Profolio of Researchers and Research Teams

THYROID AND METABOLIC DISEASES UNIT

(Last update 03/04/2013)

Code: GI-1698
Department: Medicina
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Center of Innovation and Technology Transfer
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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**

<table>
<thead>
<tr>
<th>Name</th>
<th>Position</th>
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<tbody>
<tr>
<td>Araújo Vilar, D.</td>
<td>Coordinator</td>
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<tr>
<td>Lado Abeal, J.J.</td>
<td>Member</td>
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<td>Rodríguez Requena, J.</td>
<td>Member</td>
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**External collaborators**

<table>
<thead>
<tr>
<th>Name</th>
<th>Institution</th>
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<tr>
<td>Alfonso Rodríguez Pérez</td>
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<td>Sofía Sánchez Iglesias</td>
<td>Sociedad Española de Lipodistrofías</td>
</tr>
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**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 FISss

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

**Main investigator:** Rodríguez Requena, Jesús

**Title:**
Discover-multidisciplinary project investigating the neurobiology of chronic

**Tipology:** Proyectos FISss

**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:** Acciôns 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 natrual cycles of postnatal lymphoid organ development in C%BL/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-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 fectures of lipomatous and non-lipomatous adipose tissue in familial portand.
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 etiopatogenia.

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 bocós multinodulares tóxicos y adenomas tóxicos en Galicia

Date of dissertation: 19/01/2009
Director: José Joaquín Lado Abeal
Author: Fernando Alberto Palos Paz
Treatment for thyroid disease or disorders, depend on the type that caused it. Some thyroid problems will resolve with treatment; however, many cannot be cured and will require life-long treatment. The thyroid has important roles to regulate numerous metabolic processes throughout the body. Different types of thyroid disorders affect either its structure or function. The thyroid gland is located below the Adam's apple wrapped around the trachea (windpipe). Your thyroid gland may be affected by a number of diseases, upsetting your hormone balance. Reviewed by a board-certified internal medicine physician. Thyroid-attacking antibodies that your body produces, including thyroid peroxidase antibodies (TPO) and thyroglobulin antibodies (TgAb). Hashimoto's disease typically causes inflammation and the gradual destruction of your thyroid gland over time. Most people with Hashimoto's end up with hypothyroidism, a condition of insufficient thyroid hormone. Hashimoto's can, however, cause periods or phases of overactivity, known as hyperthyroidism. Thyroid disease is a general term for conditions that prevent your thyroid from making the right amount of hormones. Symptoms can include changes in your heart rate and body weight. There are several different types of thyroid disease, including hyperthyroidism, hypothyroidism, thyroiditis and Hashimoto's thyroiditis. Appointments & Access. Contact Us. Thyroid Disease Menu. Overview Diagnosis and Tests Management and Treatment Outlook / Prognosis Living With. What is the thyroid?