



Profolio of Researchers and Research Teams

THYROID AND METABOLIC DISEASES UNIT

(Last update 03/04/2013)

Code: GI-1698

Department: Medicina

Web: www.usc.es/medicina/UETeM.html

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Research field

- Molecular basis and clinical characterization of familial lipodystrophies
- Etiopathogenic bases, physiopathological mechanisms and therapeutical principles of the metabolic syndrome
- Biochemical analysis and characterization of post-translational modifications in proteins involved in physiopathology
- Neurodegenerative process associated to seipin mutations
- Molecular-genetic mechanisms involved in thyroid pathology.

Technology services

- Level-2 biosafety laboratory adequate for work with type "scrapie" prions.
- Genotyping, proteomic analysis and crystallography in thyroid pathology, including cell culture and real time PCR

- Genotyping and proteomic analysis in familial lipodystrophies, including cell culture and real time PCR
- Clinical evaluation of rare lipodystrophic syndromes.
- Treatment with human recombinant leptin of rare lipodystrophic syndromes.

Key words

Protein post-translational modification; thyroid molecular pathology; metabolic syndrome; lipodystrophies; adipogenesis; neurodegeneration; diabetic neuropathy

Researchers

Name	Position
Araújo Vilar,D.	Coordinator
Lado Abeal,J.J.	Member
Rodríguez Requena,J.	Member

External collaborators

Alfonso Rodríguez Pérez
Sofía Sánchez Iglesias

SERGAS. CHUS
Sociedad Española de Lipodistrofias

RTD PROJECTS (period: 2009 - 2013)

Title:

Un nuevo síndrome neurodegenerativo asociado a la mutación R265X en el gen BSCL2: caracterización clínica y molecular.

Tipology: Proyectos Xunta

Duration: 10/08/2010 - 30/09/2013

Main investigator: Araújo Vilar, David

Title:

Caracterización clínica y mecanismos moleculares de un nuevo síndrome neurodegenerativo asociado a la mutación R265X en el gen de la seipina

Tipology: Proyectos FISs

Duration: 24/05/2010 - 23/06/2010

Main investigator: Rodríguez Requena, Jesús

Title:

Discover-multidisciplinary project investigating the neurobiology of chronic

Tipology: Proyectos FISs

Duration: 01/01/2010 - 31/12/2013

Main investigator: Rodríguez Requena, Jesús

Title:

Protecting animal and human health from prions in food, feed and the environment (PRIORITY)

Tipology: Contratos coa UE

Duration: 01/10/2009 - 30/09/2014

Main investigator: Rodríguez Requena, Jesús

Title:

International meeting on familial lipodystrophies

Tipology: Accións complementarias

Duration: 03/04/2009 - 02/04/2010
Main investigator: Araújo Vilar, David

SCIENTIFIC PRODUCTION (period: 2007 - 2011)

Articles in scientific journals

Article:

Hyperglycaemic hyperosmolar non-ketotic state as a cause of low gonadotropin levels in postmenopausal diabetic women: a role for severe hypernatraemia

Journal: JOURNAL OF NEUROENDOCRINOLOGY, ISSN: 0953-8194
2007

Article:

Stress-induced alterations in the programmed natural cycles of postnatal lymphoid organ development in C57BL/6 mice: evidence for a regulatory feedback relationship between bone marrow and thymus

Journal: IMMUNOBIOLOGY, ISSN: 0171-2985
2007

Article:

Sensitive, preclinical detection of prions in brain by nanospray liquid chromatography/tandem mass spectrometry

Journal: RAPID COMMUNICATIONS IN MASS SPECTROMETRY, ISSN: 0951-4198
2007

Article:

Mass spectrometric detection of attomole amounts of the prion protein by nanoLC/MS/MS.

Journal: JOURNAL OF THE AMERICAN SOCIETY FOR MASS SPECTROMETRY, ISSN: 1044-0305
2007

Article:

Prevalence of mutations in TSHR, GNAS, PRKAR1A and RAS genes in a large series of toxic thyroid adenomas from Galicia, an iodine-deficient area in NW Spain.

Journal: EUROPEAN JOURNAL OF ENDOCRINOLOGY, ISSN: 0804-4643
2008

Article:

Hypothyroidism is associated with increased myostatin expression in rats.

Journal: JOURNAL OF ENDOCRINOLOGICAL INVESTIGATION, ISSN: 0391-4097
2008

Article:

Identification of molecular mechanisms related to nonthyroidal illness syndrome in skeletal muscle and adipose tissue from patients with septic shock.

Journal: CLINICAL ENDOCRINOLOGY, ISSN: 0300-0664
2008

Article:

Pendred syndrome in two Galician families: insights into clinical phenotypes through cellular, genetic, and molecular studies

Journal: JOURNAL OF CLINICAL ENDOCRINOLOGY & METABOLISM, ISSN: 0021-972X
2008

Article:

Prevalence of mutations in TSHR, GNAS, PRKAR1A and RAS genes in a large series of toxic thyroid adenomas from Galicia, an iodine-deficient area in NW Spain

Journal: EUROPEAN JOURNAL OF ENDOCRINOLOGY, ISSN: 0804-4643
2008

Article:

Identification of molecular mechanisms related to nonthyroidal illness syndrome in skeletal muscle and adipose tissue from patients with septic shock

Journal: CLINICAL ENDOCRINOLOGY, ISSN: 0300-0664
2008

Article:

A novel phenotypic expression associated with a new mutation in LMN gene, characterized by partial lipodystrophy, insulin resistance, aortic stenosis and hypertrophic cardiomyopathy

Journal: CLINICAL ENDOCRINOLOGY, ISSN: 0300-0664
2008

Article:

Combination of cardiorespiratory reflex parameters and heart rate variability power spectrum analysis for early diagnosis of diabetic cardiac autonomic neuropathy.

Journal: DIABETES & METABOLISM, ISSN: 1262-3636
2009

Article:

Oligomer assembly of the C-terminal DISC1 domain (640-854) is controlled by self-association motifs and disease-associated polymorphism S704C.

Journal: BIOCHEMISTRY, ISSN: 0006-2960
2009

Article:

Cardiac autonomic neuropathy, estimated cardiovascular risk, and circadian blood pressure pattern in diabetes mellitus.

Journal: CHRONOBIOLOGY INTERNATIONAL, ISSN: 0742-0528
2009

Article:

Reduced adipogenic gene expression in fibroblasts from a patient with type 2 congenital generalized lipodystrophy.

Journal: DIABETIC MEDICINE, ISSN: 0742-3071
2010

Article:

Thyroid hormone receptors are down-1 regulated in skeletal muscle of patients with Non-thyroidal illness syndrome secondary to non-septic shock.

Journal: EUROPEAN JOURNAL OF ENDOCRINOLOGY, ISSN: 0804-4643
2010

Article:

Functional consequences of seven novel mutations in the CYP11B1 gene: four mutations associated with nonclassic and three mutations causing classic 11B-hydroxylase deficiency

Journal: JOURNAL OF CLINICAL ENDOCRINOLOGY & METABOLISM, ISSN: 0021-972X

2010

Article:

New insights into thyroglobulin pathophysiology revealed by the study of a family with congenital goiter

Journal: JOURNAL OF CLINICAL ENDOCRINOLOGY & METABOLISM, ISSN: 0021-972X
2010

Article:

Histological and molecular features of lipomatous and non-lipomatous adipose tissue in familial portland.

Journal: CLINICAL ENDOCRINOLOGY, ISSN: 0300-0664
2011

Article:

Probing structural differences between PrP(C) and PrP(SC) by surface nitration and acetylation: evidence of conformational change in the C-terminus

Journal: BIOCHEMISTRY, ISSN: 0006-2960
2011

Article:

Utility of Mass Spectrometry in the Diagnosis of Prion Diseases

Journal: ANALYTICAL CHEMISTRY, ISSN: 0003-2700
2011

Article:

Clinical and molecular study of five families with resistance to thyroid hormones

Journal: MEDICINA CLINICA, ISSN: 0025-7753
2011

Article:

Lipodistrofia Congénita Generalizada versus Síndrome de Berardinelli-Seip

Journal: ANALES DE PEDIATRIA, ISSN: 1695-4033
2011

Chapter:

Title:

Fundamentos de la nutrición enteral y su aplicación en el paciente diabético

Book:

Nutrición enteral y diabetes

(978-84-7429-332-6) Publicación: 2007

Editorial: GLOSA

Title:

Neuropatía Diabética

Book:

Tratado SED de Diabetes Mellitus

(978-84-7903-613-3) Publicación: 2007

Editorial: EDITORIAL MEDIC A PANAMERICANA

Title:

Etiopatogenia de la diabetes tipo 2

Book:

Tratado SED de Diabetes Mellitus

(978-84-7903-613-3) Publicación: 2007

Editorial: EDITORIAL MEDIC A PANAMERICANA

Title:

Glicación de la macromoléculas

Book:

Tratado SED de Diabetes Mellitus

(978-84-7903-613-3) Publicación: 2007

Editorial: EDITORIAL MEDIC A PANAMERICANA

Title:

Neuropatía diabética. Clasificación y etipatogenia.

Book:

La diabetes mellitus en la práctica clínica.

(978-84-7903-450-4) Publicación: 2009

Editorial: Ed. Médica Panamericana

Title:

Neuropatía diabética. Clínica y tratamiento.

Book:

La diabetes mellitus en la práctica clínica.

(978-84-7903-450-4) Publicación: 2009

Editorial: Ed. Médica Panamericana

Doctoral theses:

Title:

Aislamiento y caracterización de una fracción de PrPSc sensible a proteinasa K (sPrPSc): estudios estructurales sobre la PrPSc

Date of dissertation: 21/11/2008

Director: Jesús Rodríguez Requena

Author: Miguel Ángel Pastrana González

Title:

Identificación de mecanismos moleculares relacionados con el síndrome del enfermo eutiroideo en el músculo esquelético y el tejido adiposo de pacientes en shock séptico

Date of dissertation: 24/04/2008

Director: Julián Álvarez Escudero, José Joaquín Lado Abeal

Author: Alfonso Rodríguez Pérez

Title:

Identificación de mecanismos moleculares relacionados con el síndrome del enfermo eutiroideo en el músculo esquelético y el tejido adiposo de pacientes en shock séptico

Date of dissertation: 24/04/2008

Director: José Joaquín Lado Abeal, Julián Álvarez Escudero
Author: alfonso rodríguez pérez

Title:

Estudio de la prevalencia y mecanismos de acción de mutaciones en el receptor de TSH y subunidad alfa de la proteína Gs en bocios multinodulares tóxicos y adenomas tóxicos en Galicia

Date of dissertation: 19/01/2009

Director: José Joaquín Lado Abeal
Author: Fernando Alberto Palos Paz
