

# Abstracts presentados en congresos internacionales 2007

Por decisión del Comité Editorial de la Revista HCUCh, se publican en esta oportunidad solamente los abstracts de los académicos que facilitaron la información a la Revista y que correspondan a congresos mundiales, europeos y norteamericanos.

## CLÍNICA PSIQUIÁTRICA

### **XIII INTERNATIONAL CONGRESS ESCAP (EUROPEAN SOCIETY FOR CHILD AND ADOLESCENT PSYCHIATRY) - FLORENCIA, ITALIA.**

PSYCHOPATHOLOGY IN CHILDREN OF DEPRESSED MOTHERS IN SANTIAGO, CHILE.

María Elena Montt, Fritsch Rosemarie.

**OBJECTIVES:** to assess psychopathology and social functioning in children of currently depressed mothers. **METHOD:** A two-stage screening process was used to identify depressed mothers with children aged 6-16 years. Maternal Assessments included the mini international neuropsychiatric interview and the Hamilton depression rating scale. Child psychopathology was assessed with the Child Behavior Checklist (CBCL). **RESULTS:** The final sample consisted of 290 mother-child pairs. 49.8% (95%CI: 43.9-55.7) of children had overall CBCL psychopathology scores in clinical range. Internalizing symptoms were more prevalent than externalizing symptoms (62.2% [95%CI: 56.3-67.8] vs. 35.7% [95%CI: 30.2-41.5];  $p = 0.0000$ ). Overall CBCL psychopathology, internalizing and externalizing psychopathology scores were highly correlated with maternal HRDS scores. In several models that controlled for multiple potential confounders were significantly associated with mother's HDRS score ( $p = 0.029$ ); overall CBCL psychopathology were significantly correlated to children's self reported anxiety. **CONCLUSIONS:** A large proportion of children of depressed poor mothers attending primary care clinics in Chile had psychopathological symptom scores in the clinical range, with a predominance of internalizing symptomatology. These results are similar to those previously reported in the US. These children may benefit from psychiatric evaluations and many are in need of clinical and social services.

### **15<sup>th</sup> EUROPEAN CONGRESS OF PSYCHIATRY - MADRID, ESPAÑA.**

TREATING POSTPARTUM DEPRESSION IN PRIMARY CARE IN SANTIAGO, CHILE.

Rojas G, Fritsch R, Solis J, Jadresic E, Araya R.

We compared the effectiveness of a multi-component intervention with usual care to treat postnatal depression among low-income mothers in primary care clinics in Santiago, Chile. **Methods:** Randomised controlled trial. Two hundred and thirty mothers with major depression attending primary care clinics were randomly allocated to either a multi-component intervention or usual care. The multi-component intervention involved a psychoeducational group, systematic monitoring and treatment compliance support, and pharmacotherapy if needed. Data were analysed on an intention-to-treat basis. The main outcome measure was the Edinburgh Postnatal Depression Scale (EPDS) at 3 and 6 months post randomisation. **Results:** Approximately 90% of randomised women completed assessments. There was a marked difference in all outcome measures at 3 months, in favour of the multi-component intervention. However, these differences between groups decreased after 3 months. In our primary analysis, the adjusted difference in mean EPDS between the two groups at 3 months was -4.5, 95% C.I. -6.3 to -2.7,  $p < 0.001$ . There was a sharp decline in the proportion of women on antidepressants after 3 months in both groups. **Conclusions:** This intervention

considerably improved the outcome of depressed low-income mothers compared to usual care for the first 3 months. However, some of these clinical gains were not maintained thereafter, most likely because a large proportion stopped taking medication. Further refinements to this intervention are needed to ensure treatment compliance after the acute phase.

## DERMATOLOGÍA

### 21<sup>st</sup> WORLD CONGRESS OF DERMATOLOGY - BUENOS AIRES, ARGENTINA.

#### INNOVATIVE CANCER VACCINES.

Honeyman J.

Immune effectors can play a significant role in controlling tumor growth, either in natural conditions or in response to therapeutic manipulation, of the cascade of molecular events leading to tumor rejection by the immune system. The vaccines attempt to teach the immune system to recognize antigens that escaped the immunologic surveillance and are by it, therefore able to survive and, in time, disseminate. Recent tumor immunology advancements might change the way to design the next generation of cancer vaccines, hopefully improving the effectiveness of this therapeutic approach. Dendritic cells are the most potent antigen-presenting cells for initiating cellular immune responses. Dendritic cells are attractive immunoregulatory cells for cancer immunotherapy. Several research groups are developing cancer vaccines. They use different technologies, targeting different antigens, combining different carriers and adjuvants, and using different immunization schedules. Most of the vaccines are still experimental and not approved by the US or European Regulatory Agencies. The experience of activating dendritic cells with different new modalities specially oriented to the management of skin cancer, specially malignant melanoma is analyzed. A new tool is the chemoprevention with potential agents as the lipid-lowering drugs and the statins. The use of vaccines and chemoprevention in cutaneous melanoma can become a valid strategy of managing this cutaneous cancer.

#### A CASE OF VERTIGO ASSOCIATED WITH CUTANEOUS LESIONS.

M Gaete, M Recavarren, P Calderón, J Honeyman.

A 44 year-old woman with chronic arterial hypertension presented with a three months history of vertigo. The episodes were exacerbated by postural changes and interfered with her normal activities. She reported for the same time, asymptomatic papules on her extremities, trunk and face. From the data obtained by direct interrogation, she referred similar lesions on her lower extremities, two years ago, that had disappeared spontaneously. Physical examination revealed multiple indurate purple and reddish-brown papules and nodules distributed along her lower extremities, trunk and periorbital area. Histopathological examination showed non-caseating granulomatous inflammation without necrosis, negative for the Ziehl-Nielsen staining for acid-fast bacilli. A brain magnetic resonance imaging (MRI) revealed leptomeningeal infiltration associated with parenchymal nodular lesions in the cerebellum and involvement of the sixth, seventh and eighth brain nerves. Chest computed tomography showed mediastinal and bilateral hilar lymphadenopathy and diffuse small nodules in bilateral lung fields. The tuberculin reaction was negative. The clinical correlation of pulmonary, brain MRI findings and the skin biopsy oriented the diagnosis of systemic sarcoidosis. Laboratory test ruled out neoplastic and infectious etiology. After 2 months of steroid therapy, symptoms improved with partial regression of the cutaneous lesions and remission of vertigo. Sarcoidosis is a chronic, granulomatous disease with involvement of multiple organs, characterized by a coexistence of cutaneous anergy associated to a hypersensitivity reaction in the sarcoid lesions<sup>1</sup>. Systemic sarcoidosis usually affects lungs, eyes, lymphatic nodes and skin. Neurosarcoidosis occurs infrequently, in 5 – 15% of cases<sup>2</sup>. In the present case vertigo was related to neurological sarcoidosis. In conclusion, although neurosarcoidosis is very rare in Chile, it should be considered in patients with suggestive clinical manifestations, compatible imaging findings and the histological evidence of granulomatous organ involvement.

#### IMMUNOPATHOLOGY OF DERMATOSES OF PREGNANCY.

JF Honeyman, M Gosch, J Larrondo, R Cárdenas, S Zapata.

During pregnancy, several dermatoses may occur, and in some cases the initial diagnosis is difficult. Among this diseases can be mentioned pruritus gravidarum with skin lesions, polymorphic eruption of pregnancy (PUPP), urticaria of pregnancy, pemphigoid of pregnancy and a rare variety of pemphigus of pregnancy. Some of these diseases have immunological mechanisms that are involved in the production of the skin lesions. The immunopathological studies show different patterns

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that help to the diagnosis of the different varieties of these skin disorders. We studied with the immunofluorescence techniques biopsies of 43 cases of dermatoses of pregnancy. Fluorescent labeled antibodies for IgG, IgM, IgA, complement C3, C4 and fibrinogen were performed in all the cases. The clinical diagnosis was Pemphigoid gestationis, PUPP and Pemphigus of pregnancy. Two cases of neonatal pemphigoid and one of neonatal pemphigus were found. The different clinical and immunological features observed are presented.

### AGE, SEX AND CLINICAL FORMS OF ONYCHOMYCOSIS: A FIVE-YEAR SURVEY.

R De La Parra, J Larrondo, M Gosch, S Zapata, JF Honeyman.

Background: This study presents the etiological agents of onychomycosis and analysis its distribution in 2877 patients of different age, gender and involved sites. Patients and Methods: Nail specimens were collected from patients with suspected clinical onychomycosis attending the Dermatology Department from 1998 to 2002. Both direct microscopy and cultures of the nail material were performed to confirm the diagnosis. Fungal cultures were observed prospectively and analyzed to identify the causative agent. Results: A total of 1031 patients with cultures, resulted positive (36%). Dermatophytes were isolated in 725 cases (70%), and yeast in 306 (30%). There was a significant toenail onychomycosis ( $p < 0.01$ ). The most common agent was *T. rubrum*, followed by *Candida* sp and *T. mentagrophytes*. There is a predilection of toe nails infections on men and finger nails in women. ( $p < 0.0001$ ). The incidence of infection increase with age. Conclusions: Clinicians should consider that pathogens of onychomycosis may vary in different patients according age, gender and clinical forms, to choose optimal antifungal agents accordingly.

### PEMPHIGUS FOLIACEOUS: AN ATYPICAL PRESENTATION.

C Ramírez, P Calderón, P Segura, J Szot.

An 80 year old female patient, with past medical history of diabetes and hypertension, presented to us with a 20 years history of rosacea, using topical corticosteroids, intermittently. She complained of erythematous and scaling plaques on her legs since 3 months and also severe facial burning erythema and crusting developed few days before. She was recently treated with terbinafine 250 mg daily for 2 months for tinea corporis and the face rash developed during the last week of treatment. On physical examination she had a severe facial erythema, with hematic crusts that involved the entire facial surface. On her lower extremities there were annular, erythematous, well defined plaques with mild desquamation. Small vesicles were found on the borders of some of these lesions. Acute generalized exanthematic pustulosis v/s facial pyoderma and figurated erythema were suspected, and the patient was admitted to our hospital. Within laboratory examination, the abnormal parameters were: white blood cells count of 10.300, 18% of eosinophils and ESR of 108; alkaline phosphatase of 349 mg/dl, gamma glutamyl transferase of 284 mg/dl and creatinine of 1.7 mg/dl. The patient was started on ceftriaxone and clindamycine and topical sterile petrolatum for the face lesions, with mild response. A skin biopsy from the lower extremities lesions was performed, which showed presence of superficial blisters with fibrin, neutrophils and acantholytic keratinocytes, with spongiosis of the adjacent epidermis. Within the dermis there was a mixed inflammatory infiltrate, consisting mostly of lymphocytes and eosinophils. Direct immunofluorescence showed intercellular deposits of IgG in the superficial epidermis; all these findings being consistent with pemphigus foliaceus. This case is presented because of its unusual presentation leading to a difficulty in the diagnosis.

### CONJUNCTIVAL MALIGNANT MELANOMA: ONE CASE REPORT.

P Calderón, P Segura, J Szot.

A 68 years old caucasian male, who worked as a farmer in Northern Chile (Vicuña, 30° 12' South - 69° 51' West) consulted for a 5 years old asymptomatic pigmented lesion on his left eye. He was otherwise healthy. His physical examination, showed a brown-black 10 mm exophytic tumor, affecting bulbar and palpebral conjunctiva of his left eye. There was no evidence of corneal, orbital or regional lymph node involvement. Blood and urinary work-up were normal. Encephalic MRI showed a tumor compromising the left eye's medial canthus. Additional study excluded other organs tumoral infiltration or metastasis. Histopathology showed prominent nesting of atypical melanocytes with epithelioid cells showing pleomorphic nuclei, prominent nucleoli, atypical mitoses, and abundant cytoplasm. The tumor thickness was 800µ. Necrosis and focal ulceration was present. Complete removal of the eye globe was indicated, but he refused it, not ever coming back for follow up. Conjunctival melanoma is a condition of concern because of its rarity and lethal potential. Also, a number of clinical entities should be considered in its differential diagnosis: e.g.: conjunctival nevi, melanosis, oculodermal melanocytosis, papilloma and squamous cell carcinoma. Dermatologists should always assess the eye conjunctiva and follow closely pigmented suspicious lesions and consider early referral to ophthalmologist for biopsy in growing or atypical lesions.

## CUTANEOUS VASCULITIS AND HYPOCOMPLEMENTAEMIA IN PRIMARY SJÖGREN SYNDROME.

J Roa, P Calderón.

We present a 50-year-old woman who was treated in our clinic in May 2005 for cellulitis of her left low extremity. In March 2006 she was started with terbinafine treatment for Onicomycosis, within 30 days of therapy she developed a purpuric papular erythematosus rash on her lower extremities, interpreted as an allergic drug reaction; terbinafine was stopped. The rash cleared during the next few days. In May 2006 she presented the same rash on her lower extremities that was not drug related. Prednisone (20 mg/day) was started for 3 months, with excellent clinical response. Punch biopsy of the affected skin was compatible with leukocytoclastic vasculitis (LCV) with eosinophilic infiltrate. The direct immunofluorescence study of lesion skin biopsy showed granular deposits of immunoglobulin M (IgM), C3, C4 and fibrinogen within the walls of the dermal vessels as well as granular deposits of IgM in the basal membrane. The erythrocyte sedimentation rate was 46 mm/h; high titer of antinuclear antibody (ANA) (homogeneous type) at 1 : 160 (normal < 1 : 40), as well as the rheumatoid factor (RF) 8192 (normal < 8); the complement level of C3 was normal and C4 was low 1.94 (16-44). Anti-RNP, anti-Sm, anti-SS-A(Ro), and anti-SS-B(Ro), antibodies for antineutrophil cytoplasmic antibody (ANCA), Venereal Disease Research Laboratory (VDRL) test, hepatitis C virus and antistreptolysin antibody were negative. Biochemical analysis of serum, complete blood cell count and urine were within normal limits. Because ANA and RF were positive, we asked the patient if she had experienced a dry sensation in her eyes and mouth, or any other rheumatologic disease symptom related. As she reported a dry sensation of both eyes and mouth, we referred her to the rheumatology clinic. The Schirmer's test was positive. Lip biopsy revealed the accumulation of numerous lymphocytes in the salivary gland. Thus, our patient fulfilled four criteria of the European multicenter study, suggesting a definitive diagnosis of Sjögren's syndrome (SS). Currently, her treatment includes prednisone (20 mg/day) and hydroxicloroquine 50 mg BID. We present the case as an uncommon association of these 2 entities, with important prognostic implications, which requires a close follow up, since in the future this patients may develop Systemic Lupus Erythematosus or other connective tissue diseases. Furthermore, hypocomplementaemia is closely associated with systemic expression and adverse outcomes (lymphoma development and death) in patients with primary SS.

## HIV POSITIVE PATIENTS BEARING ANOGENITAL CONDYLOMA: A CLINICAL, VIROLOGICAL AND HISTOPATHOLOGICAL ASSESSMENT OF THE LESIONS.

Martínez M, Giacaman P, Chnaiderman J, Ramis C, Santander E, Garmendia M.

Condylomata acuminata is one of the most frequently sexually transmitted diseases (STD) in Chile. When these lesions appear in human immunodeficiency virus (HIV) infected individuals, they adopt particular characteristics that make them more prone to progress to neoplastic lesions. Objective: The aim of this study was to determine whether HIV infected subjects exhibit more risk factors associated with the onset of cancer than seronegative people. We also aimed to study if higher frequency of oncogenic genotypes could be isolated from condylomata acuminata of HIV infected than HIV negative patients, and to evaluate if seropositive patients presented more neoplastic lesions as compared to seronegative controls. Material and methods: 89 subjects with condylomata acuminata, 47 HIV infected and 42 HIV negative, participated in the study. Incisional biopsies were performed in all of them, followed by histopathologic analysis and PCR to detect human papillomavirus (HPV) DNA. The clinical charts and a questionnaire, that each participant was invited to fill, were used to assess epidemiological variables and previously reported risk factors associated with cancer. Results: Several risk factors were found to be different in HIV infected patients than in the HIV negative controls. The most frequently identified were; the gender male, homo or bisexual, promiscuity, drug use, anal intercourse, recurrent condyloma, perianal localization of the condyloma and history of having more than 1 STD. Likewise, significantly different frequencies of HPV oncogenic genotypes ( $P < 0.05$ ) were found in 19.1% and 2.38% of the HIV positive individuals and HIV negative patients, respectively. 5 subjects showed intraepithelial neoplasia, 4 of them were HIV infected individuals. Furthermore, an association was found between the detection of oncogenic genotypes and the occurrence of anogenital cancer (OR=16.5). Conclusion: HIV infected patients exhibit more risk behaviors for the onset of anogenital cancer. In these patients, more oncogenic HPV genotypes were isolated, as well as a higher number of neoplastic lesion in their condylomata acuminata. We found that the presence of HPV oncogenic genotypes in the anogenital warts is associated with a higher likelihood of having intraepithelial neoplasia.

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### YOUNGER PEOPLE HAVE MORE SCABIES: A FIVE-YEAR SURVEY.

Saavedra T, Larrondo J, Gosch M, Zapata S, Honeyman JF.

Background: Epidemiological features of scabies of 745 patients attending the Dermatology Department from 1998 to 2002, with positive acarotest were evaluated. The distribution of patients of different ages and sex. Results were observed and analyzed retrospectively Results: Among the positive cases, 362 were male (49%) and 383 female (51%). Mean age of the patients was  $25 \pm 24$  with a range of 0-97. The disease was more frequent in younger patients ( $p < 0,0001$ ). Patients less than 19 y-old 54% of cases, 20-59 y-old 35% and over 60 y-old 11%.

### PARANEOPLASTIC AMYOPATHIC DERMATOMYOSITIS.

Brant M, Saavedra T, Brant S, Santander R, Valenzuela C, Prado B.

Objectives: analysis of a case of amyopathic dermatomyositis associated with occult neoplasia. Methods: description of clinical findings, laboratory, imaging and histopathology of paraneoplastic amyopathic dermatomyositis, found in a clinical case at Hospital Clínico Universidad de Chile, Santiago, Chile, along with a bibliographical revision of the topic. Results: a 53-year-old woman presented with a 3 years history of pruritic, erythematous eruption affecting her hands, knees, upper back and occipital region. Physical examination was significant for erythematous-violaceous papulae affecting metacarpophalangeal joints of both hands, as well as posterior neck, knees and hips. Classic peringual erythema and telangiectasies of the proximal nailfolds were also present. Proximal muscle strength was conserved. Laboratory examination revealed a normal complete blood count, and creatin kinase of 108 U/L. Antinuclear antibodies, extractable nuclear antibodies, C3, and C4 were either negative or normal. Antitumoral marker CA-125 was elevated (2260 U/mL). Transvaginal ultrasound revealed a right adnexal mass with intraperitoneal fluid in the pouch of Douglas. A biopsy specimen from the hand lesions resulted compatible with dermatomyositis. Dermatomyositis is an idiopathic vascular disorder characterized by inflammatory muscular and cutaneous disease. Clinically, it manifests with proximal muscle weakness and characteristic skin lesions, including peringual telangiectasies and poikilodermatous, violaceous macules distributed in a shawl distribution over the shoulders, upper back and forearms. Histopathological evaluation of skin lesions reveals perivascular lymphocytic infiltrate, increased dermal edema and mucin deposition. Dermatomyositis has a 25% risk of association with underlying neoplasia in adult patients, and it may increase up to 50% in patients with amyopathic dermatomyositis. Amyopathic dermatomyositis is a clinical subtype of dermatomyositis, which refers to patients exhibiting the classic cutaneous manifestations of dermatomyositis but without muscle weakness or laboratory evidence of muscle involvement. The neoplasias most commonly associated with amyopathic dermatomyositis are ovarian, breast, lung, stomach and urinary tract cancer. Conclusions: the clinical history describes a very uncommon case, but described in the literature nonetheless, where dermatological findings allow the diagnosis of an occult neoplasia.

### NON CUTANEOUS MANIFESTATIONS OF KAPOSI SARCOMA: ONE CASE REPORT.

Szot J, Segura P, Calderón P, Saavedra T.

A 32 years old homosexual Hispanic man, consulted for 2 months fever, weakness and weight loss of 10 kg. On the physical examination he was afebrile, with mucocutaneous jaundice, painless swelling of neck lymph nodes, and multiple dark red-purple tumors 0.5-1 cm thick, located in his nose, neck, trunk, forearms and thighs. Laboratory findings included: Hemoglobin 11.6 g/dL, white cell count 6.1 K/uL, 45% lymphocytes, ESR: 40. Total bilirubin: 10.8 mg/dL, conjugated bilirubin: 9.8 mg/dL. AP: 963 mg/dL, SGGT: 417 mg/dL, SGPT: 134 mg/dL. 100% prothrombin. CRP: 32. HIV testing: positive. Viral load: 352,000 eq/mL. CD4 counts: 158. PPD, VDRL, HBsAg and Hepatitis C antibodies: negative. Urine sample was normal. Chest CT scan showed mediastinic and axillary lymph nodes enlargement. Abdominal scan showed: liver and spleen enlarged and extra/intra-hepatic bile duct dilatation without stones, retroperitoneal, mesenteric and inguinal lymph nodes positives. Upper gastrointestinal endoscopy demonstrated infiltrated red-purple spots distributed along the duodenum tract. A biopsy specimen was collected which revealed spindle cells and capillary proliferation with atypia. There were some inflammatory cells and erythrocytes surrounding the lesions. These findings were consistent with Kaposi's sarcoma (KS) of duodenum. Cutaneous biopsy of lesions was also compatible with KS. HAART (AZT + 3TC + lopinavir and ritonavir) was initiated. Also, papillotomy and 7F-12 cm stent insertion into the biliary tract was placed. All parameters of obstructive jaundice were improved. Patient is currently followed. Because of increasing in the incidence and prevalence of HIV, this report aimed to alert to Dermatologist community regarding non cutaneous manifestations of SK.

## LIFE QUALITY AND CLINICAL ANALYSIS OF PSORIATIC PATIENTS IN A CHILEAN HOSPITAL.

Valenzuela FA, Silva PL, Valdés MP, Saavedra DE, Villalba V, Brant M, Brant S, Figueroa A, Roa, Leiva J.

Psoriasis is a prevalent chronic inflammatory disease of partially known etiology that can compromise large cutaneous areas with or without joint involvement. There are no Chilean studies about the impact of psoriasis on patients' quality of life. Objective: to investigate the clinical profile of psoriatic patients in control at the Clinical Hospital of the University of Chile, their present psoriatic episode, remote history, joint involvement and impact of psoriasis on patients' quality of life. Patients and Method: a voluntary and confidential survey was applied to psoriatic patients controlled at the Dermatology Department from September 2006 to January 2007. The survey consisted of 2 parts, the first one related to the clinical aspects of the present episode and remote history. The second one referring to the impact of the disease on quality of life during the previous 7 days by means of 10 questions from the Dermatology Life Quality Index (DLQI), related to work, leisure, daily activities and personal relationships, among others. All surveys were applied by dermatology residents. Results: 43 women and 40 men were interviewed (n: 83), the average age was 42.5. 33.6% of the patients had a relative with psoriasis. Psoriasis vulgaris was the predominant type (66%), followed by guttata (15.7%), inverse and palmoplantar (4.8% each), one single patient displayed pustular psoriasis. 27% had less than 10% of body surface involvement and approximately 10% had 50% or more. Trunk and lower extremities were the most frequently affected corporal segments, followed by superior extremities and scalp. Among therapies, 86.8% of patients used lubrication, 66.3% topical corticosteroids, 20.5% methotrexate, and 10.8% phototherapy. Acitretin and cyclosporin were used only by 1 patient each, and no one received biological agents. Of the 24 patients who had joint complaint, only 9 had been studied and catalogued as psoriatic arthritis, mainly asymmetric oligoarticular involvement. Our results show stress as the most important triggering factor of psoriatic exacerbation (67.5%), followed by treatment suspension (28.9%) and infections (20.5%). On relation to quality of life, globally more than 40% of the interviewed patients had a marked alteration ("very much" or "moderately") in the aspects investigated. The most frequently complaint was pruritus, pain or stinging sensation (61.4%), embarrassed or self-consciousness (55.4%), influence on the clothes they wore and on their social or leisure activities (over 40% each). Psoriasis affected slightly their work and sexual life (20%). 32.5% of patients answered that treatment interfered a lot with their quality of life. Conclusions: the clinical aspects of this sample of psoriatic patients is concordant with literature. Because of the descriptive nature of this study, we can't make "cause-effect" conclusions. We verify that psoriasis is a disease of great impact on patients' quality of life, an important aspect to consider for integral management.

## FOREIGN BODY GRANULOMATOUS REACTION ASSOCIATED TO SQUAMOUS CELL CARCINOMA.

Saavedra D, Schwartz R, Donoso MT.

A 82-year-old woman presented with a 1-month history of a tumour on her left cheek. The patient referred that this lesion appeared few days following an accidental facial trauma. He was otherwise healthy without any other underlying medical conditions. Physical examination revealed a 3 cms tumour, with a superficial ulcer and tender to palpation. Considering the rapid growth of the lesion our initial clinical diagnosis was a foreign body granulomatous reaction (FBGR) and a shave biopsy was performed. Histopathologic sections revealed a well differentiated squamous cell carcinoma (SCC) with a desmoplastic stroma, and an inflammatory infiltrate composed by polymorphonuclear cells, histiocytes, lymphocytes and giant multinucleated cells. The latter findings were consistent with a FBGR. FBGR is an inflammatory reaction against high molecular weight, inorganic and organic material within the skin. It can be acquired accidental, self-inflicted, secondary to surgery or in relation to topical medications. There are very few reports on SCC in association to FBGR, and most of them are related to intralesional bleomycin or radiotherapy. There are no previous reports showing an association between SCC, FBGR and local physical trauma as we observed in our patient. A brief review of the possible role of giant multinucleated cells within a SCC is discussed.

## ELEPHANTIASIS VERRUCOSA NOSTRA.

Brant M, Saavedra T, Brant S, Santander R, Valenzuela C, Prado B.

Objectives: analysis of a case of elephantiasis verrucosa nostra with various cutaneous manifestations. Methods: description of dermatological clinical findings in a patient with elephantiasis verrucosa nostra of the lower extremities, found in a clinical case at Hospital Clínico Universidad de Chile, Santiago, Chile, along with a brief bibliographical revision of the topic. Results: a 57-year-old man presented with a history of bilateral chronic lymphedema secondary to congenital lymphatic atrophy, beginning at 10 years of age. He received surgical treatment in two opportunities with parcial resolution in Chile and Brazil. Currently under dermatologic control for a recent case of cellulitis, skinfold fissures and verrucous lesions, wich were biopsied to rule out carcinoma (lymphangiosarcoma). Physical examination revealed skin hyperpigmentation as well as significant edema of

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the lower extremities, and verrucous lesions on skinfold areas, along with skin fissures and some isolated dermatofibromas and millium cysts. Chronic lymphedema is the accumulation of lymph on the subcutaneous tissue which leads to chronic inflammation and secondary scarring, and it is commonly associated with complications such as bacterial infections, and less frequently with lymphangiosarcoma, an extremely rare but lethal malignant tumor. Elephantiasis verrucosa nostra is a well-known chronic lymphedema complication that appears about 10 years after the onset of the chronic lymphedema, with progressive trophic changes on the skin similar to those described on the patient above. Elephantiasis verrucosa nostra often ends with the amputation of the affected extremities. Conclusions: the clinical history describes an uncommon case, with the classical dermatological manifestations. Regular medical controls are important for an early diagnosis and adequate management of the numerous complications of this disorder.

### ADULT HENOCH-SCHÖNLEIN PURPURA, ITS TREATMENT AND COMPLICATIONS CLINICAL CASE.

Valenzuela FA, Saavedra T, Honeyman J.

A 34-year-old man consults for symmetrical palpable purpura on lower limbs, thighs and lower abdomen in addition to sneezing, articular pain in ankles, wrists, knees and elbows, and muscular and abdominal pain. In his remote history: hepatitis during childhood. He received anti-flu vaccine 3 weeks before skin manifestations onset, and Ketoprofen a couple of days before. Direct immunofluorescence from the left foot dorsal skin showed an IgA granulate deposit in vessel walls concordant with Anaphylactoid purpura. Blood analysis including ANA and HIV were normal, Urinary Sediment never showed proteinuria and there was no evidence of malignancy. Treatment regimen began with absolute rest and 0.8 mg/kg/d prednisone per 5 days, and then 0.6 mg/kg/d per three weeks. By the second week of treatment, liver enzymes were transiently mildly elevated. He completed three months with oral corticosteroids and rest. By the fourth month, the patient began with bilateral hip pain especially in activity and purpura reappeared with less intensity. Pentoxifylline 400 bid and hyperbaric camera sessions were indicated, but the hip compromise grew. MRI indicated bilateral avascular necrosis of femoral heads, which was appropriately repaired with osteosynthesis. After seven months, skin lesions still appear on lower limbs, with a minimal intensity. Henoch-Schönlein purpura (HSP) is an Ig A-mediated small-vessel vasculitis that largely affects children but also is seen in adults. Many antigens have been involved in the pathogenesis including microorganisms, drugs and neoplasm, but the exact mechanism is unclear. Clinical manifestations include palpable purpura, arthralgia or arthritis, abdominal pain, gastrointestinal bleeding, and nephritis. Primary skin lesion may begin as erythematous macular or urticarial lesions, progressing to blanching papules, and later, to palpable purpura, usually 2-10 millimeters in diameter. The lesions are typically symmetric and tend to be distributed in dependent body areas, such as ankles and lower legs. Recurrences occur in the same sites as prior lesions. The most serious long-term complication is progressive renal failure, fortunately infrequent. HSP is more common in males with a rate of 1.5-2:1. Only some studies have tried to determine the association between adult HSP and malignancy, particularly in men older than 40 years who develop this disease without a precipitating factor. The treatment is based in corticosteroids. In the present case, patient presented an avascular bi-femoral head necrosis secondary to this drugs, not because of an overdose, thus he may be susceptible to this complication on therapeutical doses. His conditions has rapidly improved. The transient increase on liver enzymes was considered as an uncommon sign of vasculitis. We have made a close follow-up case to detect non-evident cancer or connective tissues disease, without positive findings.

### FREQUENCY OF SKIN DISEASES IN DOWN SYNDROME'S PATIENT IN SANTIAGO, CHILE.

Aranibar L, Villagrán B, Merino D, Hernández E, Espinoza M, Honeyman J.

Introduction: down syndrome (DS) is associated frequently with several non defined specific dermatologic conditions. The more frequently seen are xerosis, Folliculitis, alopecia areata and millia. Purpose: evaluate frequency of dermatoses in a children population with DS and compare this without this syndrome. Methodology: 252 cases with DS were compared with the control group. Statistical analysis was performed with statigraph like medium and a confidence interval of 0.5%. To define the difference between this two populations it was used the bivariate analysis with the Fisher test with the significant p value 0.05, using the Stata 9.0 program. Results: the more frequent skin disease are: keratosis pilaris 52% ( $p < 0.0001$ ); xerosis 39% ( $p < 0.0001$ ); seborreic dermatitis 29% ( $p < 0.0001$ ); atopic dermatitis 22% ( $p < 0.0001$ ); onychomycosis 21% ( $p < 0.0001$ ); cheilitis 19% ( $p < 0.0001$ ); tinea pedis 13% (0.002); syringoma 12% (0.0001); acne 10% ( $p < 0.02$ ); Folliculitis 10% (0.0001); fissured tongue 8% (0.0001) and diaper dermatitis 6% ( $p < 0.02$ ). There are no difference between both groups in the frequency of nevus 11% ( $p = 0.2$ ) and cafe au lait spot 3% ( $p = 0.6$ ). Conclusion: patient with DS have higher frequency of several skin diseases that the dermatologist should consider to have better management.

## EPIDERMOMYCOSES IN CHILDREN.

Merino D, Gosch M, Larrondo J, Zapata S, Honeyman JF, De La Parra R.

A survey of the etiological agents of epidermomycoses in 1936 children (0-12 years-old) is presented. The distribution according age, sex and involved sites is studied. Samples for fungal cultures were obtained from children with suspected clinical epidermomycoses attending the Dermatology Department from 1998 to 2006. Results: 615 cultures resulted positive (32%). The most frequent clinical forms (66%) were tinea unguium and tinea capitis. The most common causative agents (80%) were *M.canis* and *T. rubrum*. Etiological fungi are different according to the site of infection. ( $p<0.0001$ ). Female have more frequently *Candida* sp infection and male *T.rubrum* infection ( $p<0.05$ ). *M.canis* is more frequently founded in children less than 6y-old and *T. rubrum* predominates in older children. Conclusions: dermatophytes were responsible for most of the infections. Involved sites, gender and age appear to be associated to certain etiological agents in epidermomycoses.

## HERPES SIMPLEX VIRUS TYPE 2 PREVALENCE IN HIV+ PATIENTS OF THE NORTH AREA OF SANTIAGO, CHILE.

Luzoro A, Martínez MJ, Santander E, Gubelin W, Afani A.

HIV infection stands as a mayor issue in public health with a beyond-national-frontiers relevance. Actually is considered a chronic infection with a continuous worldwide rising in prevalence. This fact makes urgent to know and prevent conditions that help its transmission. Herpes Simplex Virus type 2 (HSV-2) is one of the most frequent opportunist infections in HIV+ patients. It is also the first cause of genital's ulcers worldwide. This constitutes a mayor epidemiologic concern since it is well documented that genital's ulcers increase the chances of infection by HIV and makes worse the course of the disease. Given the significance of measuring HIV & HSV-2 co-infection, we undertake a study to determine its incidence in a population representing a large area in Santiago, Chile. Objectives: to determine the incidence of HSV-2 infection in HIV+ patients in the North area of Santiago (Chile). To assess the relation between risky history and behaviors to the infection by HSV-2. Methods: a cohort study was made in 400 patients who were under control in San Jose's Hospital, Santiago, Chile. A socio-demographic and sexual behaviors questionnaire along with a blood sample was applied. HSV-2 infection was assessed using ELISA technique. This procedure was repeated six month and a year after initial trial. Preliminary results, corresponding to the first sampling are shown here. Results: In the first sample, 77% of HIV+ subjects test positive for HSV-2. Significance differences were found between HIV & HSV-2 co-infection vs HSV-2 negative, in marital status (67% single vs 52% in HSV-2 negative), sexual orientation (65% homosexual vs 37% in HSV-2 negative) and number of sexual partners (mean 37 vs 27). Discussion: the high prevalence of HSV-2 infection among HIV+ is in agreement with previous studies. The same stands for risk factors known for this co-infection. These results reinforce the idea that HIV infection is a worldwide concern and that general guidelines and education should reduce HSV-2 co-infection's rate and improve HIV+ patient's life quality.

## BULLOSIS DIABETICORUM AS DIABETES MELLITUS TYPE 2 PRESENTATION.

Luzoro A, Cárdenas R, Carreño L, Calderón P.

We present a 47 year old woman, with no previous medical history, who consulted for one-week history of pain and edema on her distal left leg. On physical examination, she presented a warm and well-defined red plaque with confluent blisters on its surface containing clear fluid. Initially diagnosed as Erysipela, she was started on 12 MU/day Penicillin treatment. Laboratory study revealed glicemia 213 mg/dL. Since no improvement was noted on the third day of therapy, Cefazolin EV replaced Penicilin. A skin biopsy was performed, revealing a large subepidermal blister with minimal lymphocyte infiltration, dermis with edema and fibrosis that extended through hypodermis. Small blood vessels were PAS (+) with thickened walls, some of them surrounded by lymphocytes inflammatory infiltration compatible with diabetic microangiopathy and Bullous Diabeticorum (BD). While hospitalized insulin schedule was started, followed by oral hypoglycemic agents on discharge. Skin signs related to Diabetes Mellitus may be present up to 30% of diabetic patients. Although rare, some bullous lesions may be found within the characteristic DM's lesions. BD presents as spontaneously developing bulla on diabetic patients. They are generally asymptomatic and no previous trauma is needed. They are usually located on distal lower limbs. Although their pathogenesis is still unknown, circulatory-related-factors such as diabetic microangiopathy are suspected. Histologically an intra or subepidermal blister is usually found. Differential diagnosis such as cutaneous porfiria, bullous pemphigoid and adverse drug reactions, among others, should be ruled out. This case is presented because BD is strongly associated with DM and its presence should lead to a specific detection of this pathology.

## ABSTRACTS

### PREVALENCE STUDY OF CONTACT SENSITIZATION IN PSORIATIC PATIENTS COMPARED WITH NON-PSORIATIC PATIENTS.

Muñoz C, Valdés P, Szot J, Espinoza M, Zapata S.

Background: the relation between psoriasis and allergic contact dermatitis (ACD) is conflicting. Studies of the frequency of ACD among psoriatic in comparison to non-psoriatic have given contradictory results. Objective: we aimed to determine the difference in the prevalence of contact sensitization in psoriatic patients compared with non-psoriatic patients. Methods: we performed a descriptive, exploratory study on 100 adult's patients population in Santiago, Chile. Fifty of the patients were psoriatic (Group I) and 50 were non-psoriatic (Group II). Both groups answered questionnaire about personal and familiar medical past history. All patients were patch tested with 24 standard allergens (European Standard Series). Results: both groups were comparable in sex, age and past medical history. There were differences in their familiar medical history, familiar psoriasis history and alcohol consumption ( $p < 0.05$ ). 47 patients had a positive patch test (PT), 51.1% were from Group I, average age 48.8 years. Most common allergens in this group were Niquel sulphate (58.3%), Cobalt chloride (41.7%) and Potassium dichromate (12.5%). Group II represented 48.9% of positive PT, average age 56.5 years. Most common allergens were Niquel sulphate (56.5%), Cobalt chloride (39.1%) and Fragance Mix II (26.1%). There were no differences between common allergens on both groups ( $p > 0.05$ ). We found no differences between patients with positive PT and negative PT from Group I ( $p > 0.05$ ). Conclusions: our results showed an increased incidence of contact sensitization within psoriatic and non-psoriatic patients. We did not find any risk factor in psoriatic for presenting contact sensitization. It would be useful to continue this study using patch test with psoriatic specific allergens.

### SKIN CANCER IN ANTOFAGASTA, CHILE (1998 –2004).

Carrasco P, Zamora R, Zemelman V.

In Chile, skin cancer incidence rates showed an increase between 1993 and 2001 due to the increase of sun exposure. On the other hand, inorganic arsenic in drinking water has been associated with skin cancer. The population of Antofagasta (23°38' S) has been exposed to high UV radiation and also to high concentrations of arsenic in drinking water (1950-1970). The purpose of this study was to analyze the incidence rates of Basal Cell Carcinoma (BCC), Squamous Cell Carcinoma (SCC) and Malignant Melanoma (MM) in Antofagasta between the years 1998 and 2004 and characterize the tumors according to age, sex, histopathological type and anatomical localization. The data was collected from the Antofagasta's Health Service Cancer Registry Program. A total of 1790 patients with skin tumor were collected (52% males and 48% females). The total number of BCC was 1356, mean of 59.2 years old, 51% of BCC were females. Regarding anatomic localization, BCC was mainly located in the face (37% in females, 27% in males) followed by trunk (11% in females, 14% in males) and scalp (8% in females, 7% in males). The BCC incidence rates in Antofagasta decreased from 45/100.000 in the year 1998 to 30/100.000 in the year 2004. These incidence rates are much higher than in Santiago (10.8–21.5; 1998-2001) and Concepción (14.3–16.4; 1998-2001). Regarding localization, a higher percentage of trunkal BCC was observed in Antofagasta, in both sexes than in Santiago (5%) and Concepción (6%). The total number of SCC was 329, mean age of 62.56 years old, (36% female, mean 61.1 years old, 64% male, mean 64 years old). Concerning anatomic localization, SCC in males was mainly located in the face (20%), arms (14.5%) and trunk (11.6%); SCC in females was mainly located in arms (17%), face (16%), trunk (11%). The SCC incidence rates in Antofagasta decrease from 14/100.000 in the year 1998 to 9/100.000 in the year 2004. These incidence rates are also higher than in Santiago (6–6.3; 1998-2001) and Concepcion (4–5.5; 1998-2001). Regarding localization a higher % of SCC in the trunk was observed in Antofagasta (11.4%) than in Santiago (6.1%) and in Concepción (7%). The total number of MM was 101, mean 55.1 years old (47.2% males, mean 55 years old, 52.5% females, mean 55.3 years old). Concerning localization, MM in males was mainly located in the face (29%), trunk (16.6%); MM in females was mainly located in lower limbs (26%), face arms (19%). The MM incidence rates in Antofagasta are 4–5/100.000 (1988-2004), a beat higher than in Santiago (3.2–2.4; 1998-2001) and Concepción (2.8–1.9; 1998-2001). Regarding localisation, a lower % of MM in the trunk was observed in Antofagasta compared with Concepción (12% vs 23%) and similar % with Santiago. The higher BCC and SCC incidence rates observed in Antofagasta are partly due to a higher sun exposure and also due to the chronic exposure to arsenic from this population. The higher % of trunkal BCC and SCC in Antofagasta, especially in females, showed the effect of a long term exposure to arsenic. This study shows the relationship between arsenic exposure and skin cancer in a region from Chile.

#### PAEDIATRIC SKIN DISORDERS ENCOUNTERED AT THE EMERGENCY DEPARTMENT OF THE CLINICAL HOSPITAL UNIVERSITY OF CHILE.

Leiva A, Yutronic J, Espinoza M, Bello MP, Correa F, Saavedra T, Zemelman V.

Background / Objectives: children skin diseases are frequent among general population in Chile; however, there is little information about the pediatric skin disorders encountered at an Emergency Department in our country. The purpose of this study was to analyze the pattern and frequency of children skin disorders at the Emergency Department from our hospital, during the year 2005. Methodology: medical reports from all the children patients (17.899) attending the Emergency Department (ED) during the year 2005 were analyzed retrospectively. The data was classified by sex, age, month of the year and clinical diagnosis. For the statistical analysis, the Fisher test was used. P values <0.05 were considered significant. Results: skin disorders represented 9.96% (1782) of all pediatric emergency care unit visits (43.88% females and 56.12% males). The average age was 5.12 years old (95% CI; 4.94-5.31). The clinical diagnoses were classified in: allergies (34.1%), infectious diseases (38.78%), immunological disorders (2.58%), tumors (1.29%) and other skin disorders not classified under the previous groups (22.84%). No significant differences by gender were observed in all groups. Within the allergies, we observed a 41.13% due to insect bites, a 20.61% due to allergic rash and a 16.74% corresponding to acute urticaria. Among the infectious skin diseases, those of viral (70.9%) and bacterial (25.03%) origin were the most frequent. In general, for a specific diagnosis, the frequency was: wounds and contusions (15.66%), stings of insects (14.2%), herpangina (9.37%), chickenpox (7.18%), allergic rash (7.13%), acute urticaria (5.78%) and herpetic gingivostomatitis (4.26%). Both, the allergic and infectious diseases presented seasonal variation, being the allergic diseases more frequent in spring-summer and the infectious diseases more frequent in autumn. The results obtained in our study are different from the results of adults skin disorders encountered at the ED at the same year. More children than adults visited the ED with allergies, infectious and immunological diseases (p<0.001). Conclusions: unlike results published in the international literature, we obtained a high frequency of skin disorders in children attending the ED from our hospital, these disorders increased dramatically in the summer period. To know the pattern of the children skin diseases at the ED in our hospital may help to conduct a dermatological education towards clinician non dermatologists working at ED and also at primary health level in Chile.

#### ANALYSIS OF REGRESSION IN BASAL CELL CARCINOMA AFTER INCOMPLETE EXCISION.

Silva P, Zemelman V, Sazunic I.

Basal Cell Carcinoma (BCC) incident rates have increased in Chile in the last decades. Total surgical excision is the most frequent treatment for BCC. Histological evidence of incompletely removed BCC is a common fact after surgery. The purpose of this study was to analyze 52 incompletely removed BCC who underwent re-excision. Regressed and non-regressed tumors were studied to assess the influence of sex and age of the patient, anatomic location, histopathological type and size of the tumor. No significant differences were found between the two groups of tumors according to the studied variables. 40 % of the BCC showed no residual tumor in the re-excision samples, implying that some event in the interval between the first and the second surgery may lead to eradication of residual tumor. The exact mechanism is unclear, but the inflammation and immunological changes after the first surgery play an important role.

#### ADULT SKIN DISORDERS ENCOUNTERED AT THE EMERGENCY DEPARTMENT OF THE CLINICAL HOSPITAL UNIVERSITY OF CHILE.

Yutronic J, Leiva A, Espinoza M, Bello MP, Correa F, Saavedra T, Zemelman V.

Background / Objectives: adult skin diseases are frequent among general population in Chile; however, there is little information about the adult skin disorders encountered at an Emergency Department in our country. The purpose of this study was to analyze the pattern and frequency of adult skin disorders at the Emergency Department from our hospital, during the year 2005. Methodology: medical reports from all the adults patients (32307) attending the Emergency Department (ED) during the year 2005 were analyzed retrospectively. The data was classified by sex, age, month of the year and clinical diagnosis. For the statistical analysis, the Fisher test was used. P values <0.05 were considered significant. Results: skin disorders represented 8.84% (2857) of all adult emergency care unit visits (48.58% females and 51.42% males). The average age was 40.48 years old (95% IC; 39.81-41.15). The clinical diagnoses were classified in: allergies (29.68%), infectious diseases (27.06%), immunological disorders (1.19%), tumors (1.96%) and other skin disorders not classified under the previous groups (40.11%). Significant differences by gender were observed in allergic diseases, with a higher frequency in females (60.14%) than males (39.86%); (p<0.001). Likewise, immunological disorders were more

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frequent in females (67.65%) than males (32.35%); ( $p=0.037$ ). No significant differences by gender were observed in the other groups. Within the allergies, we observed a 33.49% due to allergic rash, a 19.81% due to acute urticaria and a 16.98% corresponding to insect bites. Among the infectious skin diseases, those of bacterial (72.31%) and viral (23.93%) origin were the most frequent. In general, for a specific diagnosis, the frequency was: wounds and contusions (27.97%), cellulites (11.8%), allergic rash (9.94%), acute urticaria (5.88%), stings of insects (5.04%), herpes zoster (3.33%) and burns (4.26%). Both, the allergic and infectious diseases presented seasonal variation, being both more frequent in spring-summer. The results obtained in our study are different from the results of children skin disorders encountered at the ED at the same year. More children than adults visited the ED with allergies, infectious and immunological diseases ( $p<0.001$ ). Furthermore, within the infectious diseases, the frequency of bacterial infections was higher in adults than in children. Conclusions: the results of our study are similar to that published in the international literature. The allergies and infectious diseases encountered in adults at the ED increased in the summer period. To identify the pattern of the adult skin diseases at the ED in our hospital may help to conduct a dermatological education towards clinician non dermatologists working at ED and also at primary health level in Chile.

## GENÉTICA

### 3<sup>rd</sup> INTERNATIONAL CONFERENCE ON BIRTH DEFECTS AND DISABILITIES - RÍO DE JANEIRO, BRASIL.

#### SINDROME FEMUR FIBULA ULNA. PRESENTACIÓN DE UN CASO CLINICO.

Cares C, Sanz P, Tellerías L, Zúñiga ML.

El término complejo o síndrome FFU (Fémur Fibula Ulna), se emplea para denominar una asociación de malformaciones de extremidades, que comprometen fémur, peroné y cúbito. Se caracteriza porque los defectos de las estructuras se combinan en forma muy variable, pudiendo presentarse junto a anomalías de dedos de manos y/o pies en el lado del cúbito y/o peroné afectado. Su prevalencia se ha estimado entre 1,1 a 2 en 100.000 RN vivos. Se describe como entidad a partir de 1967 por existir una tendencia de los defectos que lo caracterizan a ocurrir juntos en el mismo paciente, mucho más frecuentemente de lo que podría esperarse sobre la combinación de los defectos simples. La ocurrencia de esta entidad es esporádica, siendo rarísimos los casos familiares, tampoco existe un factor ambiental involucrado. Las mutaciones somáticas se han planteado como causa probable. Se presenta el caso de un paciente de sexo masculino, de dos años de edad, hijo de padres sanos, sin consanguinidad, y sin antecedentes de exposición a teratógenos durante el embarazo, con residencia en Melipilla, sector rural cercano a Santiago. Al examen físico destaca asimetría de cráneo, micrognatia, gran defecto en las cuatro extremidades: amelia bilateral de extremidades superiores, acortamiento bilateral de extremidades inferiores, con mayor compromiso a derecha, sindactilia de segundo y tercer dedos de pie derecho, pie valgo bilateral. Al examen radiológico destaca hipoplasia proximal bilateral del fémur, con mayor compromiso a derecha, hipoplasia tibial bilateral y agenesia bilateral de peroné. Se descartaron malformaciones cardíacas y cerebrales.

#### DELETIONS OF THE HOMEBOX GENE SHOX (SHORT STATURE HOMEBOX) IN CHILDREN WITH IDIOPATHIC SHORT STATURE IN CHILE.

Aravena T, Ureta P, Cares C, Escribano G.

Height is result of interactions of several factors including those of genetic origin. Short stature is a fairly frequent disorder in children, with an incidence of 3 in 100. Idiopathic short stature refers to patients who are short due to various unknown reasons. Mutations of a human homebox gene, SHOX (Short Stature Homeobox), have been shown to be associated with the short stature phenotype in patients with Turner syndrome, most patients with Leri-Weill dyschondrosteosis and in some individuals with idiopathic short stature. Around 80% of SHOX mutations are complete gene deletions, whereas diverse point mutations account for the rest. The aim of this study was to estimate the prevalence of SHOX deletions in Chilean patients with idiopathic short stature. All patients had normal karyotype, absence of chronic or syndromic disease, and their height for chronological age were below the third percentile of national height standards. We analyzed the SHOX deletions by PCR-amplified with fluorescent primers of highly polymorphic microsatellite markers located around the SHOX coding region in 21 patients. Analysis of parental DNA was performed. Fluorescence in situ hybridization (FISH) was performed in the probands who had only one fragment size of each marker.

Complete SHOX gene deletions were detected in 2 of the 21 patients. The prevalence of short stature due to SHOX gene deletions among children with short stature in Chile appears to be similar to that of the other international studies. The screening of SHOX deletions should be included in the routine genetic testing of children with idiopathic short stature.

#### CLINICAL FEATURES OF CHROMOSOME 22Q11.2 DELETION SYNDROME PATIENTS FROM CHILE.

Guzmán ML, Aracena M, Mellado C, Astete CP, Aravena T, Repetto GM.

Chromosome 22q11.2 microdeletion syndrome (del22q11) is the most common known microdeletion syndrome, with an estimated incidence of 1/4000-5000 newborns. It has variable clinical presentations. Objective: To illustrate the clinical features of Chilean patients with del22q11. Methods: Retrospective review of medical records of patients with FISH-demonstrated deletions from 3-4 tertiary centers from Santiago. Results: 58 patients with del22q11 were identified, 44,8 % male. The mean age ( $\pm$ SD) at diagnosis was  $5,17 \pm 16,76$  years, ranging from newborn to 38 years. 34 individuals (58,6%) had CHD, and 4 others had structural anomalies not requiring surgery. The most common CHD were Tetralogy of Fallot (29,4% of patients with CHD), interrupted aortic arch (IAA) (8,8%), and VSD (29,4%). Mean ages ( $\pm$  SD) at diagnosis for patients with and without CHD were  $4,82 \pm 6,41$  and  $10,25 \pm 10,44$  years, respectively. Height/age was below the 50th centile in 37,9%, and below the 5th centile in 22,4%. Calcium levels were recorded in 24 patients; 10 had neonatal hypocalcemia. Otolaryngeal problems were present in 34 patients, and included submucous cleft palate (n= 11), velopharyngeal insufficiency (n= 5), overt cleft palate (n= 9), and laryngeal membrane (n= 2). Conclusions: though probably biased by the retrospective nature of the study and inclusion of only live patients, the results provide useful data for patient counselling and care. Frequency and types of common medical complications are similar to published US and European series, but diagnosis tends to occur later, in particular in patients without CHD. The non-cardiac presentation likely requires a higher index of suspicion by treating clinicians.

#### HUMAN CHIMERAS: TWO POSSIBLE CASES

Castillo S, Soto V, Seebach C, Cifuentes L, Acuña M, Jorquera H.

A chimera is an individual with two or more populations of genetically distinct cells that originated in different zygotes; if the different cells emerged from the same zygote, it is called a mosaicism. We have seen two possible cases product of natural conceptions and without antecedents of having been twin at the beginning. First case (P.Z.C.): Diagnosis: sexual ambiguity. First child of healthy, young and unrelated parents. There is no familial history of similar manifestations. Uneventful pregnancy and delivery, birthweight 3.296g(p50), birthlength 50cm(p50), cranial circumference 36cm(p50) and Apgar 9 and 10. Clitoromegaly and slight dysmorphies: coarse facial features, long philtrum and big mouth. Bilateral inguinal hernias. Camptodactyly on both hands. Caryotype: 45,X[36](76,6%)/46,XY,r(18)(p 11.3; q 21.3)[9](19,1%) /46,XY,r(18)(p11.3 ; q ?)[2](4,3%). Positive SRY. Prognosis: difficult to establish considering unknown proportions of cells with different chromosomal constitutions in tissues. Treatment: surgery of the inguinal hernias and gonadal biopsies. Gonadectomy if testicle. Esthetical correction of external genitalia if necessary. Feminine hormonal restitution around puberty. Reproduction may be possible with donated ova. Female sex was assigned considering that: genetically, two peripheral blood karyotypes and one oral mucosa X/Y FISH have shown an approximate 80% of 45,X cells; endocrinologically, male testosterone levels have diminished to half since newborn; anatomically, external genitalia show slight virilization of female structures, ultrasound confirms the presence of an uterus; and, surgically, correction to female is easier and has better functional prognosis. Both parents need to be studied cytogenetically for estimating the recurrence risk. Case 2 (F.Z.): Diagnosis: Down syndrome with atypical features. Two peripheral blood karyotypes show one cell line 47,XY,+21 in 86,1% (68 mitosis) and 45,X (11 mitosis) in 13,9%. Short tandem repeats genotyping using ten loci (D3S1358,vWA,FGA,AMEL,D8S1179, D21S11,D18S51,D5S818,D13S317,D7S 820) revealed only one pair of alleles for each locus in both samples. At a first sight, this favors a mosaicism hypothesis.

#### CHARGE SYNDROME: DESCRIPTION OF A CLINICAL CASE.

Escribano G, Aravena T, Castillo S.

CHARGE Syndrome represents an important cause of congenital anomalies with an approximate birth incidence of 1 in 120000. It was reported in 2004 that most of CHARGE Syndrome cases had mutations in the gene CHD7 located in chromosome 8 which is expressed in early embryonic development in specific areas of brain, kidney and skeletal muscle. CHARGE syndrome also has been related to mutations in gene SEMA3E located in chromosome 7, which has an important role in brain development. Clinical features of CHARGE Syndrome are: eye coloboma, heart anomaly,

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choanal atresia, anal stenosis, genital anomaly, ear anomalies, deafness, growth and development retardation. The case we present is a 5 month old male without significant family history, product of a second dizygotic twin from a non complicated pregnancy with a healthy first twin. His parents are young, healthy and not blood related. He was born from C section with birth weight of 2080g , height 39,5 cm, and APGAR score of 8 – 10. On physical examination he has multiple malformations including: brachycephaly, large bregma, complete and bilateral lip-palate cleft, protruding and incomplete folded ears with sharp helix, bilateral iris coloboma, umbilical hernia, psychomotor retardation, transition crease on right palm, upper and lower limb mild hypertonia. Complementary studies show: G banding Karyotype 46, XY, first Echocardiogram shows atrial septal defect, follow up echocardiogram done later was normal, Brain CT Scan and ultrasound were normal, complete spinal X ray and renal ultrasound were normal. Molecular studies are still pending.

### SYNDROMATIC ASSOCIATION: POLAND, GOLDENHAR, MOEBIUS, KLIPPEL FEIL.

Cares C, Aravena T.

The clinical case we present is a 10 years old male patient, who has in his physical exam: left hemifacial microsomia, left severe microtia, mandibular left branch shortening, periferical left facial paralysis. Short and wide neck with limitation in lateral movements. Left pectoralis major absence with thoracic asymmetry. Left upper limb absence of the radio and thumb. It also presents severe left deafness secondary to severe microtia . He has normal intellectual development. Without affectation of the whole rest of organs and systems. Spine X-ray shows C3-C4 hemivertebrae and C4-C5 fusion. Patient's diagnoses are: Poland Syndrome, Moebius Syndrome, Klippel Feil Syndrome , Goldenhar Syndrome. SASDS (subclavian artery supply disruption sequence) was proposed in 1986 by Bouwes-Bavinck and Weaver, as a cause of a spectrum of affections that commit the region irrigated by the subclavian artery in the embryonic period. They suggested that between the 4<sup>th</sup> and 6<sup>th</sup> weeks of gestation, the alteration of subclavian artery different branches can generate premature regression, obstruction, that drive to lost of vascular circulation and enervation, fibrosis and atrophies of the affected segment. This sequence was think like a cause of a group of pathologies that could share the same pathogenia: Poland Syndrome, Klippel-Feil Syndrome, Moebius Syndrome and Goldenhar Syndrome. Since then several cases of association have been reported among some of these syndromes. In our knowledge it has not been reported cases that present the four syndromes until now.

### HEREDITARY STICKLER SYNDROME AND DE NOVO PEUTZ JEGHERS SYNDROME IN A PATIENT.

Aravena T.

Stickler syndrome is an autosomal dominant disorder with characteristic ophthalmological and orofacial features, deafness, and arthritis. Abnormalities of vitreo are a pathognomonic feature, usually associated with myopia. Peutz Jeghers syndrome is an autosomal dominant disorder characterized by melanocytic macules of lips and buccal mucosa, multiple gastrointestinal hamartomatous polyps, and an increased risk of various neoplasms. We present a female patient with prenatal history of intrauterine growth restriction (mother developed preclampsia during the pregnancy). At birth he presented a weight of 1940 g, a height of 43 cm, and a cranial circumference of 33 cm. The physical exam at 4 year of age highlighted a flat midface with depressed nasal bridge, short nose, anteverted nares, and light micrognathia, submucous cleft palate and joint hypermobility. The cardiac exam showed a holosystolic murmur grade 2/6 at the left lower sternal border. Slender extremities and long fingers. Stature and intellect are normal. Cardiac US was normal. Otorhinolaryngology evaluation confirmed submucous cleft palate and high frequency sensorineural hearing loss. Caryotype: 46,XX. X-rays was normal. When de patient was 6 year of age pyperpigmentation of lips and oral mucosa was observed, and the intestinal endoscopy revealed the presence of jejunal polyps. Colonoscopy of the patient revealed no polyps. When the patient was 7 year 1 month of age, intestinal obstruction was detected and diagnosed radiologically as jejunal intussusception. Jejunal surgical resection was performed. Pathologic examination of polyps confirmed the diagnosis of Peutz Jeghers syndrome-associated polyps. Mother exam showed myopia and astigmatism, and articular pain in conjunction with joint space narrowing and osteophytes. The family history of intestinal disorder was negative.

## HEMATOLOGÍA

### 11<sup>th</sup> INTERNATIONAL MYELOMA WORKSHOP / IVTH INTERNATIONAL WORKSHOP ON WALDENSTRÖM'S MACROGLOBULINEMIA - KOS, GRECIA.

GENOMIC ABERRATIONS IN MONOCLONAL GAMMOPATHY OF UNDETERMINED SIGNIFICANCE (MGUS) AND SMOLDERING MULTIPLE MYELOMA (SMM) AND THE RISK OF PROGRESSION TO MULTIPLE MYELOMA (MM).

Van Wier SA, Larson DM, Chng W, Rempel R, Ahmann GJ, Henderson KM, Figueroa G, Kyle RA, Rajkumar SV, Dispenzieri AD, Bergsagel PL, Stewart AK, Therneau TM, Greipp PR, Fonseca R.

Introduction. Molecular cytogenetic defined subgroups of patients with MGUS and SMM may differ in their risk of progression to MM; but to what extent is not known. These subgroups have been firmly established in MM as associated with pathogenesis and clinicopathologic features. Similar abnormalities have been observed in MGUS. Materials and methods. We studied patients diagnosed with MGUS or SMM defined by accepted criteria. Patients were excluded if they had active MM. Patients provided written informed consent for the collection of a research bone marrow aliquot collected at the time of routine bone marrow procurement. Cytospin slides were made and studied for translocations: t(4;14), t(11;14), t(14;16), IgL-light chain (+22q11) and deletions of chromosome 13q (D13) and 17p. In cases where we did not find a primary IgH translocation we looked for +14q32 NOS using a break-apart strategy. We used interphase fluorescence in situ hybridization (FISH) combined with cytoplasmic immunoglobulin (cIg-FISH) as previously described by us. Results. We studied the following cohorts of patients: 215 for t(4;14) and abnormal in 20 (9.3%); 200 for t(11;14) and abnormal in 36 (18.0%), 192 for t(14;16), and abnormal in 5 (2.6%), 201 for D13 and abnormal in 77 (38.3%), 173 for 17p and abnormal in 5 (2.9%), 180 for IgL-light chain translocations and abnormal in 11 (6.1%) and 149 for +14q32 and abnormal in 41(27.8%). A total of 168 patients were evaluable for all assays. Twenty-seven patients (16%) progressed to active MM at a median of 20.2 months. In our initial set of observations we discern a higher risk of progression for those with t(4;14) (31.3% progressed) and lower in patients with t(11;14) (4.2% progressed). D13 or 17p13 (with low power due to few cases) did not seem have a discernible effect on progression. Conclusion. Additional follow up time and cases will be needed to better define the role of genetic markers to predict progression to MM. Although translocations of IgH and IgL- $\lambda$  are found in similar frequency in MGUS/SMM to that of MM, the subtypes would appear to dictate differential risk of progression. In this larger study we find a lower prevalence of D13 than in MM consistent with it being a progression event (38% versus 54%). Deletions of 17p13 are also less common and also indicative of a progression event (3% versus 11%).

## INMUNOLOGÍA

### XXVI CONGRESS EAACI (EUROPEAN ACADEMY OF ALLERGOLOGY AND CLINICAL IMMUNOLOGY) - GOTEMBURGO, SUECIA.

RECALCITRANT WARTS TREATED WITH CIMETIDINE AS IMMUNOSTIMULANT.

Marinovic M, Guzmán M.

Background: it is believed that most warts are self-limiting and generally require little or no treatment, however there is a group of patients who show no therapeutic response to multiple treatments that includes cryosurgery, dinitrochlorobenzene, imiquimod, etc. Cimetidine is a histamine 2 receptor antagonist that has been used mainly to treat peptic ulcer disease. In the last years it has been reported to be useful for the treatment of herpes simplex, herpes zoster, mucocutaneous candidiasis, genital and laryngeal condiloma, gastrointestinal cancer and warts because of its immunomodulatory effects, but its effectiveness is still debated. The purpose of the study was to evaluate treatment with oral cimetidine in patients with recalcitrant flat and vulgar warts with at least 1 year of regular treatment. Patients and methods: We treat five patients age 7–30 years with recalcitrant and multiple flat and vulgar warts derived to the Immunology Unit of San Borjas Arriaran Hospital, Santiago Chile. All of them were studied with blood count, immunoglobulin count, lymphocytes population and HIV serology before treatment. They received cimetidine 200 mg three times a day for 3 months, and no other associated treatment. Results: All the patients were men, two of them were seven and eleven years old and the other three were adults between 25 and 30 years old. Four of all had the immunologic parameters between normal ranges. Just one from 25 years old had hypogammaglobulinemia and mild CD 4 lymphopenia. The entire group responded between the second week and the second

## ABSTRACTS

month of treatment, with complete resolution at the end of 3 month of therapy. None of them showed adverse reactions to cimetidine. The follow up was at least one year for each patients, without recurrence. Conclusion: In view of some recent publications, it seems that cimetidine activates Th1 cells to produce IL-2 and IFN-g and that their expression correlates with warts remission because they stimulate the cellular immune response. With our results we conclude that cimetidine is a safe, cheap and effective treatment for recalcitrant warts in adults and children and because 3 is a small group of patients without a control group we propose the need for a randomized bigger control trial.

### **4<sup>th</sup> CONFERENCE IAS (INTERNATIONAL AIDS SOCIETY) ON HIV PATHOGENESIS, TREATMENT AND PREVENTION - SIDNEY, AUSTRALIA.**

RESISTANCE TO ANTIRETROVIRAL THERAPY IN PEDIATRICS HIV/AIDS CHILEAN PATIENTS, 2002-2005.

Acevedo W, Álvarez A, Hernández M., Comité Nacional de SIDA pediátrico.

Objectives: Resistance limits the efficacy of antiretroviral drugs for treatment of HIV. At this moment, in Chile there are 171 children living with HIV/AIDS. Since 2001 there is a free access to highly active antiretroviral therapy (HAART). The aim of this study is to determine drug resistance associated mutations in pediatrics patients. Methods: Between 2002-2005, 42 pediatric patients with therapeutic failure were studied for genotypic resistance (Trugene HIV-1, Bayer). Results: The resistance only to nucleoside reverse transcriptase inhibitors (NRTI) was 10%; resistance only to non nucleoside reverse transcriptase inhibitors (NNRTI) was 6.7%; resistance only to protease inhibitors (PI) was 0%; resistance to all drugs (NRTI, NNRTI, PI) was 16.7%; resistance to all NRTI and NNRTI was 33%. Individual mutations most frequently found for reverse transcriptase were T215Y (54.8%), M41L, D67N, V118I and M184V (40.5%), L210W (35.7%); for protease were L63P (81%), L10I/V/F (52.4%), I54V (28.6%), V82A/T (28.6%), L90M (21.4%). Conclusions: This is the first Chilean study about mutations patterns associated to resistance in antiretroviral treated pediatric patients with virological failure diagnosis. This data provides important information about the molecular epidemiology of drug resistance mutations and its application for design new strategies of an effective HAART. The results show diminished possibilities for treatment of an important group of children and the necessity of new drugs and formulations for these patients.

## MEDICINA NUCLEAR

### **INTERNATIONAL CONFERENCE IAEA (INTERNATIONAL ATOMIC ENERGY AGENCY) ON CLINICAL PET AND MOLECULAR NUCLEAR MEDICINE) - BANGKOK, TAILANDIA.**

GASTROINTESTINAL STROMAL TUMORS (GIST) ASSESSMENT USING 18F-FLUOR-DEOXYGLUCOSE.

Massardo LT, Jofré MJ, Canessa J, Sierralta, González P, Jaimovich R.

Introduction: stromal gastrointestinal tumors (GIST) are relatively infrequent soft sarcomas, although correspond to the most common mesenchymal tumor in the gastrointestinal tract. Surgery is the main therapy during initial stages. Nearly a third of them could be malignant (higher risk) depending on the localization, size and histological parameters. Chemotherapy and radiotherapy have low therapeutic value. Specific molecular therapy associated to surgery using imatinib-mesylate (GLIVEC®) – a selective transducer signal inhibitor for some tyrosine-kinase receptors – is currently use. It is helpful in non-resectables, recurrent or metastatic tumors. Metabolic fluorine18-deoxyglucose (FDG) allows to characterize tumor behavior demonstrating good predictive value. Promising results have been obtained using adjuvant and neoadjuvant protocols. There are some multicenter in-course trials including FDG in order to evaluate early response to GLIVEC® therapy. Other recently developed molecules such as sunitinib malate (SUTENT®) are used in non-responders. Method: we have performed 18 FDG studies to 15 GIST patients referred from different centers in a period of 48 months, corresponding approximately to 1% of all cancers in adults. and to 6% of gastrointestinal tumors. The mean age of the group was 57±10.6 y.o., ranging from 33-72 years, 60% of the patients were male. GIST primary localization corresponded to jejunum or ileum (4), duodenum (3), esophagus, stomach (1) besides, 2 retroperitoneal/extra intestinal cases and 5 disseminated cases with no clear origin site. Eight out of fifteen patients presented known dissemination when FDG was performed. PET-FDG was performed to assess: a) medical therapy control in 9 cases: 7 with GLIVEC®, 1 with SUTENT® post GLIVEC® and 1 post chemotherapy, b) restaging in 6 and c) staging in the other 3 cases (1 submitted to surgery and 1 extensive tumor to decide GLIVEC® therapy). All but one patients already had surgery performed with a mean 12±10m prior to their first FDG. GLIVEC® therapy ranged between 3-43 m. Mean FDG dose was 481±74

MBq injected with a mean serum glucose level of  $89 \pm 9$  mg/dl. Images were obtained with a dedicated PET system Siemens ECAT EXAT HR+. Quantitative analysis using SUV measurements were used for metabolic follow-up. Results: 44% of FDG studies were positive for malignancy. Within the staging group, 2 recent post-surgery patients were negative and the disseminated non operated was positive. Concordance with recent anatomical or functional images [Computed Tomography (CT), bone scintigraphy, abdominal echography or prior FDG]. was observed in 61% of the studies. New unknown lesions were found in 7 studies (39%) located in mediastinum, liver, peritoneum and skeleton. Only 3/8 cases with molecular therapy had negative FDG for active tumor; other 2 cases presenting 20% and 50% of CT regression had clearly positive FDGs without new lesions. Three patients had FDG follow-up: one remained negative, other progressed with new lesions and the non-operated case with extensive dissemination presented total FDG regression. Conclusion: in our initial experience, functional images with PET-FDG demonstrated great value for metabolic control of molecular therapy, staging and restaging post surgery of GIST tumors.

#### QUALITY CONTROL AND LEARNING EXPERIENCE IN CLINICAL NUCLEAR CARDIOLOGY AT A TEACHING HOSPITAL FACILITY.

Jaimovich R, Massardo LT, González P, Alliende I.

INTRODUCTION: traditional Nuclear Medicine training includes seminars, guided practices and self-learning modules (continuing education and internet-based). We have developed recently a combination of training, quality control and active clinical research, with postgraduate nuclear medicine residents and pregraduate technology students. Goal: a) to assess the reproducibility of diverse techniques, among staff specialists and residents; and b) software application quality control. This also in order to accomplish international parameters, mainly in nuclear cardiology training. METHODOLOGY AND RESULTS: four main cardiovascular subjects were selected: A) Reproducibility in Lung V/Q Scans Interpretation. 401 studies from 382 patients with a possible pulmonary embolism were analyzed retrospectively; a blind lecture was performed by 6 independent observers with different experience level. Interpretation was based on individual experience and revised PLOPED criteria. Original reports included 27.2% high probability and 67.3% low probability. Interobserver agreement range was: 73-86% and correlation with original report: 74-82%. Excellent interobserver concordance and kappa was found, higher in experienced observers. B) Perfusion SPECT in Coronary Artery Disease (CAD). 60 cases with recent myocardial infarction with successful thrombolysis were blindly interpreted by 2 independent specialists and also by 5 in-training observers from different universities. Excellent interobserver agreement was obtained by specialists for normal/abnormal perfusion and wall motion (98.3% and 93.3%, respectively). Agreement between perfusion and wall motion was adequate, as well as assigned artery analysis. There was good correlation interpreting myocardial perfusion SPECT at both centers, with better adjustment in more experienced observers. Currently, we are developing another study comparing interobserver reproducibility for exercise electrocardiogram and perfusion images including 100 known and unknown CAD patients. Four (local and IAEA) different experienced residents interpretations are compared to both specialist's report. C) Reproducibility of Myocardial Spect with Artifacts. Patient motion and extracardiac tracer activity may affect myocardial perfusion SPECT interpretation in CAD assessment. Interpretation changes after applying motion correction software, as well as reproducibility of automated functional parameters in presence of extracardiac activity (intestinal loops or liver) were analyzed. C.1. Motion Correction: 160 selected CAD  $^{99m}\text{Tc}$ - sestamibi studies processed with automatic and manual motion correction were divided according to presence of i) motion severity during standard acquisition and ii) perfusion defects. Motion correction must be used with caution to optimize myocardial perfusion SPECT CAD specificity. It's use must be decided according to individual performance. Cases with severe motion should be repeated due to variable results. C.2. Extracardiac Activity: 100  $^{99m}\text{Tc}$ -Sestamibi- Dipyridamol studies, 50 with and 50 without extracardiac activity; each included 25 with and 25 without perfusion abnormalities. They were processed automatically and by 4 independent operators with commercial software. Extracardiac activity affected automatic QGS-QPS assessment, even with manual intervention. Reproducibility worsened when significant activity was close to the myocardium. Perfusion abnormalities did not interfere with reproducibility. It was higher for functional than for perfusion parameters. D) Reproducibility in Planar and SPECT Radionuclide Angiography. 83 radionuclide angiography using labelled  $^{99m}\text{Tc}$ -red cells were classified according to ventricular dilation, wall motion abnormalities and systolic dysfunction. Both ventricular ejection fraction and volumes were obtained using available software for planar and SPECT. Good correlation existed between SPECT and planar studies for left ejection fraction and volumes. SPECT was useful in cases with functional abnormalities, however, was less reliable with smaller cavities. Excellent interoperator reproducibility for both techniques was observed, better for left ventricle. CONCLUSIONS: This academic task performed during the last 3 years -accomplished using local databases and daily routine exams- was mainly a practical training for our nuclear medicine residents in order to ammeliorate self-confidence in the interpretation. Less frequently performed techniques were also reviewed this way.

**NEFROLOGÍA****WORLD CONGRESS OF NEPHROLOGY - RÍO DE JANEIRO, BRASIL.**

OUTCOME OF 648 HEMODIALYSIS CATHETERS PLACED BY NEPHROLOGISTS OVER 36 MONTHS AT THE UNIVERSITY OF CHILE CLINICAL HOSPITAL.

Torres R, Laguna J, Pacheco A, Sanhueza ME, Briones E, Segovia E, Cotera A.

Introduction: The present study was aimed at evaluating the clinical experiences, rate of success, and mechanical complications in the catheterization for hemodialysis over 36 month of following. Methods: During a 36-month period we prospectively collected data on success rate and mayor and minor complications, for all the temporary and tunnelled Dacron-cuffed catheters placed by Nephrologists in 509 adult patients admitted for hemodialysis at University of Chile Clinic Hospital from January 2004 to December 2006. Results: In the present study we report the results of 680 hemodialysis catheters at our centre, placed in 509 uremic patients, 262 males and 247 females, with a mean age of  $60.4 \pm 15.5$  years (mean  $\pm$  SD). There were 103 patients that required more than one catheter and 406 that required only one. There were 274 tunnelled and 374 temporary catheters. Of the tunnelled devices 2 were translumbar catheters. The indications for temporary access were uremic syndrome 60%, vascular access dysfunction 34%, and 6% others. For tunneled catheters 50% of indication was vascular access dysfunction, 40% uremic syndrome, and 10% others. Overall success rate was 98.3%, (97.8 and 98.6 for tunnelled and temporary catheters). The main place of insertion was supraclavicular (62% for temporary and 90.5% for tunnelled catheters). Overall complication rate was 10%, that included pneumothorax (3 patients), arterial puncture (53 patients) and haematoma (27 patients). There were no deaths or sequelae related with the procedure. The overall rate of complications was 11.2% in temporary access, and 9.4% in tunnelled catheters. The rate of complications between years 2004 and 2006 has decreased from 14.6% to 8.3% in temporary access, and 11.7% to 6.7% in tunnelled catheters. Conclusion: We conclude that in our experience rate of success is fairly good (98%) without difference between temporary and tunnelled catheters. The overall rate of complications that is 10%, decreases along the time of following, especially in the tunnelled catheters, indicating a curve of learning.

**HEART RATE VARIABILITY IN END-STAGE RENAL PATIENTS IN CHRONIC HEMODIALYSIS.**

Venegas R, Cano M, Torres R, Alvo M, Sanhueza ME, Pacheco A.

Introduction: decreased heart rate variability (HRV) have been described as an independent predictor of death from cardiac and non-cardiac causes, in patients that have myocardial infarction (MI), coronary disease, diabetes and ESRD. Uremics have heart and vessels dysfunction with a restricted response to haemodynamic changes. Using spectral analysis we have measured HRV in a Chilean uremic patients, for evaluation and risk stratification purposes, in order to define specific clinical interventions. Methods: We studied 27 (16 male, 11 female) chronic hemodialysis (HD) patients from University of Chile Clinical Hospital ( $49.5 \pm 13.5$  y/o), with  $6.4 \pm 6.2$  years HD. 74% of the patients were using antihypertensive drugs (18.5% one drug, 55.5% two or more). 75% of the antihypertensive drug users have a beta blocker. Haematocrit was  $33.0 \pm 7.0$ , serum  $K^+ = 4.9 \pm 0.5$  mEq/L and  $KT/V = 1.5 \pm 0.3$ . Only patients carrying a pacemaker were non candidates for the evaluation. The HRV was measured pre-dialysis from 5 minutes ECG records in supine resting position, for analysis in time and frequency domain. For time domain analysis the measurements were: a) Mean RR (time in seconds from normal to normal QRS, no ectopic beats), b) SDNN (standard deviation of all normal to normal r-r intervals of each patient), c) Triangular index (total number of normal intervals divided by the height of the histogram) d) SD1 and SD2 (short and long term variability in Poincare plot). For frequency domain the measurements were: a) Low frequency (LF) power in absolute ( $ms^2$ ) and normalized units (nu) in the range of 0.04 to 0.15 Hz, b) High frequency (HF) power in absolute ( $ms^2$ ) and normalized units (nu) in the range of 0.15 to 0.4 Hz and c) LF/HF index (ratio LF/HF). Normal values were collected from the Heart Rate Variability Standards from the TASK FORCE European and North American Society. Results: time domain analysis: Mean RR =  $0.743 \pm 0.118$  s (normal value-NV-  $> 0.75$  s). SDNN =  $0.017 \pm 0.012$  s (NV  $0.141 \pm 39$  s). Triangular Index =  $0.038 \pm 0.023$  (NV  $0.0163 \pm 0.0047$  for 5 min records). Frequency domain analysis (parametric): LF =  $99.8 \pm 210.6$   $ms^2$  (NV  $1170 \pm 416$   $ms^2$ ). LFnu =  $54.3 \pm 33.6$  % (NV  $54 \pm 4$  %). HF =  $34.0 \pm 124.6$   $ms^2$  (NV  $975 \pm 203$   $ms^2$ ). HFnu =  $17.5 \pm 18.8$  % (NV  $29.3 \pm 3$  %) and LF/HF ratio = 15.82 (NV 1.5-2.0). Conclusion: HRV measurement was significantly different in chronic HD patients, compared with standardized normal data in healthy subjects. Patients undergoing chronic HD have highly depressed heart rate variability, which involve cardiovascular high risk stratification. It is suggested that these patients must undergo some cardiovascular rehabilitation interventions.

## RESPIRATORY QUOTIENT (RQ) UTILITY IN THE FITNESS ASSESSMENT IN HAEMODIALYZED PATIENTS.

Negróni O, Cano M, Torres R, Sanhueza ME, Pacheco A.

Introduction: the anaerobic threshold (AT) is a fitness marker which can be measured using the respiratory quotient (RQ). It is the ratio between CO<sub>2</sub> production and O<sub>2</sub> consumption (VCO<sub>2</sub> / VO<sub>2</sub>). At the ESRD patients are exposed to an acid charge permanently and tend to metabolic acidosis. The haemodialyzed patients (HP) get correct this problem with the dialysis solution which has bicarbonate as alkaline source. Bicarbonate in plasma produces the following reaction: H<sub>2</sub>CO<sub>3</sub> ↔ CO<sub>2</sub> + H<sub>2</sub>O. Since the bicarbonate level in the HP depends on the bicarbonate mass transfer through the dialysis membrane and the CO<sub>2</sub> production by the metabolism, its level can change. HP has got a higher CO<sub>2</sub> level in their blood and therefore AT measuring method, though RQ, could have a low security level. The objective of this work was to compare the RQ (basal and under exercise) in healthy volunteers and HP. Methods: This was a prospective case-control study; 17 patients (33.8 ± 6.7 y/o, BMI 24.3 ± 3.6 Kg/m<sup>2</sup>) in maintenance chronic hemodialysis program (3 per week) with Kt/V = or > 1.2 were evaluated and compared with 18 healthy volunteers (30.61 ± 7.7 y/o, BMI 24.3 ± 4.2 Kg/m<sup>2</sup>). Furthermore, both groups did not show differences with gender distribution. All of them signed an informed consent and this study was approved by an ethic committee. Both groups participate in an exercise trial. This trial consists in a computerized cycloergometric measure of their physical performance, under increased charge of work. This protocol had an incremental rate of 15 Watt/min, and it was stopped when the subjects got a heart rate frequency at 85% of their maximum, extreme fatigue or a RQ = or > 1.3. Ventilation and gaseous exchange (O<sub>2</sub> uptake and CO<sub>2</sub> production) was registered by the "breath by breath" method. The results were analyzed using the software SPSS 12.0 and were showed as average and standard deviation (+/-). Results: Basal RQ for HP was 1.01 ± 0.18 and for healthy volunteers was 0.94 ± 0.1 (p = 0.16); RQ at 100 Watt of HP was 1.10 ± 0.07 and 0.95 ± 0.13 for healthy volunteers (p = 0.003). At the end of the test, 8/17 patients did not finish this assessment, and it was significantly different in healthy group where just 1/18 patients could not finish it. We did not find differences in basal RQ but there were significant differences at the end of the test (100 Watt) and the number of subject who were able to conclude the trial. Conclusion: RQ may not be a reliable method to measure AT in HP; there were significant differences with healthy population that can be explained by their physical performance or another reason as the CO<sub>2</sub> product.

## HIGH PREVALENCE OF SEROLOGICAL EVIDENCE FOR HEPATITIS B INFECTION IN CHILEAN PRE DIALYSIS END STAGE RENAL DISEASE PATIENTS.

Alvo M, Bolstansky A, Elgueta L, Tambo S.

Introduction: hepatitis B (HB) infection continues to represent a major hazard in hemodialysis patients. Hepatitis B surface antigen (HBsAg) is the usual test utilized to identify infecting patients. Complementary serology and viral DNA allows detecting past infections and hidden forms of hepatitis B. The purpose of the present study was to determine the prevalence of hepatitis B virus (HBV) infection in end stage renal disease (ESRD) patients before initiation of dialysis. Methods: Every patient with ESRD before initiating dialysis that attended to the Clinical Hospital of University of Chile during 2002 to 2004 was included. At admission to study all patients answered a survey to detect risk factors for HB infection. Blood samples were drawn for HBsAg and total anti core antibodies (HBcAb). If both were negative no more studies were done. If only HBcAb was positive, IgM HBcAb, anti HBsAg and HBV DNA were determined. If HBsAg and HBcAb were positive, IgM HBcAb and HBV DNA were determined. If only HBsAg was positive, HBV DNA was determined. Results: One hundred and eight patients were admitted to the study. Only one patient was positive for HBsAg, in this patient HBcAb and HBV DNA were negative. In 14 patients HBcAb was positive, from these 9 tested positive for anti HBsAg and negative for HBV DNA. The other five were negative for anti HBsAg and HBV DNA. No differences in risk factors for HB infection were found between patients which positive and negative serology. Conclusion: In our cohort of pre dialysis patients we found that the only one patient with an HBsAg positive was a false positive. In 8.3% of the patients evidence of past and solved infection with HBV was found. A group of 4.6% of the patients had a serology of difficult interpretation (HBcAb positive with HBsAg and HBV DNA negative) this may represent a false positive for HBcAb, an infection with low or intermittent viremia, or a infection with a mutant virus. In this cohort of Chilean pre dialysis ESRD patients serological evidence of past contact with HBV and difficult interpretation serology are frequent.

## ABSTRACTS

### NEONATOLOGÍA

#### **3<sup>rd</sup> INTERNATIONAL CONFERENCE ON BIRTH DISEASES AND DISABILITIES - RÍO DE JANEIRO, BRASIL.**

##### **POLYDACTYLY: THE CLINICAL CHARACTERISTICS, GENE PENETRANCE AND PREVALENCE IN A CHILEAN SAMPLE.**

Lucía C, Julio N.

Polydactyly has been recognized as an autosomal dominant illness since centuries, nevertheless, its inheritance remain unclear until our days. It has different clinical characteristics in different populations according their ethnic origin, even more, the gene responsible has been mapped to different chromosomal regions in different families. The purpose of this study is to determine the clinical characteristics, prevalence and gene penetrance of polydactyly in a Chilean Hospital, member of the ECLAMC (HCUCH, Hospital Clínico of University of Chile). We studied every polydactyly case detected in the 37.008 newborns occurred from 1991 until 2006 at the HCUCH. We found 92 cases of polydactyly, 28 of them were associated to others congenital malformations. The prevalence at birth of the isolated polydactyly was 1.7 x 1.000 births (13 preaxials and 46 postaxials cases), similar to the frequency describe in Mexico and lower than the figures described for Black populations. The polydactyly frequency was higher in male than females, even this predominance didn't reach the statistical significance. The prevalence was similar in hands and foot. The 30.4 % of cases related a relative with polydactyly. Based on these familial cases we estimated a gene penetrance of 62.6 % for postaxial polydactyly; this high family recurrence agree with a dominant inheritance, with modifier genes, but not with a multifactorial model, like some authors stated.

### NEUROLOGÍA Y NEUROCIRUGÍA

#### **11<sup>th</sup> INTERNATIONAL CONGRESS OF PARKINSON'S DISEASE AND MOVEMENT DISORDERS - ESTAMBUL, TURQUÍA.**

##### **CLINICAL CHARACTERIZATION OF A CHILEAN FAMILY WITH KUFOR RAKEB DISEASE AND MUTATIONS IN ATP13A2, A LISOSOMAL ATPASE.**

Behrens MI, Chana P, Parrao T, Venegas P, Miranda M, Rojas CV, Ramírez A.

Objective: To present the clinical description of 5 members of a Chilean family with Kufor Rakeb disease and mutations in ATP13A2. Background: Kufor Rakeb is a rare autosomal recessive disease of juvenile onset characterized by slowly progressive parkinsonism, pyramidalism, and cognitive deterioration. Herein, we describe a family affected with this disorder in which we have found two loss-of-function mutations in ATP13A2, a neuronal P-type ATPase gene (Ramirez et al, Nat Genet 2006;38:1184). Methods: Data correspond to a prospective clinical examination of affected and non affected members of a Chilean family over a period of 10 years. Medical records, neuropsychological testing and CT scans were performed with previous informed consent and approval of the ethics committee of the Hospital Sotero del Rio. Results: The family is composed of non consanguineous parents and 17 children from an area near Santiago, Chile. The last five siblings had normal development until ages around 12-18, when insidiously started to show slowness of movements and decay in school performance, with rigidity and difficulty walking. When first evaluated, 2-5 years after disease onset, there was bradykinesia, subtle tremor, cogwheel rigidity, stooped gait and spasticity. Some showed visual hallucinations and upward gaze palsy. There was little response to levodopa, although it was tried late in the disease course. Disease evolution was slow, with progressive deterioration of motor and mental activity until wheelchair bound and anarthric 15-20 years after. In advanced stages they showed facial-facial-finger mini myoclonus, insomnia and epilepsy. Three have died from pneumonia after 20-27 years of disease. Some of the unaffected members show subtle ocular dysmetria and fine tremor. Conclusions: The affected family members showed a homogeneous pattern of juvenile onset parkinsonism, pyramidal signs, dementia, supranuclear vertical gaze palsy, insomnia and facial-facial-finger myoclonus, very similar to the description of the Jordanian family recently reported (Williams et al, Mov Disord 2005;20:1264). Interestingly, the presence of subtle ocular dysmetria and fine action tremor in some unaffected carriers suggests that haploinsufficiency may cause neurological symptoms.

## 5<sup>th</sup> WORLD CONGRESS OF FEDERATION OF SLEEP RESEARCH AND SLEEP MEDICINE SOCIETIES - CAIRNS AUSTRALIA.

NOCTURNAL LOW OXIGEN SATURATION AS A MAIN FACTOR OF EXCESSIVE DAYTIME SOMNOLENCE.

Herrera A, Aguilera L, Ocampo A, Díaz M.

The main objective is to evaluate Apnoea-hypopnea index (AHI) and sleep time in oxygen saturation below 90% (Sat90) as predictors of subjective excessive daytime somnolence (EDS). Polysomnographic recordings and Epworth Scale test were performed in 105 patients with suspected OSAS. Four groups were established according to AHI values: No OSAS (n = 19), mild OSAS (n = 34), moderate OSAS (n = 24) and severe OSAS (n = 28) when AHI was <5, 5–19, 20–49 or >50 respectively. In our sample, Epworth Scale Score (ESS) does not interact with age, total sleep time, sleep latency, REM sleep latency, sleep efficiency, REM sleep or snoring index. Averages scores of Epworth scale by groups were 11.8, 12.3, 13.9 and 16.5 for no OSAS, mild OSAS, moderate OSAS and severe OSAS respectively (Kruskal–Wallis test,  $p < 0.01$ ). ESS is inversely related to Slow Wave Sleep amount (Spearman test,  $\rho = -0.21$ ,  $p < 0.05$ ), and directly with the AHI and Sat90 (Spearman test,  $\rho = 0.29$  and  $0.28$  respectively,  $p < 0.01$ ). In the No OSAS group the ESS is associated to Sat90 (Spearman test,  $\rho = 0.58$ ,  $p < 0.01$ ), but it is not with AHI. Nocturnal oxygen desaturation is a mayor determinant of EDS among OSAS and non-OSAS patients.

## PEDIATRÍA

### 45<sup>th</sup> ANNUAL MEETING OF IDSA (INFECTIOUS DISEASES SOCIETY OF AMERICA) – SAN DIEGO, USA.

SAFETY, TOLERABILITY, AND IMMUNOGENICITY OF MOTAVIZUMAB IN YOUNG CHILDREN AFTER A SECOND SEASON OF RSV PROPHYLAXIS.

Abarca KG, Fernández P, Harris B, Losonsky G, Connor EM.

Background: Palivizumab, an RSV specific monoclonal antibody (MAb) is recommended for various children at high risk for serious RSV disease for 1 to 2 seasons. Motavizumab, an investigational enhanced potency anti-RSV MAb, has been studied during a first season of dosing. Methods: In this phase I/II, randomized, double-blind study, high risk preterm infants aged 24 months who received /3 motavizumab doses in their first RSV season received monthly motavizumab (n = 66) or palivizumab (n = 70) 15 mg/kg IM in their (150 days); serum Mab concentrations and antiMAB antibody were evaluated before the first, second, and fifth dose, and at 30 days and 90 to 120 days post-last dose. Results: The number of children with a reported adverse event (AE) was similar: 84.8% motavizumab and 88.6% palivizumab. Transient mild injection site erythema was the most common drug-related AE in each group (15.2% vs 11.4%). Serious AEs (SAEs) were reported in 5 patients: 4 occurred in the motavizumab group (6.1%) compared with 23 (10.6%) in the first season of motavizumab therapy. One SAE was considered related to study drug, an event of transient acute hypersensitivity post dose 3 in a motavizumab recipient. Mean serum trough motavizumab concentrations increased during dosing (54.6 and 86.2 mg/mL after dose 1 and 4, respectively), similar to that seen in season 1. There were no treatment group-specific antibody responses detected during or after dosing (titer /1:10) to the MABs received during the second season. Conclusions: The safety profile of motavizumab when given for a second consecutive season in high risk children was consistent to that seen during the first season of dosing and comparable to palivizumab.

## RESPIRATORIO

### ANNUAL CONGRESS OF EUROPEAN RESPIRATORY SOCIETY - ESTOCOLMO, SUECIA.

EFFECT OF INSPIRATORY MUSCLE TRAINING ON LUNG FUNCTION, DYSPNEA, EXERCISE TOLERANCE AND QUALITY OF LIFE IN COPD PATIENTS.

Mendoza L, Leiva A, Jover E, Cavada G, Lisboa C.

Introduction: the benefic to finspiratory muscle training (IMT) on the Chronic Obstructive Pulmonary Disease (COPD) is still controversial. Objectives: To Determine the effect sof IMT on muscle function, dyspnea, exercise tolerance and quality of life in COPD. Methods: A randomize double-blind placebo controlled essay was undertook in stable COPD patients that were trained with pressure thresholdf or 10 weeks, randomly assigned to IMT with minimal load: 7 cmH2O (low load group, LLG) or

## ABSTRACTS

with 35% of sustained inspiratory maximal pressure in one second (SIMP) (high load group, HLG). We studied 25 patients, age  $70 \pm 7$  y, FEV1  $142.5 \pm 14\%$  predicted, SMIP  $49 \pm 10$  cmH<sub>2</sub>O. Prior to and at the end of the study, we measured maximal inspiratory pressures, incremental threshold loading, inspiratory capacity (IC), dyspnea with the Medical Research Council (MRC) and Mahler's scores, six-minute walking test (6MWT) and the quality of life with Saint George Respiratory Questionnaire (SGRQ). Results: In the total group, there was an increase of peak maximal inspiratory pressure (PI<sub>max</sub>), SMIP, incremental threshold loading, with a decrease of dyspnea's scores. In contrast, no differences were found between LLG and HLG. This could be explained because the low load group was trained at 16% of SIMP and this value can induce respiratory training. There were no significant differences on SGRQ, IC and 6MWT. Conclusions: IMT produced significant changes on inspiratory muscle function and dyspnea in the total group of COPD patients despite the load employed.

## REUMATOLOGÍA

### CONGRESS EULAR (EUROPEAN LIEGE AGAINST RHEUMATISM) - BARCELONA, ESPAÑA.

BASAL ANTI-CYCLIC CITRULLINATED PEPTIDE (ANTI-CCP) ANTIBODIES LEVELS AND A DECREASE IN ANTI-CCP TITRES ARE ASSOCIATED WITH CLINICAL RESPONSE TO ADALIMUMAB THERAPY IN RHEUMATOID ARTHRITIS PATIENTS.

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Background: Recent reports suggest that a decrease in anti-cyclic citrullinated peptide (anti-CCP) antibodies titres might be a useful adjunct in assessing the efficacy of anti-TNF-alpha treatment in rheumatoid arthritis (RA), although this finding was not confirmed by other groups. The present study was designed to investigate the effect of adalimumab treatment on anti-CCP antibodies in patients with RA. Objectives: To evaluate in a prospective manner the effect of adalimumab treatment in serum anti-CCP antibody levels in a group of 70 RA patients who had an inadequate response to methotrexate or to other disease modifying antirheumatic drugs (DMARDs). Methods: 70 RA patients who failed treatment with DMARDs received 40 mg adalimumab subcutaneously every other week during 24 weeks. Serum samples were collected at baseline and at weeks 8, 16 and 24 before the corresponding adalimumab dose. The serum anti-CCP levels were tested by enzyme linked immunosorbent assay. Results: At baseline, 52 of the 70 patients (74.3%) were positive for anti-CCP antibodies. 60% of the anti-CCP positive patients and 44.4% of the anti-CCP negative patients were ACR 20 responders at week 24 ( $p < 0.049$ ). The serum levels of anti-CCP antibodies decreased significantly after 24 weeks of adalimumab treatment for the whole group of patients ( $p < 0.00069$ ). The serum level of anti-CCP antibodies decreased significantly only in those patients who met ACR 20 response criteria at week 24 ( $p < 0.00044$ ). Differences between baseline anti-CCP titers and those at 8, 16 and 24 weeks were all statistically significant ( $p < 0.014$ ,  $0.003$  and  $0.019$  respectively). No statistically significant changes in the anti-CCP levels were observed in patients who did not meet the ACR 20 response criteria. Conclusion: Basal anti-CCP antibodies levels correlate with clinical response to adalimumab. A decrease in anti-CCP levels on time was observed in patients showing also clinical improvement, suggesting that serum anti-CCP antibodies determination may be useful in assessing treatment efficacy in RA patients.

## UPC

### 36<sup>th</sup> CRITICAL CARE CONGRESS - ORLANDO, USA.

ANALGESIA AND SEDATION IN CRITICALLY ILL PATIENTS ON MECHANICAL VENTILATION IN CHILE.

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Introduction: despite evidence-based guidelines, wide variation in sedation schemes and practice are reported in ICUs. Hypothesis: our aim was to evaluate the use of sedative drugs, quality of sedation and outcome in patients undergoing mechanical ventilation (MV) in Chile. Methods: prospective observational study during 10 weeks in 13 ICU, involving all adult patients requiring ventilatory support. Nervous system diseases, previous liver or renal failure, second episode of MV, and expected MV shorter than 48 hours were main exclusion criteria. Nurse training was implemented for using a sedation/agitation scale (SAS, Crit Care Med 1999;27:1325) at 12 h interval, and sedative, analgesic and muscle relaxants

were registered on a daily basis for the first 7 days, and weekly thereafter. Stepwise logistic regression was used to identify variables independently associated with mortality at 28 days. Results: we evaluated 635 patients, which 155 (24%) fulfilled inclusion criteria: 57% male,  $60 \pm 18$  yo, APACHE II  $19 \pm 6$ , and SOFA  $7.8 \pm 3.1$ . Main diagnosis were sepsis 63%, ALI/ARDS 47%, COPD 19%, congestive heart failure 17%, and trauma 9%. Ventilatory parameters at 24 hours were Pa/FiO<sub>2</sub>  $225 \pm 97$ , PEEP  $7.7 \pm 3.1$ , and Vt  $7.5 \pm 1.7$  ml/kg. Of 1907 SAS measurements 55.4% were 1-2 (deep sopor/coma), 37.1% 3-4 (mild sopor/awake) and 7.4% 5-7 (agitation). Midazolam (86%) and fentanyl (81%) were the most frequently used drugs. Muscle relaxants after intubation were used in 30% by bolus or continuous infusion. Mortality (35.5) was independently associated to SOFA scores ( $p=0.016$ ), medical condition ( $p=0.004$ ) and use of muscle relaxants ( $p=0.012$ ). Conclusions: in Chile, patients on MV are frequently deeply sedated. Midazolam and fentanyl are the preferred drugs, but still 30% of patients use muscle relaxants. Our findings support protocols to improve quality of sedation and outcome of patients on MV.