated and received full chemotherapy and was discharged free of disease. A 80 year-old patient that was operated and died due to a multiple organ failure, 50 days after hospital admission and a 54 year-old female that was operated but rejected chemotherapy and was lost from follow up.

REV. CHILENA DE CIRUGÍA. VOL 61 - N° 1, FEBRERO 2009; PÁG. 83-88.

**OBESIDAD MÓRBIDA ASOCIADA A QUISTE BRONCOGÉNICO Y LEIOMIOMA SUBCARDIAL QUE SIMULA GIST. CASO CLÍNICO.**

**Jaime Rappoport S., Guillermo Watkins S., Attila Cséndes J., Manuel Lillo C., Iván Gallegos M., Daniel Rappoport W.**

La patología intramural gástrica tiene una frecuencia menor al 1%, en cirugía gástrica. Alrededor del 50% corresponden a leiomiomas. Los quistes broncogénicos tienen origen embriológico y son excepcionales, y la mayor parte de los reportes corresponden a pacientes pediátricos. Se presenta un caso clínico de paciente portadora de Obesidad mórbida (IMC = 52), asociado a lesión subcardial que simula GIST, cuyo estudio histopatológico resultó ser un quiste esofágico de origen broncogénico de 40 mm, asociado a leiomioma subcardial de 10 mm. Fue sometida a gastrectomía total más anastomosis esófago-yeunal en Y de Roux, con asa de 180 cm. No presentó morbi-mortalidad postoperatoria. Se discuten los métodos de estudio preoperatorio y las alternativas terapéuticas. No hemos encontrado en la literatura otro caso de asociación de estas raras patologías.

REV. CHILENA DE CIRUGÍA. VOL 61 - N° 3, JUNIO 2009; PÁG. 223-228.

**EMPIEMA PLEURAL: ETILOGÍA, TRATAMIENTO Y COMPLICACIONES.**

**Jorge Salguero A., Gonzalo Cardemil H., Juan Carlos Molina F., Hanns Lembach J., Jaime Fernández R.**

Introducción: Definido como la presencia de infección bacteriana en el espacio pleural, determina una importante morbimortalidad a pesar de los avances en el tratamiento antibiótico y las nuevas técnicas quirúrgicas. Objetivos: Comparar resultados de las distintas estrategias terapéuticas para el Empiema Pleural (EP). Material y Métodos: Revisión retrospectiva de fichas clínicas de pacientes con diagnóstico de EP manejados en el Hospital Clínico Universidad de Chile entre enero de 1994 y junio de 2007. Se consideró significación estadística con  $p < 0,05$ . Resultados: De 77 pacientes, 48 fueron hombres y 29 mujeres, con edad promedio de  $52,6 \pm 19,9$  años. La causa más frecuente de EP fue la neumonía (74%). En 55 pacientes se realizaron exámenes imagenológicos complementarios a la radiografía de tórax. Treinta y dos pacientes se manejaron inicialmente con pleurostomía percutánea (PP). Siete de ellos requirieron una nueva intervención por empiema residual. Once se manejaron con toracotomía abierta (TA) y 34 con Toracoscopía Vídeo asistida (TVA). La tasa de complicaciones y mortalidad fue: 34,37% y 25% para PP, 11,7% y 0% para TVA y 36,36% y 0% para TA, respectivamente (diferencias significativas). El tiempo de hospitalización promedio fue de 22,8 días, y el de tubo pleural de 9,21 días. Se encontró diferencias significativas entre los días con tubo pleural de los pacientes sometidos a TVA ( $X = 5,56$ ) respecto de los sometidos a PP ( $X = 14,24$ ) y TA ( $X = 12,4$ ). No se observan diferencias en tiempo de hospitalización. Conclusiones: La TVA demuestra obtener mejores resultados que la PP y la TA al evaluar mortalidad, complicaciones y días de tubo pleural.

REV. CHILENA DE CIRUGÍA. VOL 61 - N° 1, FEBRERO 2009; PÁG. 97-100.

**DIÁSTASIS DE LOS RECTOS. TÉCNICA QUIRÚRGICA ORIGINAL.**

**Jorge Bezama M., Aníbal Debandi L, María M. Haddad A., Pablo Bezama U.**

Introducción: La diástasis de los rectos constituye una entidad distinta de las hernias, no tiene anillo y la posibilidad que ocurra un evento obstructivo, atascamiento o estrangulación es poco probable; sin embargo, es un particular defecto de la pared que produce más bien problemas de tipo estéticos. Se describe una técnica, original del autor principal de esta presentación, para reparar este defecto cuando es de hasta de 3 cm de ancho, el que se realiza mediante un acceso a través de una pequeña incisión en la cicatriz umbilical. Los defectos mayores por su parte requieren de un acceso directo a través de una incisión cutánea realizada en la línea media supraumbilical, sobre el defecto.

REV. CHILENA DE CIRUGÍA. VOL 61 - N° 3, JUNIO 2009; PÁG. 261-265.

**OPERACIÓN DE PICKRELL O GRACILOPLASTÍA NO ESTIMULADA. ¿TIENE UN ROL EN EL MANEJO QUIRÚRGICO ACTUAL DE LA INCONTINENCIA ANAL SEVERA?**

**Christian Jensen B., Héctor Chiong L., José Luis Llanos B., Miguel Ángel Cumisile G.**

Se presenta la casuística de Operación de Pickrell o graciloplastía no estimulada realizada por uno de los autores (CJB). Se analizan sus indicaciones, técnica, complicaciones, manejo postoperatorio y resultados. Se presentan 5 pacientes (4 hombres y una mujer) operados

por incontinencia anal severa, realizándose una graciloplastía no estimulada u operación de Pickrell. Las principales complicaciones fueron dehiscencia cutánea en 2 pacientes, trombosis venosa profunda y dolor crónico de la zona de desinserción en un caso. No hubo mortalidad. En el postoperatorio se envió a estimulación eléctrica del gracilis. Se evaluó la presión del esfínter con manometría anorrectal pre y postoperatoria. La incontinencia fue medida con la escala de Jorge y Wexner. En esta serie, hubo mejoría significativa de la Presión de Contracción Voluntaria y disminución significativa del puntaje de incontinencia. La graciloplastía es alternativa a una ostomía definitiva o al implante de un esfínter artificial en aquellos pacientes en que la incontinencia anal severa no es posible de manejar con técnicas habituales (esfinteroplastía), que fracasaron a la esfinteroplastía, o que presentan inexistencia de esfínter o de una lesión anatómica que reparar.

REV. CHILENA DE CIRUGÍA. 2009; 61:467-470.

#### **CUERPO EXTRAÑO PULMONAR: RESECCIÓN POR VIDEOTORACOSCOPIA. CASO CLÍNICO.**

**Jorge Salguero A., Andrés Marambio G., Fernanda Baeza G., Juan P. Marambio G.**

Se reporta el caso de un paciente hombre de 18 años con un cuerpo extraño pulmonar de presentación tardía, que consultó por un cuadro de 2 años de evolución de episodios de hemoptisis escasa y recurrente, sin otros síntomas asociados. Se realizó estudio con radiografía y TAC de tórax evidenciándose una imagen sugerente de un cuerpo extraño metálico en los segmentos posteriores del lóbulo inferior derecho. Dado que la fibrobroncoscopia resultó frustra se decidió realizar una videotoracoscopia con resección en cuña del parénquima pulmonar afectado, encontrándose una lesión capsular de tejido inflamatorio que en su interior contenía un chinche. La evolución clínica fue favorable, con radiografía de tórax de control con adecuada reexpansión pulmonar y dándose de alta a las 48 hrs postoperatorias.

REV. CHILENA DE CIRUGÍA. 2009; 61:560-565.

#### **TUMOR FIBROMIXOIDES OSIFICANTE SIMULANDO UN CÁNCER ANAPLÁSICO DE TIROIDES. REPORTE DE UN CASO.**

**José Amat V., Andrés Marambio G., Francisco Rodríguez M., Patricio Gac E., Patricio Cabané T., María E. Villanueva L., Al. Juan Pablo Marambio G.**

El tumor fibromixoide osificante corresponde a una neoplasia infrecuente, de comportamiento benigno pero que presenta recurrencia en un tercio de los casos, comportándose como un sarcoma de bajo grado. Se reporta el caso de una paciente operada de tiroides en dos oportunidades en 9 años, cuyo diagnóstico final fue tumor fibromixoide osificante.

## **DEPARTAMENTO DE PSIQUIATRÍA Y SALUD MENTAL**

REV MÉD CHILE 2009; 137: 466-474.

#### **ADOLESCENTES CON TRASTORNO POR CONSUMO DE SUSTANCIAS: UNA CARACTERIZACIÓN DE PERSONALIDAD A TRAVÉS DE LAS NORMAS CHILENAS DEL MACI.**

Eugenia V. Vineta, Ximena Faúndez, **Marcela Larraguibel.**

**Background:** Adolescence is the most vulnerable development stage to start drug abuse in our country. Moreover, adolescent personality may be an individual risk factor for drug abuse. **Aim:** To characterize the personality of adolescent drug abusers using the Millon Adolescent Clinical Inventory (MACI) and its Chilean norms. **Patients and methods:** Adolescents consulting in a mental health facility were studied. MACI was applied to 46 subjects, aged  $16 \pm 1$  years (30 males) with a history of drug abuse and compared with 58 peers aged  $15 \pm 2$  years (34 males) without such history. **Results:** Male and female drug abuser adolescents had a transgressor type of personality. Males had a high degree of sociability and unrestrained sexuality while females present signs of sexual abuse, suicide risk, and a more severe personality disorder. **Conclusions:** The personality traits detected in this group of drug abuser adolescents using Chilean norms is similar to those detected abroad and should help to plan rehabilitation.

## **DEPARTAMENTO DE MEDICINA**

### **ENDOCRINOLOGÍA**

REV MÉD CHIL. 2009 JAN;137(1):106-14.

#### **ALTERACIONES METABÓLICAS ASOCIADAS AL USO DE TERAPIA ANTIPSICÓTICA.**

**Paula Rojas G., Catalina Poblete A., Ximena Orellana G., Karen Rouliez A., Claudio Liberman G.**

The advent of new antipsychotic drugs has improved the treatment of schizophrenic patients as well as those suffering from other severe psychiatric disorders. Its widespread use, however, has been associated to the development of obesity

and metabolic disturbances such as diabetes mellitus, dyslipidemia and increased coronary risk. This has caused a serious concern, due to the high cardiovascular mortality that prematurely affects these patients. The etiology of these abnormalities is still a matter of debate, although it is generally believed that the new antipsychotic drugs have a control stimulating effect on appetite, and their use is associated to an increased level of cortisol and to an insulin-resistance state. In addition, there is an increase in inflammatory mediator and cytokine production, induced by the pathophysiology of the schizophrenic process itself and also caused by the direct action of the antipsychotic drugs. In spite of the mounting evidence, the metabolic management of these patients is still deficient. A close follow-up in the initial stages of the antipsychotic treatment is recommended, as well as giving advice about diet and physical exercise. Finally, when obesity or other conditions associated to metabolic syndrome appear, the recommendation is to switch to drugs with less secondary effects or to add adjuvant medications to improve the overall evolution of these patients.

REV MÉD CHIL. 2009 JUL;137(7):972-81.

#### **TRATAMIENTO FARMACOLÓGICO O QUIRÚRGICO DEL PACIENTE CON SEBREPESO U OBESIDAD.**

**Carrasco N F, Manrique M, de la Maza MP, Moreno M, Albala C, García J, Díaz J, Liberman C.**

This is an updated review of the available treatments for obesity, which can be used when lifestyles modifications fail. Using the available information and the experience of the members of this advisory group, a recommendation is given about the most useful treatments, according to the severity of obesity and its complications. With regards to pharmacological treatments, only sibutramine and orlistat are approved on a worldwide basis for the treatment of obesity. These medications achieve a 10% higher weight reduction than lifestyles modification. A third medication, rimonabant, is also more effective than lifestyles modifications, but it was withdrawn due to psychological safety issues. The indications for surgical treatment and a brief description of the available techniques, success rates and complications are outlined. Finally, the need to have followed up protocols for patients and the formation of multidisciplinary treatment teams is underscored.

REV MÉD CHIL. 2009 JUL;137(7):963-71.

#### **DIAGNÓSTICO, EVALUACIÓN Y TRATAMIENTO NO FARMACOLÓGICO DEL PACIENTE CON SOBREPESO U OBESIDAD.**

**Manrique E M, de la Maza MP, Carrasco F, Moreno M, Albala C, García J, Díaz J, Liberman C.**

The risk of complications of obesity is proportional to body mass index and is higher in severe or morbid obesities and when abdominal or visceral fat is predominant. In Chile the prevalence of obesity is increasing. According to the World Health Organization, obese subjects must reduce at least a 5% of their weight to reduce the risk of complications. Although this amount of reduction is seldom achieved with non pharmacological treatments, better results are obtained with multidisciplinary/ approaches that include a medical, psychosocial and laboratory assessment, to determine obesity level and different factors involved and the associated complications. In a second stage, goals of treatment are set and a personalized treatment is designed including dietary changes and physical activity. The aim is to obtain perdurable lifestyles modifications.

## **GASTROENTEROLOGÍA**

REV MÉD CHIL. 2009 JUL;137(7):906-11.

#### **SEROPREVALENCIA DE VIRUS HEPATITIS B EN NIÑOS CON CÁNCER EN TRATAMIENTO QUIMIOTERÁPICO EN 6 HOSPITALES DE SANTIAGO DE CHILE.**

Marcela Zubietta, María E Santolaya, **Carmen Hurtado**, Ana M Alvarez, Carmen L Avilés, Ana Becker, **Javier Brahm**, Carmen Salgado, Pamela Silva, Santiago Topelberg, Juan Tordecilla, Mónica Varas, Milena Villarroel, Tamara Viviani.

**BACKGROUND:** Children under oncological therapy are at risk of infection by hepatitis B virus (HBV). **AIM:** To determine the prevalence of infection of HBV in children with cancer who have undergone chemotherapy or have had a hematopoietic stem cell transplant. **MATERIAL AND METHODS:** Collaborative, multi-centric study. Serum samples were collected from 281 children with cancer and episodes of febrile neutropenia, from 6 hospitals belonging to the public health network in the Metropolitan Region, between June 2004 and August 2006. These samples were stored at -70 degrees C. In September 2006, 200 samples were randomly chosen and 170 analyzed to determine hepatitis B virus surface antigen (HBsAg) and anticore total antibodies (anti HBc) by fluorescent ELISA (Enzyme Linked Immunosorbent Assay). In five cases in which a low volume of sample was available, only one marker was studied (HBsAg in two and anti HBc in three). **RESULTS:** Samples came from children aged 4 months to 18 years, 104 males (61%). They had received an average of 38 transfusions (range 3-107) from 65 donors (range 3-345). Twelve children were found positive for some marker of HBV: HBsAg in three (1.8%) and anti HBc in ten (7%). In 5 patients that had negative HBsAg and positive anti HBc, anti surface antigen antibodies (anti HBs) were determined and resulted positive in four. **CONCLUSIONS:** The prevalence of HBV in this sample was 7% if both serologic markers are considered and 1.8% if only HBsAg is considered.

REV MÉD CHILE 2009; 137: 185-192.

## **POLIMORFISMO DEL GEN DE LA TIOPURINA S-METILTRANSFERASA EN DONANTES DE SANGRE DE UN HOSPITAL UNIVERSITARIO.**

**Luis Álvarez L, Mauricio Venegas S, Milton Larrondo L, Natalia Becerra B, Ariel Castro L, Rodrigo Quera P.**

**Background:** Thiopurine S-methyltransferase (TPMT) is a cytosolic enzyme that catalyzes the S-methylation of 6-mercaptopurine and azathioprine. Low-activity phenotypes are correlated with polymorphism in the TPMT gene. Patients with low or undetectable TPMT activity could develop severe myelosuppression when they are treated with standard doses of thiopurine drugs. Since ethnic differences in the TPMT gene polymorphism have been demonstrated worldwide, its assessment in the Chilean population is worthwhile. **Aim:** To investigate the TPMT gene polymorphism in a Chilean blood donor individuals. **Subjects and Methods:** The frequency of four allelic variants of the TPMT gene, \*2 (G238C), \*3A (G460A and A719G), \*3B (G460A) and \*3C (A719G) were analyzed in 210 Chilean blood donors, using polymerase chain reaction (PCR), restriction fragment length polymorphism (RFLP) and allele-specific PCR-based assays. **Results:** TPMT variants associated to low enzymatic activity, were detected in 16 subjects (8%), who had a heterozygous genotype (\*3A in 12; \*3C in three and \*2 in one subject). No TPMT\*3B allelic variant was found. The normal allele (wild-type) was found in 92% of studied individuals. **Conclusions:** The allele TPMT\*3A, is the most prevalent in this group of Chilean blood donors, as in Caucasian populations.

REV CHIL INFECT 2009; 26 (3): 258-262.

## **ENDOCARDITIS INFECCIOSA EN PACIENTES CON DAÑO HEPÁTICO CRÓNICO. SERIE DE 4 CASOS CLÍNICOS.**

**Dan Oksenberg R, Anna Castelli T. y Alberto Fica C.**

La asociación entre daño hepático y endocarditis infecciosa es infrecuente. Para analizar los factores predisponentes de esta asociación, la etiología microbiana y evolución clínica, se efectuó un análisis retrospectivo de los egresos por endocarditis infecciosa en pacientes con cirrosis hepática desde 1995 a junio de 2008. Se identificaron cuatro casos, asociados a categoría Child A en tres y en todos había una cardiopatía predisponente. Las manifestaciones clínicas fueron clásicas excepto en un caso que se presentó como descompensación hepática. Sólo un caso se asoció a un agente típico, otros a un agente nosocomial y *Corynebacterium diphtheriae*. En un caso no se identificó la etiología. Uno de los pacientes requirió cirugía de reemplazo valvular. Tres pacientes se recuperaron (Child A) y el paciente en categoría Child C falleció. Aunque infrecuente, la asociación cirrosis y endocarditis ocurre en la práctica clínica, se puede asociar a agentes inhabituales y tener manifestaciones encubiertas.

REV MÉD CHIL. 2009 JUN;137(6):729-36.

## **CISTATINA C Y ADIPONECTINA EN PACIENTES DIABÉTICOS TIPO 2 CORONARIOS Y NO CORONARIOS.**

**Wolff C, Durruty P, Espinoza J, Ripamonti S, Díaz J.**

**BACKGROUND:** Patients with type 2 diabetes have a high incidence of coronary artery disease, which is even higher among those with renal failure. A serum level of cystatin C are used to assess renal function and is a potential cardiovascular risk factor. Adiponectin is an anti-atherogenic factor. **AIM:** To measure cystatin C and adiponectin in type 2 diabetic patients with and without coronary artery disease. **MATERIAL AND METHODS:** Nine diabetic patients with coronary artery disease aged 76+/- 10 years, 20 diabetics without coronary artery disease aged 61 +/- 5 years and 20 non diabetic subjects aged 57+/- 10 years, were studied. **RESULTS:** Serum levels of cystatin C (mg/L) were 1.5 (range 0.89-219), 0.81 (range 0.71-1.08) and 0.68 mg/L (range 0.55-0.75) in diabetics with and without coronary artery disease and controls, respectively ( $p <0.0001$ ). No differences in adiponectin between groups and no association between cystatin C and adiponectin, were observed. No association between both parameters and body mass index orglycosilated hemoglobin Ale was observed. Cystatin C had a positive correlation with serum creatinine ( $r =0.57 p <0.001$ ). **CONCLUSIONS:** Diabetics with coronary artery disease have higher levels of cystatin C, that are closely correlated with serum creatinine levels.

## **GENÉTICA**

BIOL RES. 2009;42(4):461-8. EPUB 2010 JAN 29.

## **VEGFA POLYMORPHISMS AND CARDIOVASCULAR ANOMALIES IN 22Q11 MICRODELETION SYNDROME: A CASE-CONTROL AND FAMILY-BASED STUDY.**

Calderón JF, Puga AR, Guzmán ML, Astete CP, Arriaza M, Aracena M, **Aravena T, Sanz P, Repetto GM.**

Microdeletion 22q11 in humans causes velocardiofacial and DiGeorge syndromes. Most patients share a common 3Mb deletion, but the clinical manifestations are very heterogeneous. Congenital heart disease is present in 50-80% of patients and is a significant cause of morbidity and mortality. The phenotypic variability suggests the presence of modifiers. Polymorphisms in the VEGFA gene, coding for the vascular endothelial growth factor A, have been associated with non-syndromic congenital heart disease, as

well as with the presence of cardiovascular anomalies in patients with microdeletion 22q11. We evaluated the association of VEGFA polymorphisms c.-2578C>A (rs699947), c.-1154G>A (rs1570360) and c.-634C>G (rs2010963) with congenital heart disease in Chilean patients with microdeletion 22q11. The study was performed using case-control and family-based association designs. We evaluated 122 patients with microdeletion 22q11 and known anatomy of the heart and great vessels, and their parents. Half the patients had congenital heart disease. We obtained no evidence of association by either method of analysis. Our results provide further evidence of the incomplete penetrance of the cardiovascular phenotype of microdeletion 22q11, but do not support association between VEGFA promoter polymorphisms and the presence of congenital heart disease in Chilean patients with this syndrome.

## HEMATOLOGÍA

REV MÉD CHILE 2009; 137: 552-558.

### LA GENÉTICA COMO FACTOR PRONÓSTICO Y TERAPÉUTICO EN EL MIELOMA MÚLTIPLE.

**Guillermo Conte L,** Esteban Braggi<sup>2</sup>, **Gastón Figueroa,** Rafael Fonseca.

The search for prognostic factors in multiple myeloma has identified the genetic profile of the tumor as the main determinant of patient survival and response to treatment. There is an association between a dismal prognosis and the presence of t(4;14) translocations or 17p deletion, determined by fluorescent in situ hybridization (FISH) or the detection of chromosome 13 deletion using conventional cytogenetic techniques. These alterations define a subpopulation that comprises 25% of patients with a bad prognosis even if they are treated with high dose chemotherapy. These patients should be early derived to more specific therapies. In the other hand, the other 75% of patients without a genetic risk factor, have a higher probability of success with conventional treatment.

## NEFROLOGÍA

BIOL RES. 2009;42(2):189-98. EPUB 2009 AUG 20.

### ANÁLISIS DE LOS POLIMORFISMOS DE NUCLEÓTIDO ÚNICO INTRÓNICAS RS# 466452 DEL GEN DE LA NEFRINA EN PACIENTES CON NEFROPATÍA DIABÉTICA.

González R, Tirado A, Rojas LA, Ossandón FJ, **Alvo M**, Wolff C, Seelenfreund D, Durruty P, Lobos S.

We present the analysis of an intronic polymorphism of the nephrin gene and its relationship to the development of diabetic nephropathy in a study of diabetes type 1 and type 2 patients. The frequency of the single nucleotide polymorphism rs#466452 in the nephrin gene was determined in 231 patients and control subjects. The C/T status of the polymorphism was assessed using restriction enzyme digestions and the nephrin transcript from a kidney biopsy was examined. Association between the polymorphism and clinical parameters was evaluated using multivariate correspondence analysis. A bioinformatics analysis of the single nucleotide polymorphism rs#466452 suggested the appearance of a splicing enhancer sequence in intron 24 of the nephrin gene and a modification of proteins that bind to this sequence. However, no change in the splicing of a nephrin transcript from a renal biopsy was found. No association was found between the polymorphism and diabetes or degree of renal damage in diabetes type 1 or 2 patients. The single nucleotide polymorphism rs#466452 of the nephrin gene seems to be neutral in relation to diabetes and the development of diabetic nephropathy, and does not affect the splicing of a nephrin transcript, in spite of a splicing enhancer site.

## REUMATOLOGÍA

REV MÉD CHILE 2009; 137: 1333-1340.

### ENFERMEDAD DE BEHÇET EN CHILE: ANÁLISIS CLÍNICO DE 44 CASOS.

**Pamela Wurmann, Gonzalo Díaz, Francisca Sabugo, Lilian Soto,** Federica Solanes, Sandra Pino, Guillermo Merino, Juan Ignacio Verdaguer, Francisco Villaruelo, **Miguel Cuchacovich.**

Background: Behget's disease (BD) is a rare multisystemic inflammatory disease that is potentially disabling and may cause death. Aim: To describe the characteristics of BD patients from two Chilean centers. Patients and method: Retrospective review of the clinical records of patients with BD attended in two rheumatology services between 1985 and 2007. The "Behget's Disease Research Committee of Japan" (BDCJ) and the "International Study Group for Behget's Disease" (ISG) diagnostic criteria were applied. Results: We found 44 cases (25 males), diagnosed as BD. The mean age at the onset of symptoms was 26± 12 years. According to BDCJ criteria, 13 patients had complete BD, 24 had incomplete BD and 7 had a suspected BD. Thirty two patients fulfilled the ISG criteria. Forty two patients (95%) had oral ulcers, 33 (75%) had genital ulcers and 29 (66%) had ophthalmological involvement. Eleven and three patients had symptoms of central and peripheral nervous system involvement, respectively. No gender differences were detected. Conclusions: The clinical characteristics of these patients were similar to those described abroad, except for a higher frequency of peripheral nervous system involvement and a lower rate of arthritis.

## OFICINA DE APOYO A LA INVESTIGACIÓN CLÍNICA - OAIC

REV MÉD CHIL. 2009 FEB;137(2):255-8.

### ALTO SUBDIAGNÓSTICO DE SÍNDROME DE PIERNAS INQUIETAS EN LA CONSULTA AMBULATORIA NEUROLÓGICA Y PSIQUIÁTRICA.

Miranda C M, Fabres O L, Contreras S A, **Torres Ch T.**

**BACKGROUND:** Restless legs syndrome (RLS) is a neurological condition that is characterized by the irresistible urge to move the legs and is very common. In the last decade, much attention has been focused on RLS, given its high occurrence, underdiagnosis, and impact on quality of life. **AIM:** To determine the frequency of RLS in a neurologic-psychiatric outpatient clinic. **PATIENTS AND METHODS:** We interviewed patients attending a private neurological outpatient clinic, using a standardized validated questionnaire, and an additional phone interview to confirm diagnosis. **RESULTS:** Of approximately 800 people attending the clinic, the questionnaire was answered by 238 subjects (168 females). Fifteen percent of respondents were affected by RLS and none had been diagnosed before. Most patients had a severe form that probably required treatment. **CONCLUSIONS:** A low awareness of RLS exists in Chile, even among specialized physicians.

REV MÉD CHILE 2009; 137: 1388-1394.

### BENEFICIOS DEL EJERCICIO DE LA AUTONOMÍA Y CONSENTIMIENTO INFORMADO: EJEMPLO DE LOS TESTIGOS DE JEHOVÁ.

Sr. Avelino Retamales P, Dr. **Gonzalo Cardemil H.**

The respect for self-determination has represented a great challenge for the medical community. This debate has resulted in laws, codes of ethics, international treaties, and administrative guidelines, all with the purpose of protecting such right. In the medical field, the "Convention for the Protection of Human Rights and Dignity of the Human Being with Regard to the Application of Biology and Medicine: Convention on Human Rights and Biomedicine", known as the Oviedo Convention plays a crucial role. The doctrine of Informed Consent (IC) exists to enforce it. This principle is considered as law in some countries. In Chile, the IC is considered in the legal ordinance and in documents that are used as guidelines in the field of ethics. Jehovah's Witnesses invoke such precepts when they demand respect for their decisions. The present article outlines their position regarding blood transfusions and their contribution to the practice of bloodless medicine and surgery, which promotes the respect for patient's self determination. The experience of Jehovah's Witnesses has led to a dignified treatment of the patient and has promoted a better participation in decision-making, focusing on the patient. All these benefits can be conveyed to other patients, resulting in the protection of the dignity of the individual.

REV MÉD CHILE 2009; 137: 1482-1487.

### ATROFIA CORTICAL POSTERIOR.

**Carolina Delgado D, Archibaldo Donoso S.**

syndrome, usually due to Alzheimer's disease. The first symptoms are progressive impairment of visuo spatial (Balint's and Gerstmann's syndromes) or visuo perceptive (visual agnosia, alexia) function. Episodic memory and executive function are spared until later stages. We report two males aged 51 and 55 years and three females aged 50, 54 and 56 years, with posterior cortical atrophy. Ophthalmologic study was normal in all. Presenting signs and symptoms were visual ataxia, simultagnosia, agraphia, acalculia, spatial disorientation and unilateral neglect (Balint's and Gerstmann's syndromes). Apperceptive visual agnosia, aphasia, apraxia and alexia were also observed. One female had cortical blindness. Structural images were inconclusive, but PET scan and SPECT disclosed functional impairments in occipitotemporal or occipitoparietal areas.

## UPC

REV MÉD CHIL. 2009 OCT;137(10):1351-6.

### VENTILACIÓN EN POSICIÓN PRONO PROLONGADA COMO ALTERNATIVA EN EL TRATAMIENTO DEL SÍNDROME DE DISTRÉS RESPIRATORIO AGUDO GRAVE POSNEUMONECTOMÍA. CASO CLÍNICO.

**Cornejo R, Romero C, Goñi D, Luengas R, Llanos O, Gálvez R, Castro J.**

Management of patients with severe respiratory failure is mainly supportive, and protective mechanical ventilation is the pivotal treatment. When conventional therapy is insufficient to improve oxygenation without deleterious effects, other strategies should be considered. We report a 53 year-old male who presented a severe respiratory failure refractory to conventional management after pneumonectomy. Prone position ventilation was used for 36 hours. Respiratory variables improved and he did not show hemodynamic instability. He was returned to the supine position without worsening of oxygenation parameters. Extended prone position ventilation could be considered in patients presenting with unresponsive severe respiratory failure after pulmonary resection.

## DEPARTAMENTO DE DERMATOLOGÍA

REV MÉD CHILE 2009; 137: 53-62.

### FACTORES PSICOBIOLÓGICOS EN VITILIGO INFANTIL: POSIBLE ROL EN SU GÉNESIS E IMPACTO EN LA CALIDAD DE VIDA.

**Rodrigo Schwartz, Juan Enrique Sepúlveda, Teresa Quintana.**

**Background:** The exposure to stressing situations may play a role in the appearance of vitiligo. Patients with the disease have a greater sensitivity to environmental stress and a lower threshold to generate catecholamine mediated responses. **Aim:** To evaluate the temperament and character of patients with vitiligo and explore the relationship of the disease with negative life events and life quality impairment. **Material and methods:** The study population were 21 patients with vitiligo aged 5 to 12 years, and two control groups (G1 and G2). G1 was composed by 14 healthy siblings of vitiligo patients. G2 was composed by 21 age and gender matched healthy students from two schools in Santiago, Chile. The Júnior Temperament and Character Inventory (JTCI), the Qualitative Psychosocial Development Survey (QPDS), the Life Event Checklist (LEC) and the Childrens Life Quality index (CDLQI) were applied (LEC only to vitiligo patients). **Resulite:** On the temperament dimensions, vitiligo patients scored high on the "harm avoidance" scale in comparison to G2 (13.7 v/s 10.6). Compared with G1, QPDS showed in vitiligo patients a higher frequency of fear to strangers (71% and 36%, respectively) and a predominant feeling of fear and shyness in response to changes in a close relative (80% and 8%, respectively). There was a negative correlation (protective factor) between the character dimension "self-directedness" and CDLQI score ( $r = -0.703$ ). **Conclusions:** In this group of patients, we found a possible relationship between a specific temperament dimension, vitiligo and its impact on life quality.

REV CHIL INFECT 2009; 26 (3): 248-257.

### ACAROSIS Y ZOONOSIS RELACIONADAS.

Leonor Jofré M, Isabel Noemí H, Patricia Neira O, **Tirza Saavedra U, Cecilia Díaz L.**

Los ácaros de animales (acariasis) pueden transmitirse en forma accidental al hombre al estar en contacto con ellos, produciendo lesiones características (pápulas o vesículas) y ocasionando una dermatitis pruriginosa (acarosis). Existen diversas especies de ácaros, que se encuentran en perros, gatos, roedores, aves y reptiles. Los ácaros hematófagos o los que se alimentan de linfa tienen el potencial de transmitir importantes agentes zoonóticos. La presencia de lesiones sin una etiología clara y el antecedente de contacto con animales, tanto mascotas como animales silvestres, ayudan a plantear esta etiología. El diagnóstico se realiza con la visualización del ácaro, morfología y tipo de hospedero animal. El conocimiento de estas acarosis y el control responsable de mascotas y animales, son las principales medidas de prevención.

## DEPARTAMENTO DE NEUROLOGÍA Y NEUROCIRUGÍA

REV MÉD CHILE 2009; 137: 289-295.

### PERSPECTIVAS EN LA PREVENCIÓN Y TRATAMIENTO FARMACOLÓGICO DE LA ENFERMEDAD DE ALZHEIMER.

**Archibaldo Donoso S, Carolina Delgado D.**

The pharmacological interventions for Alzheimer disease should be based in its pathogenic mechanisms such as amyloidogenesis, tau hyperphosphorylation, disturbances in neurotransmission and changes in neuronal tropism. Other therapies derive from epidemiological observations, such as antioxidants and anti-inflammatory drugs, estrogens, statins and anti hypertensive drugs. Some life style interventions, such as changes in diet, exercise and brain stimulation could also be beneficial for the prevention of Alzheimer disease. Ongoing research on pathogenic mechanisms promises the discovery of more effective therapies. Healthy life style should always be recommended due to its benefit and lack of untoward effects.

REV MÉD CHILE 2009; 137: 900-905.

### DEMENCIA FRONTOTEMPORAL: EXPERIENCIA CLÍNICA.

**Archibaldo Donoso S, Cristián Figueroa V, Rodrigo Gómez R, M. Isabel Behrens P.**

**Background:** Front temporal dementias (FTD) are neurodegenerative disorders characterized by alterations in behavior, affection and language, with relative sparing of episodic memory. There are three major forms of FTD: the frontal or behavioral form, progressive non-fluent aphasia and semantic dementia (that may begin as a fluent progressive aphasia). **Aim:** To report a retrospective clinical experience of patients with frontotemporal dementia. **Material and methods:** Review of 3,700 records of neuropsychological assessments of patients with behavioral disturbances, studied between 1981 and 2008. Of these, 63 patients (59% females) complied with the criteria for frontotemporal dementia. **Results:** There were 47 cases with the frontal variant, four with non-fluent progressive aphasia and six with fluent progressive aphasias (2 evolved to semantic dementia). The mean age of onset was 60±11

years. There were no familiar cases of FTD. Conclusions: It is clinically difficult to diagnose FTD, since evaluation of attitude or language is required. In addition to structural images, functional images were helpful in some cases, but the definitive diagnosis is anatomical.

## CENTRO DE IMAGENOLOGÍA

REV MÉD CHILE 2009; 137: 1602-1606.

### **ROMBOENCEFALITIS POR LISTERIA MONOCYTOGENES. PATOLOGÍA EMERGENTE EN RELACIÓN AL BROTE EPIDÉMICO. PRESENTACIÓN DE TRES CASOS CLÍNICOS.**

**Gonzalo Miranda G, Patricia Orellana P, Holvis Dellien Z, Margarita Switt R.**

An unusual number of cases of rhomb encephalitis have occurred in Chile because of the increased frequency of infections caused by Listeria monocytogenes. We report three females aged 36, 40 and 55 years, with the disease. All presented with a prodrome characterized by headache, nausea, vomiting and fever, followed by ataxia and unilateral palsies of the third, seventh and twelfth cranial nerves. One patient presented also a hemi-hypoesthesia. Magnetic resonance showed lesions in the posterior aspect of the brain stem, specifically in relation to the floor of the fourth ventricle. Blood cultures were positive for Listeria monocytogenes.

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### **FIBROMATOSIS MAMARIA, DIAGNÓSTICO DIFERENCIAL DEL CARCINOMA INVASOR. REPORTE DE UN CASO.**

**M. Lorena Díaz C., Patricia Arancibia H., Leonor Moyano S., Nelson Burgos S.**

Introducción: La fibromatosis o tumor desmoide, es una lesión infrecuente del tejido conectivo, que corresponde al 0,2% de los tumores mamarios. No tiene potencial de dar metástasis pero presenta alta tasa de recidiva local debido a sus márgenes infiltrativos. Su etiología es desconocida. La presentación clínica e imagenológica es casi indistinguible a la de un carcinoma mamario. El diagnóstico es histológico y el tratamiento de elección es la resección quirúrgica. Reporte de caso: Mujer de 31 años que consulta por retracción cutánea en mama derecha. No presenta antecedentes quirúrgicos ni de traumas previos. Se realiza ecografía mamaria que muestra, en relación al sitio de retracción, un nódulo irregular que se extiende hacia la piel. La mamografía muestra discreta retracción de la piel y aumento de densidad del tejido subcutáneo adyacente. Se realiza biopsia percutánea CORE bajo ultrasonido, que muestra proliferación estromal fusocelular, sugerente de un tumor filodes. Posteriormente, se realiza mastectomía parcial cuya histología muestra una proliferación mesenquimatosa de bordes infiltrativos, concordante con fibromatosis mamaria. Conclusión: La fibromatosis mamaria es una entidad poco frecuente y localmente agresiva. Su importancia radica en que las características clínicas, radiológicas y citológicas simulan con frecuencia tumores malignos de la mama. El tratamiento de elección es la cirugía, presentando gran tendencia a la recurrencia, incluso con márgenes libres. Se han descrito poco más de 100 casos en la literatura. Palabras clave: Fibromatosis mamaria, tumor desmoide.

## DEPARTAMENTO DE UROLOGÍA

REV CHIL INFECT 2009; 26 (5): 445-451.

### **ABSCESOS RENALES Y PERI-RENALES: ANÁLISIS DE 44 CASOS.**

**Juan Fullá O., Oscar Storme C, Alberto Fica C\*, M. Antonieta Varas P, José Flores M., Fernando Marchant G. y Daniel Varas F.**

Los abscesos renales son eventos infrecuentes pero potencialmente letales. Objetivo: Conocer sus características clínicas, diagnóstico y evolución. Metodología: Se efectuó un trabajo descriptivo-retrospectivo con los casos detectados entre 1996 y el 2006 en un centro universitario. Resultados: Se identificaron 44 pacientes (edad promedio 49,9 años) asociados en algunos casos a diabetes mellitus (38,6%), litiasis urinaria (36,4%) o infección urinaria previa (11,4%). Los microorganismos más frecuentes fueron Enterobacteriaceae (44,4%) y 33,3% de los cultivos fueron polimicrobianos. *Staphylococcus aureus* se identificó infrecuentemente. La estrategia terapéutica principal fue el uso de técnicas mínimamente invasoras (pigtail, drenaje percutáneo o nefrostomía; 50%), y luego quirúrgicas (nefrectomía o aseos quirúrgicos; ~30%). Sólo 20,5% fue tratado exclusivamente con antimicrobianos. Los procedimientos mínimamente invasores se usaron en forma progresiva después del 2001 ( $p < 0,005$ ). La letalidad en esta serie fue 4,5% ( $n = 2$ ) y 13,6% ( $n = 6$ ) desarrolló shock séptico. La nefrectomía se aplicó en 9 casos (20,5%). Los pacientes seleccionados para nefrostomía tuvieron menos riesgo de ingresar a UCI (Odds Ratio 0,083 IC95 0,008-0,911). Conclusiones: Los abscesos renales son causa de morbilidad mayor aunque de baja letalidad. Su estrategia terapéutica ha ido cambiando en los últimos años a favor de procedimientos mínimamente invasores como los drenajes percutáneos y/o endoscópicos.